To Mary Michelle, to Frederick, Kimberly, Robert, Charles, Nichols, and Helena, and to Dr. Bruce Herndon, who daily, throughout our joint eye residencies, asked me about another "weird" syndrome
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Preface

This book has three main purposes: (1) to aid the busy ophthalmologist in identifying all ocular manifestations of a given syndrome, disease, or inherited disorder; (2) to provide references so that the ophthalmologist will know where to look for additional information; and (3) to promote greater observation of the characteristics of ocular syndromes, systemic diseases, and inherited disorders. This book is designed for quick reference while the patient’s eyes are being dilated or immediately following an examination. Practicality is the objective; conciseness, logical arrangement, and authenticity are the main features.

I have endeavored to include all systemic syndromes, diseases, and inherited disorders that have ocular manifestations. The titles are arranged alphabetically and are numbered. Other names of the same disorder are also given. A general definition, clinical ocular findings, clinical systemic findings, and several references are included for each entry. Each entry is dealt with as completely as space and practicality allow. Laboratory findings have not been considered; the book is designed to serve as the ophthalmologist’s primary source of information. If any syndromes, systemic diseases, or inherited disorders with ocular manifestations have been omitted, I urge you to inform me of this so that any future editions of the book may include them.

The Ocular Differential Diagnosis book provides comprehensive lists of causes for symptoms or findings. Frequently more information is needed, and it is hoped that, Ocular Syndromes and Systemic Diseases will furnish the physician with additional information to make a better diagnosis.

I am indebted to many individuals who assisted with the preparation of this manuscript, including: Renee Tindall, Dr. Fernando Murillo, Dr. Scott Lowery, Dr. Kae Chatman, Angie Brown, and Louise Geer.
A Esotropia Syndrome

Esotropia greater looking up by 15 prism diopters than looking down; an overaction of superior oblique muscles or underaction of inferior rectus muscles; fusion may be obtained by chin elevation; mongoloid (upward) slant of lid fissures; may be accommodative, nonaccommodative, or paralytic esotropia components.


A Exotropia Syndrome

Exotropia greater looking down by 15 prism diopters than looking up; mongoloid (upward) slant of lid fissures; alternating sursumduction and associated vertical divergence; overaction of superior oblique muscles or underaction of inferior oblique or inferior rectus muscles; fusion obtained by chin depression.


Aarskog Syndrome (Facial-Digital-Genital Syndrome)

General: X-linked recessive; males fully affected; females exhibit partial features; normal birth weight and length.

Ocular: Telecanthus; hypertelorism; unilateral or bilateral blepharoptosis; strabismus; hyperopic astigmatism; large cornea.

Clinical: Short stature; triangular facies; deformity of hands and feet; anomalies of external genitalia; inguinal hernia; protruding umbilicus; abnormal cervical vertebrae; cryptorchidism.


Aarskog-Scott Syndrome (Faciogenital Dysplasia)

General: Sex-linked; characterized by ocular hypertelorism, anteverted nostrils, broad upper lip, and saddlebag scrotum.

Ocular: Ptosis; hypertelorism.

Clinical: Hyperextensibility of fingers; genu recurvatum; flat feet; hypermobility in cervical spine with neurologic deficit; cleft lip and palate; anteverted nostrils; broad upper lip; abnormal penoscrotal relations; "saddlebag scrotum."


Abdominal Typhus (Enteric Fever; Typhoid Fever)

General: Causative agent, Salmonella typhi.
Ocular: Conjunctivitis; chemosis; corneal ulcer; tenonitis; paralysis of extraocular muscles; endophthalmitis; panophthalmitis; optic neuritis; retinal detachment; central scotoma; central retinal artery emboli; iritis with or without hypopyon; choroiditis; retinal hemorrhages; bilateral optic neuritis; abnormal ocular motility (likely secondary to thrombotic infarcts affecting the ocular motor nerve nuclei, fascicles, brainstem, or cerebral hemispheres).

Clinical: Fever; headache; bradycardia; splenomegaly; maculopapular rash; leukopenia; encephalitis. Salmonella may produce an illness characterized by fever and bacteremia without any other manifestations of enterocolitis or enteric fever, which is particularly common in patients with acquired immunodeficiency syndrome (AIDS).


Aberfeld Syndrome (Schwartz-Jampel Syndrome; Congenital Blepharophimosis Associated with Generalized Myopathy Syndrome; Ocular and Facial Abnormalities Syndrome)

General: Etiology not known; autosomal recessive inheritance, although there are reports of dominant inheritance; progressive disorder.

Ocular: Blepharophimosis; exotropia; myopia; congenital cataracts; microcornea.

Clinical: Myopathy; bone deformities; arachnodactyly; dwarfism; hypoplastic facial bones; hypertrichosis; kyphoscoliosis.


Acanthamoeba

General: Caused by *Acanthamoeba polyphaga* and *Acanthamoeba cartel* (see Herpes Simplex Masquerade Syndrome); all types of contact lenses have been associated with acanthamoeba keratitis, particularly daily-wear soft contact lenses.

Ocular: Hypopyon; uveitis; conjunctivitis and chemosis; keratitis; pannus; corneal ring abscess; papillitis; vitreitis; retinal perivasculitis; secondary glaucoma; postkeratoplasty acanthamoeba keratitis may present as an infectious crystalline keratopathy in the periphery of the graft.

Clinical: Meningoencephalitis; meningitis; hemorrhagic encephalitis.


Acanthosis Nigricans

General: Rare skin disease of unknown etiology; occurs at any age; equal frequency in males and females; thickening and hyperpigmentation of the skin of the entire body, especially in flexural areas; autosomal dominant inheritance.

Ocular: Conjunctivitis; pigmentation of palpebral conjunctiva; tumors on lids and lid margins; madarosis; trichiasis.

Clinical: Hyperkeratotic lesions on face, neck, oral mucosa, axillae, groin, antecubital fossae, and umbilicus.


**Accommodative Insufficiency (Accommodative Effort Syndrome)**

**General:** Male or female; 10 to 35 years of age.

**Ocular:** Asthenopia with near vision appearing within a few minutes after reading, sewing, or observing a near object; increased amplitude of accommodative adduction; abnormal relaxation of accommodation induced by relative divergence at close distances; latent convergence insufficiency; may result secondary to ciliary dysfunction associated with Adie pupil.

**Clinical:** It may be secondary to a systemic disorder such as Parkinson disease or oral lithium.


**Aceruloplasminemia**

**General:** Autosomal recessive, adult-onset

**Ocular:** Maculopathy which resembles aging macular degeneration

**Clinical:** Associated with increased levels of iron in the retina


**Achard Syndrome**

**General:** All features of Marfan syndrome, with the addition of dysostosis mandibulofacialis; arachnodactyly; receding lower jaw; joint laxity limited to the hands and feet; differs from Marfan syndrome in that the skull is broad and brachycephalic with small mandible.

**Ocular:** Myopia; lens dislocation; spherophakia.

**Clinical:** Mandibulofacial dysostosis; skeletal anomalies; arachnodactyly; high arched palate; heart disease.


**Achondroplasia**

**General:** Dwarfism; etiology unknown; occurs in both sexes; inheritance is autosomal dominant with almost complete penetrance; characterized by rhizomelic dwarfism (reduction most marked in the proximal limbs); mid-face hypoplasia; exaggerated lumbar lordosis; limitation of hip and elbow expansion; location of achondroplastic gene reported to be in the short arm of chromosome 1.

**Ocular:** Strabismus; optic atrophy; hypermetropia.

**Clinical:** Osseous impingement upon cranial nerves; rhizomelic short stature; facial features include frontal bossing, depressed nasal bridge, relative mandibular prognathism; connective tissue dysplasia; hypotonic in infancy; paraplegia may develop in second or third decade.


ACHOO Syndrome (Autosomal Dominant Compelling Helio-Ophthalmic Outburst Syndrome; Photic Sneeze Reflex; Sneezing from Light Exposure; Peroutka Sneeze) 13

General: Autosomal dominant; sneezing in response to bright light, especially sunlight; association between photic sneeze and nephropathic cystinosis has been reported.

Ocular: Photic sneeze reflex by sudden exposure of dark-adapted subject to bright light.

Clinical: Successive sneezing, as many as 43 in a row.


Acinetobacter (Mima Polymorpha; Acinetobacter Iwoffi) 14

General: Gram-negative pleomorphic bacillus Mima; generally occurs in patient with lowered resistance.

Ocular: Conjunctivitis and chemosis; corneal ulcer; blepharitis; iris prolapse; endophthalmitis.

Clinical: Meningitis; pneumonia; endocarditis; urethritis; vaginitis; arthritis; dermatitis; intracranial abscess; subdural empyema.


Ackerman Syndrome 15

General: Autosomal recessive; characterized by pyramidal molar roots.

Ocular: Juvenile glaucoma.

Clinical: Unusual upper lip and dental roots; pyramidal molar roots.


ACL Syndrome (Acromegaloïd, Cutis Verticis Gyrata, Corneal Leukoma Syndrome) 16

General: Autosomal dominant; rare; three features include cutis verticis, associated with acromegaly and corneal leukoma; onset by age 1 year.

Ocular: Bilateral corneal leukoma; keratitis.

Clinical: Unusually tall; large hands, feet, and chin; skin of hands very soft; skin of scalp lies in folds; frontal bosses; ear calcification; pituitary tumors; abnormal dermal ridge patterns; enlargement of supraorbital arch of frontal bone.

Acne Rosacea (Acne Erythematosa; Ocular Rosacea)

**General:** Etiology unknown; usually occurs in women 30 to 50 years of age; pathogenetic mechanism remains unclear.

**Ocular:** Conjunctivitis; corneal neovascularization (wedge-shaped); keratitis; meibomianitis; blepharitis; recurrent chalazion; conjunctival hyperemia; superficial punctate keratopathy; corneal vascularization, thinning, perforation, and scarring; episcleritis; scleritis; iritis; nodular conjunctivitis.

**Clinical:** Symmetrical erythema; papules; pustules; telangiectasia; sebaceous gland hypertrophy of the forehead, malar eminences, and nose.


Acosta Syndrome (Mountain Sickness; Mountain Climber Syndrome; Monge Syndrome; Soroche Syndrome)

**General:** Cause is cerebral hypoxia at high altitudes; Monge syndrome is the chronic form of mountain sickness.

**Ocular:** Acute blurred vision; difficulties in color discrimination; impaired light adaptation; retinal hemorrhage; chronic lid edema; bluish scleral injection; decreased visual acuity.

**Clinical:** Restlessness and irritability; headaches; impaired judgment at approximately 15,000 feet; confusion, cyanosis, muscular incoordination and possible loss of consciousness at approximately 18,000 to 20,000 feet; exertional dyspnea; epistaxis; gum bleeding; hemoptysis; anorexia; nausea; vomiting; tinnitus; cough; loss of libido; paresthesian extremities; coma; clubbing of fingers; hepatosplenomegaly.


Acquired Immunodeficiency Syndrome (AIDS; Acquired Cellular Immunodeficiency; Acquired Immunodeficiency)

**General:** Acquired breakdown of the immune system followed by disease that takes advantage of the body's collapsed defenses; acquired by shared drug needles or sexual intercourse; occurs most frequently in homosexually active men (75%), intravenous drug abusers (13%), and Haitian immigrants (6%).

**Ocular:** Retinal cotton-wool spots; cytomegalovirus retinitis; retinal periphlebitis; conjunctival Kaposi sarcoma; necrotizing retinitis; retinal hemorrhages; conjunctivitis sicca; orbital Burkitt lymphoma; peripheral retinochoroiditis; vitreitis; fungal corneal ulcer; hypopyon; acute glaucoma; third nerve palsy; anterior uveitis; atypical retinitis; orbital pseudotumor; herpes zoster ophthalmicus; herpes simplex keratitis; bacterial keratitis; molluscum contagiosum; cytomegalovirus retinitis; toxoplasmosis retinitis; acute retinal necrosis; human immunodeficiency virus (HIV) retinitis; syphilitic retinitis; *Pneumocystis carinii* choroiditis; fungal and bacterial endophthalmitis; fungal choroiditis; conjunctival microvasculopathy; keratitis sicca; subconjunctival hemorrhage.

**Clinical:** Because of lowered immunity, one third develop Kaposi sarcoma; pneumonia caused by *Pneumocystis carinii*; death.


**Acquired Lues (Syphilis; Acquired Syphilis; Lues Venerea; Malum Venereum)**

**General:** Causative agent, *Treponema pallidum*, usually transmitted sexually.

**Ocular:** Conjunctival chancroid; conjunctivitis; keratitis; blepharitis; ptosis; iritis; atrophy; hippocampus; dacryocystitis; optic nerve atrophy; optic neuritis; periostitis; scleritis; nystagmus; uveitis; vitreous hemorrhages; paralysis of sixth nerve; papilledema; retinal hemorrhages; retinitis proliferans; oculogyric crisis; neuroretinitis; papilledema; optic atrophy; peripheral nerve disease; diffuse or multifocal chorioretinitis; vertical supranuclear gaze palsy; Benedikt syndrome.

**Clinical:** Primary lesion associated with regional lymphadenopathy; secondary bacteremic stage associated with generalized mucocutaneous lesions; tertiary stage characterized by destructive mucocutaneous, musculoskeletal, or parenchymal lesions, aortitis, or central nervous system disease; syphilis and HIV infection often coexist in the same patient who experiences a higher incidence and greater severity of neurologic and ocular manifestations; a significant percentage of patients infected with HIV-1 and T pallidum become seronegative to syphilis testing.


**Acrodermatitis Chronica Atrophicans**

**General:** Rare familial skin disorder; autosomal recessive; both sexes equally affected; occurs in infants; not present at birth but develops during first few weeks; nutritional deficiency; there are reports of improvement following treatment with zinc suggesting an abnormality in the zinc-binding factor.

**Ocular:** Scarring of the conjunctiva; recurrent iridocyclitis; keratomalacia; cataracts; photophobia; blepharitis; punctal stenosis; corneal opacification.

**Clinical:** Vesiculobullous eruption around body orifices, skin of knees, elbows, and paronychial areas; complete alopecia; erythematous psoriasiform plaques.


**Acropachy (Hypertrophic Pulmonary Osteoarthropathy)**

**General:** Three separate components: clubbing of fingers, periosteal proliferation of distal ends of long bones, and arthritis; symptom disappear with control of disease.

**Ocular:** Exophthalmos.

**Clinical:** Finger edema; fibrous overgrowth to fingertips; nail deformity; elevated hormone levels; periosteal bone changes affecting distal radius, ulna, tibia, fibula, metacarpals, and phalanges.


Acrorenoocular Syndrome

General: Autosomal dominant; Duane syndrome with radial defects.

Ocular: Complete coloboma; coloboma of optic nerve; ptosis and Duane anomaly.

Clinical: Renal anomalies; hypoplasia of distal part of thumb with lack of motion at phalangeal joint; renal ectopia without fusion; bladder diverticula; malrotation of both kidneys; absence of kidney; clubhand or absence of thumb.


Actinomycosis

General: Gram-positive *Actinomyces israelii*.

Ocular: Hypopyon; conjunctivitis; keratitis; corneal ulcer; proptosis; uveitis; dacryocystitis; yellow nodules on conjunctiva and eyelids; occlusion of nasolacrimal canaliculi; canaliculitis; orbital abscess; endophthalmitis (rare).

Clinical: Chronic inflammatory induration and sinus formation.


Acute Follicular Conjunctivitis (Adenoviral Conjunctivitis; Pharyngoconjunctival Fever; Syndrome of Beal)

General: Infectious disease produced by adenovirus; serotypes 3, 4, 7, 8, 19, 37, and several others may cause acute conjunctivitis with or without upper respiratory tract involvement; epidemic keratoconjunctivitis has been reported worldwide associated with 11 virus serotypes, with serotypes 8, 11, and 19 being the most commonly responsible.

Ocular: Conjunctivitis; chemosis; keratitis; blepharitis; blepharospasm.

Clinical: Fever; pharyngitis; lymph node enlargement; malaise; myalgia; headache; diarrhea.


Acute Frosted Retinal Periphlebitis

General: Etiology unknown, virus suspected; involvement of veins and arteries; veins more severely affected.

Ocular: Vascular sheathing; retinal hemorrhages; exudative detachment; retinal neovascularization; thick, inflammatory infiltrates around retinal veins; macular detachment; retinal tears associated with posterior vitreous detachment; peripheral uveitis; retinal periphlebitis; associated cytomegalovirus retinitis in HIV-seropositive patients; observed as an idiopathic finding in a child.

Clinical: Associated with tuberculosis; syphilis; sarcoidosis; multiple sclerosis; HIV infection.


Acute Hemorrhagic Conjunctivitis (AHC; Epidemic Hemorrhagic Keratoconjunctivitis)

General: First reported in 1969, first epidemic in United States in 1981; enterovirus; explosive onset; usually bilateral; coxsackievirus A24 and enterovirus 70 have been implicated in the most recent outbreaks.
Ocular: Chemosis; follicular conjunctivitis; petechial bulbar hemorrhages; seromucous discharge; keratitis; lacrimation; lid edema; photophobia; preauricular lymphadenopathy.

Clinical: Systemic symptoms are rare, although several cases of lumbosacral radiculomyelitis have occurred late in the course of the disease; polio-like paralysis (associated with enterovirus 70).


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Acute Retinal Necrosis Syndrome (ARN Syndrome; Bilateral Acute Retinal Necrosis; Barn Syndrome) 28

General: Evidence of association with herpes-type deoxyribonucleic acid (DNA) virus; occurs both unilaterally and bilaterally; includes varicella-zoster virus and herpes simplex virus type 1.

Ocular: Uveitis; vasculitis; vitreitis; retinal detachment; vitreous opacification; retinal periarteritis; exudates of peripheral retina; retinal necrosis; optic nerve enlargement; papillitis; arcuate neuroretinitis; arteritis and phlebitis (affecting the retinal vasculature); necrotizing retinitis; moderate to severe vitreitis; anterior segment inflammation; optic neuritis; late retinal detachment.

Clinical: None.


---

Addison Pernicious Anemia Syndrome (Pernicious Anemia Syndrome; Vitamin B12 Deficiency Anemia; Macrocytic Anemia; Biermer Syndrome) 29

General: Autosomal dominant; female preponderance; onset between ages 30 and 50 years; lack of intrinsic factor normally produced in the fundus of stomach and important for absorption of vitamin B12 in the intestinal tract; infrequent ocular involvement.

Ocular: Central scotoma, centrocecal scotomata, and field contractions in a few cases; retinal hemorrhages (round with white center) at the posterior pole; both retina and disk may have a whitish, hazy appearance; optic neuritis (ischemic); optic atrophy; palsies of extraocular muscles; ocular hypotony; cataract; bilateral, slowly progressive optic neuropathy, unclear etiology.

Clinical: Megaloblastic anemia (chronic and progressive); hypochlorhydria; glossitis; stomatitis; constipation or diarrhea; paresthesias and numbness; incoordination; ataxia; sphincter malfunction.


---

Addison Syndrome (Addison Disease; Idiopathic Hypoparathyroidism; Adrenal Cortical Insufficiency; Moniliasis-Idiopathic Hypoparathyroidism) 30

General: Familial occurrence; association with moniliasis; onset during end of first and beginning of second decade of life; atrophy of adrenal cortex; prognosis for life is poor, with death in adrenal crisis.
Ocular: Ptosis; blepharitis; blepharospasm; keratoconjunctivitis with extreme photophobia; corneal ulcers; episcleritis; keratitic moniliasis; cataracts; papilledema.

Clinical: Moniliasis; tetany; progressive weakness; anorexia; progressive skin pigmentation; dry skin; brittle fingernails and toenails; sparse pubic and axillary hair or total alopecia; impotence.


**Adie Syndrome (Holmes-Adie Syndrome; Markus Syndrome; Saenger Syndrome; Tonic Pupil; Iridoplegia Interna; Myotonic Pupil; Pseudotonic Pupillotonia; Weill-Reys Syndrome; Ross Syndrome)**

General: Cause unknown; more frequent in females; manifested in second and third decades; abnormal sensitivity to 2.5% solution of methacholine; segmental compensatory hyperhidrosis; tonic pupil constricts, whereas normal pupils are unaffected; tonic pupil, hyporeflexia, and segmental hypohidrosis are manifestations of Ross syndrome.

Ocular: Slightly enlarged pupils; delayed or diminished direct and consensual reaction to light; usually unilateral; consensual reflex is abolished on the affected side but normal on the other; amblyopia.

Clinical: Loss of tendon reflexes, particularly ankle and knee jerk (partial or total).


**Adrenoleukodystrophy (Melanodermic Leukodystrophy; Sudanophilic Leukodystrophy)**

General: Degenerative metabolic disease in which cholesterol with long-chain fatty acids accumulates in affected cells; symptoms usually begin between ages 3 and 12 years but may have their onset in adulthood; X-linked recessive with predominantly central nervous system and adrenal dysfunction.

Ocular: Optic atrophy; retinal ganglion cell degeneration; exotropia; esotropia; cataracts; optic pallor; optic nerve hypoplasia; visual field defects; macular pigmentary changes; progressive visual loss.

Clinical: Central nervous system manifestations consisting of behavioral changes, disturbance of gait, dysarthria, and dysphagia; seizures; spastic quadriaparesis; decorticate posturing; one third of patients show adrenal insufficiency.


**African Eye Worm Disease (Loiasis)**

General: Caused by filarial nematode *Loa loa*; transmitted to humans by diurnally biting flies (deerflies) of the *Chrysops* species that live in the rain forests of West and Central Africa.
Ocular: Parasites of anterior chamber, conjunctiva, eyelid, vitreous, and choroid; conjunctivitis; keratitis; optic nerve atrophy; white, cottony mass of vitreous; central retinal artery occlusion; macular hemorrhages; paralysis of extraocular muscles; nystagmus; uveitis.

Clinical: Transient erythematous swelling; pruritus; eosinophilia; fever; urticaria; rarely neurologic involvement.


Agranulocytosis Syndrome (Pernicious Leukopenia Syndrome; Schultz Syndrome; Agranulocytic Angina Syndrome; Malignant Neutropenia Syndrome) 34

General: Caused by hypersensitivity reaction to chemicals, drugs, and ionizing radiation; may be idiopathic; more frequent in adults; female (3:1) preponderance; acute onset.

Ocular: Scleral and conjunctival icterus; conjunctival hemorrhages; retinal hemorrhages.

Clinical: Swollen, painful joints; malaise; sore throat with mucous membrane ulceration; sepsis.


Aicardi Syndrome 35

General: All symptoms present at birth; cause unknown; all findings progress with age; shows X-linked dominant inheritance.

Ocular: Microphthalmia; lid twitching; absent pupillary reflexes; round retinal lacunae up to disk size look like holes with retinal vessels crossing over them; funnel-shaped disk; chorioretinitis.

Clinical: Infantile spasms (tonic seizures in flexion); epileptic seizures; cyanosis; mental anomaly; vertebral anomalies; telangiectasia; hypotonia; head deformities with biparietal bossing, occipital flattening, and plagiocephaly; defects of corpus callosum; cortical heterotopia; characteristic electroencephalogram; dilated intracranial ventricle with leukomalacia.


Alagille Syndrome (AGS; Alagille-Watson Syndrome, AWS; Cholestasis with Peripheral Pulmonary Stenosis; Arteriohepatic Dysplasia, AHD; Hepatic Ductular Hypoplasia, Syndromatoc) 36

General: May be associated with 20p 11.2 deletion and four distinct coding mutations in Jag 1 gene.

Ocular: Posterior embryotoxon and retinal pigmentary changes; anterior chamber anomalies, associated with eccentric or ectopic pupils.

Clinical: Neonatal jaundice; prominent forehead and chin; pulmonic valvular stenosis as well as peripheral arterial stenosis; abnormal vertebrae ("butterfly" vertebrae) and decrease in interpediculate distance in the lumbar spine; absent deep tendon reflexes and poor school performance; in the facies, broad forehead, pointed mandible, and bulbous tip of the nose; and in the fingers, varying degrees of foreshortening.


Albers-Schonberg Disease (Marble Bone Disease; Osteosclerosis Fragilis Generalisata; Osteopetrosis; Osteopoikilosis; Osteosclerosis Congenita Diffusa) 37

General: Simple recessive inheritance, also dominant transmission; benign form is asymptomatic in about 50% of cases and known under the synonym Henck-Assmann syndrome; prognosis is poor for malignant form, with death usually in infancy.

Ocular: Oculomotor paralysis; cranial nerve VII palsy; optic atrophy; ptosis; exophthalmos; papilledema; nystagmus; anisocoria; congenital cataracts; hypertelorism; visual loss in infancy; nasolacrimal duct obstruction; keratoconus.

Clinical: Cartilage and bone thickening; multiple fractures; hyperchromic anemia; osteomyelitis; severe forms: jaundice, hepatosplenomegaly, skeleton sclerosis, lymphadenopathy, and hydrocephalus in infants; mild forms: nerve compression, fractures, and milder form of anemia; pancytopenia from marrow obliteration; low serum calcium; elevated phosphorus.


Albinism (Brown Oculocutaneous Albinism; Nettleship Falls Syndrome) 38

General: Congenital hypopigmentation.

1. Complete.

Ocular: Iris thin, pale blue; prominent choroidal vessels with poorly defined fovea; nystagmus; head nodding; frequently myopic astigmatism and strabismus; marked photophobia; eyelashes and eyebrows are white; optic atrophy; cataract; abnormal decussation of retinogeniculate axons at the chiasm.

Clinical: White hair, eyebrows, and skin; autosomal recessive.

2. Modified complete.

Ocular: Slight pigmentation at pupillary border; may be nystagmus, photophobia, and myopia; choroidal vessels prominent.

Clinical: Negroes; slight pigmentation; golden hair; tendency to hyperkeratoses; freckling in exposed areas of skin; autosomal recessive.

3. Ocular.

Ocular: Marked deficiency of pigment in iris and choroid; nystagmus and myopic astigmatism; iris of female carrier frequently is translucent; macular hypoplasia; photophobia; pigmentation of retinal pigment epithelium.

Clinical: Normal pigmentation elsewhere; autosomal recessive.

4. Amish.

Ocular: At birth, complete albinism with blue translucent irides and albinotic fundal reflex; nystagmus; photophobia; increasing pigmentation with age; abnormal decussation of retinogeniculate axons at the chiasm.

Clinical: White hair and skin at birth; increasing pigmentation with yellow hair and normal skin that tans; autosomal recessive.

Albright Syndrome (Fuller Albright Syndrome; Jaffe-Lichtenstein Syndrome; McCune-Albright Syndrome; Osteitis Fibrosa Disseminata; Osteodystrophia Fibrosa; Polyostotic Fibrous Dysplasia; Fibrous Dysplasia)

**General:** Etiology unknown; disease rare; manifested in children and young adults; found predominantly in females.

**Ocular:** Unilateral proptosis; papilledema; optic atrophy; lacrimal fossa mass; acute or chronic monocular visual loss.

**Clinical:** Medullary structures replaced by fibrous dysplasia; pelvic bones and lower extremities most frequently involved (spontaneous fractures); brown pigmented areas of skin, from small, freckle-like dots to large, flat patches on thighs, sacrum, upper spine, neck, and scalp; endocrine dysfunction (precocious puberty in females) with early menarche, adolescent external genitalia, and breast enlargement; loss of hearing; convulsions; mental retardation.


Alcoholism

**General:** Classified into three groups; symptoms of mental disease, physiologic poison, or result of social drinking; addiction compounds other health disorders.

**Ocular:** Congestion of conjunctiva; amblyopia; diplopia; night blindness; nystagmus; cataracts; paralysis of accommodation; paralysis of extraocular muscles; esophoria for distance fixation; acute visual loss; cotton-wool spots; cherry-red spot (associated with pancreatitis).

**Clinical:** Tremors; seizures; delirium; alcoholic hepatitis; cirrhosis; gastritis; pancreatitis; cancer of mouth and esophagus; peripheral neuropathy; organic brain disease; hypertension; cardiomyopathy; hypoglycemia; anemia; hyperuricemia; susceptibility to infections; skeletal myopathies.


Alexander Disease

**General:** Rare degenerative neurologic disorder characterized by diffuse demyelination in the presence of Rosenthal fibers; cases may resemble multiple sclerosis; neuroradiologic findings include increased cerebellar white matter hyperintensity and diffuse periventricular signal hyperintensities.

**Ocular:** Impaired smooth pursuit; gaze-evoked horizontal nystagmus; slowed saccades; ocular myoclonus.

**Clinical:** In infants, hydrocephalus, spasticity, and seizures; in juveniles and adults, bulbar palsy and hyperreflexia, intermittent neurologic dysfunction.


Alkaptonuria (Ochronosis; Garrod Syndrome)

**General:** Rare autosomal recessive metabolic disease; enzyme homogentisic acid oxidase missing; both sexes affected; onset in first few days of life; manifestations more severe in males.

**Ocular:** Pigmentation of cornea, sclera, and conjunctiva; ochronosis of sclera; oil globulation within Bowman membrane.

**Clinical:** Black-colored urine on standing; osteoarthritis; valvular heart disease; atherosclerosis (homogentisic acid oxidase deficiency); pigmentation of cartilage and other connective tissues.


**Allergic to Everything Syndrome**

**General:** "Environmentally ill" or allergic to unusual or common substances in the environment; females affected most frequently.

**Ocular:** Transient visual loss.


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**Alopecia Areata**

**General:** Unknown etiology; increased incidence of autoimmune disease, Addison disease, diabetes mellitus, and vitiligo; initially occurs episodically; most commonly in patients between the ages of 5 and 40 years; most cases repopulate with normal hair in 6 to 12 months without any specific treatment.

**Ocular:** Loss of eyelashes and eyebrows and cataract.


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**Alport Syndrome (Hereditary Familial Congenital Hemorrhagic Nephritis; Hereditary Nephritis; Familial Nephritis)**

**General:** Autosomal dominant inheritance; early death in males; normal life span in females.

**Ocular:** Anterior lenticonus (bilateral progressive); subcapsular cataracts; thinning of lens capsule; fundus albinopunctatus; retinopathy similar to juvenile macular degeneration; hyaline bodies of optic nerve head; vesicles in Descemet membrane affecting basement membrane collagen; anterior and polar cataracts.

**Clinical:** Hemorrhagic nephritis; progressive nerve deafness; deafness (high tone, sensorineural); most often transmitted as an X-linked dominant trait, although dominant and recessive transmission has been reported.


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**Allgrove Syndrome**

**General:** Autosomal dominant

**Ocular:** Distichiasis, conjunctivitis, keratitis, congenital alacrima

**Clinical:** ACTH insensitivity, achalasia


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**Alstrom Disease (Cataract and Retinitis Pigmentosa)**

**General:** Retinal lesion associated with deafness; severe visual loss in the first decade. **Ocular:** Cataract; retinitis pigmentosa; optic atrophy; salt and pepper pigment epithelial abnormalities. Electroretinogram pathognomonic findings include initially normal rod component, which can become undetectable as early as 5 years of age; undetectable cone activity at 18 months.
**Clinical:** Nerve deafness; diabetes mellitus in childhood; obesity; renal disease; baldness; hyperuricemia; hypogenitalism; acanthosis nigricans; skeletal anomalies; diabetes mellitus; deafness.


**Alzheimer Disease (Dementia)**

**General:** Diffuse brain atrophy coming on well before the senile period of life; progressive; etiology currently unknown but hereditary disorder suspected; terminally, nearly decorticate, with loss of all ability to think, perceive, speak, or move.

**Ocular:** Fixed dilated pupil; optic atrophy; decreased contrast sensitivity, color vision, and stereo vision; abnormalities of the optic nerve head and nerve fiber layer; controversy exists regarding the ability to diagnose patients with Alzheimer disease by their marked hypersensitivity in pupil dilation response to tropicamide.

**Clinical:** Emotional disturbances; depression; anxiety; antisocial behavior; aphasia; apraxic disturbances; abnormalities of space perception; shuffling gait; generalized shuffling gait with short steps; disturbances in thought process.


**Amaurosis Fugax Syndrome**

**General:** Caused by malignant hypertension; often occurs in association with heavy smoking; may indicate vascular insufficiency of the vertebrobasilar arterial system; may precede a cerebrovascular accident and not infrequently seen in vascular insufficiency problems of the carotid arterial system; the cause, if found, is commonly an abnormality in the ipsilateral carotid artery or a cardiac source of embolism.

**Ocular:** Partial blindness in short attacks to permanent complete blindness; scintillating scotoma; teichopsia; retinal arteriolar spasm; signs of arteriolar sclerosis.

**Clinical:** Malignant hypertension; atherosclerosis; expanding lesions of the frontal or temporal lobe; vascular insufficiency.


**Amblyopic Schoolgirl Syndrome**

**General:** Etiology unknown.

**Ocular:** Amblyopia; changes in visual fields; abnormal dark adaptation curve; visual field defects usually are tubular or spiral but also include central, paracentral, and ring scotomas; hemianopsias and superior and inferior field defects reported.

**Clinical:** Psychogenic disorder: affective and hysterical.


**Amebiasis (Amebic Dysentry, Entamoeba Histolytica)**

**General:** Caused by *Entamoeba histolytica*; *E. histolytica* cysts in stools are diagnostic.

**Ocular:** Conjunctivitis; iridocyclitis; hypopyon; central choroiditis; retinal hemorrhages; retinal perivasculitis; macular edema; corneal ulceration; granulomatous and nongranulomatous uveitis; vitreous hemorrhage.

**Clinical:** Chronic dysentery; abscesses of liver and brain; toxic megacolon.

Amendola Syndrome  52

**General:** Observed in Sao Paulo, Brazil; all ethnic groups are affected; endemic form of pemphigus foliaceus; possibly caused by environmental agents; autoimmune disease mediated by autoantibodies of the immunoglobulin G (IgG) class, IgG4 subclass.

**Ocular:** Blisters around eyebrows; entropion; ectropion; trichiasis; iritis.

**Clinical:** Brazilian pemphigus (fogo selvagem, "wild fire"), which resembles, because of its appearance, pemphigus foliaceus; fever; chills.


American Mucocutaneous Leishmaniasis (Cutaneous Leishmaniasis; Oriental Sore)  53

**General:** Causative agent is protozoal parasite *Leishmania brasiliensis*.

**Ocular:** Keratitis; eyelid edema; conjunctival ulcer; vascular sclerosis of choroid; granulomata of eyelid and conjunctiva; blepharoconjunctivitis.

**Clinical:** Ulcerating granulomas of the skin, nasal septum, nasopharynx, lips, soft palate, larynx, and genitals.


Aminopterin-Induced Syndrome  54

**General:** Teratogenic effect of aminopterin and derivatives on fetus; present at birth; usually fetal or postnatal death.

**Ocular:** Hypertelorism.

**Clinical:** Small body; microcephaly; hypoplasia of cranial bones; broad nasal bridge; micrognathia; cleft palate; low-set ears; mesomelic; hypodactyly; talipes equinovarus.


Amniogenic Band Syndrome (Ring Constriction; Streeter Dysplasia)  55

**General:** Caused by fetus swallowing one or more of the free-floating strands that result from amniotic rupture; the tension of these strands intraorally and extraorally produces secondary tears and deformations; no hereditary factor known.

**Ocular:** Upward slant of palpebral fissures; bilateral upper and lower lid colobomas; telecanthus; bilateral corneal opacities; microphthalmos; strabismus; hypertelorism; epibulbar choristoma; unilateral chorioretinal defects or lacuna (rare).

**Clinical:** Craniofacial and limb abnormalities.

Amyloidosis of Gingiva and Conjunctiva, with Mental Retardation (Primary Systemic Amyloidosis) 56

**General:** Autosomal recessive; primary amyloidosis differs from secondary by the mesodermal tissues being affected and nodular form of deposits; no preexisting medical condition; preferential involvement of mesenchymal tissues; variable staining of deposits.

**Ocular:** Conjunctivitis with deposits; corneal leukoma; waxy eyelid papules with purpura; ptosis; decreased vision; keratitis sicca; upper lid mass; tonic pupil; accommodative paresis; diffuse yellow conjunctival mass.

**Clinical:** Hyperplastic gingivitis, tongue, skin, and muscles; lungs with icing-like coating; mental retardation; peripheral neuropathy; congestive heart failure; polyarthropathy; spontaneous, incidental purpura; macroglossia; bleeding diathesis; idiopathic carpal tunnel syndrome.


Andersen-Warburg Syndrome (Whitnall-Norman Syndrome; Oligophrenia Microphthalmos Syndrome; Norrie Disease; Atrophia Oculi Congenital Fetal Iritis Syndrome; Congenital Progressive Oculo-Acousto-Cerebral Dysplasia) 57

**General:** Sex-linked inheritance; gross deformation of both eyes; only males affected; onset at birth; putative gene for Norrie disease has been isolated and mapped to Xp11.3.

**Ocular:** Bilateral microphthalmos with extensive destruction of all ocular structures often resembling a pseudotumor; blindness at birth; iris atrophy; iritis; corneal opacification and lenticular destruction with a mass visible behind the lens as long as the lens is still clear; malformed retina and choroid with retinal pseudotumors; retinal detachment; retrorenal vascular mass.

**Clinical:** Mental retardation ranging from imbecility to idiocy (may begin at any age) in about two thirds of cases; deafness of differing severity with onset between ages 9 and 45 years.


Andogsky Syndrome (Atopic Cataract Syndrome; Dermatogenous Cataract) 58

**General:** Inherited abnormality involving the skin and lens with an altered reactivity to antigen.

**Ocular:** Atopic keratoconjunctivitis; keratoconus; uveitis; dense subcapsular cataract developing to a complete dense opacification.

**Clinical:** Atopic dermatitis as erythematous thickening of the skin with papular hyperpigmented and scaly changes, most frequently found in regions of the wrist, popliteal fossa, neck, and sometimes forehead.


Angiolymphoid Hyperplasia with Eosinophilia (Kimura Disease) 59

**General:** Benign, slow-growing tumor involving primarily the face and scalp; etiology unknown but inflammatory reaction suspected; occurring primarily in young Oriental males.
Ocular: Proptosis; orbital tumor; upper lid swelling (may be bilateral).

Clinical: Single or multiple subcutaneous or dermal nodules; regional lymphadenopathy; peripheral blood eosinophilia; nephrotic syndrome.


Anemia

General: Ocular complications generally only seen in severe anemia.

Ocular: Palpebral conjunctival pallor; retinal hemorrhages; cotton-wool spots; retinal vein dilation; papilledema; ischemic optic neuropathy.

Clinical: Blood loss; excessive red blood cell destruction; inadequate red blood cell production; thrombocytopenia; leukemia.


Angelucci Syndrome (Critical Allergic conjunctivitis Syndrome)

General: Etiology unknown; pruriginous cutaneous and mucous reactions that appear and cease rather suddenly.

Ocular: Chemosis; conjunctivitis (papillary type); severe itching and burning; photophobia.

Clinical: Tachycardia; vasomotor lability; excitability; allergies (asthma, urticaria, edema); dystrophic conditions and endocrine disorders are frequently associated findings.


Angioedema (Angioneurotic Edema; Giant Edema; Giant Urticaria; Hives; Nettle Rash; Quincke Disease)

General: Vascular reaction involving subcutaneous tissues or submucosa; both sexes affected; allergy to various agents, including medications; emotional factor may be involved; recurrent.

Ocular: Optic neuritis; papiledema; central serous retinopathy; corneal edema; exophthalmos; nystagmus; secondary glaucoma; uveitis; periorbital and lid edema.

Clinical: Transient erythema; angioneurotic edema of loose subcutaneous tissue; sporadic urticaria; nausea; vomiting; diarrhea; cephalalgia; severe respiratory distress; polyuria.


Angular Conjunctivitis (Morax-Axenfeld Bacillus)

General: Caused by Moraxella lacunata, which frequently inhabits the nose.

Ocular: Conjunctivitis; hypopyon; keratitis; uveitis; corneal marginal ulcer.


Aniridia

General: Hereditary, recessive (two thirds of cases), can be dominant, sporadic, or traumatic; absence of the iris; rare; usually bilateral unless due to trauma.
Ocular: Absence of iris; subluxed lens; iridodialysis; cataract; glaucoma; corneal scarring, vascularization, and edema; iris colobomata; round eccentric pupils; keratoconus.

Clinical: Cerebellar ataxia; mental retardation; Wilms tumor.


### Aniridia and Absent Patella

General: Autosomal dominant; rare.

Ocular: Absence of iris; cataracts; glaucoma.

Clinical: Absence of knee cap, hypoplastic or aplastic.


### Aniridia, Cerebellar Ataxia, and Mental Deficiency (Gillespie Syndrome)

General: Autosomal recessive; onset at birth.

Ocular: Congenital cataracts; incomplete formation of iris; bilateral congenital mydriasis.

Clinical: Cerebellar ataxia; mental deficiency; delayed developmental milestones; persistent hypotonia of muscles; gross incoordination; attention tremor; scanning speech.


### Aniridia, Partial with Unilateral Renal Agenesis and Psychomotor Retardation

General: Autosomal recessive.

Ocular: Congenital glaucoma; telecanthus; absence of iris; hypertelorism.

Clinical: One kidney absent or in failure; motor effects of cerebral or psychic activity retarded or slowed.


### Ankyloblepharon Filiforme Adnatum and Cleft Palate (Hay-Wells Syndrome; AEC Syndrome)

General: Autosomal dominant; cleft palate and/or cleft lip; congenital filiform fusion of eyelids.

Ocular: Filiform fusion of eyelids; pterygium; keratoconus.
Clinical: Cleft lip and palate; paramedian mucous pits of lower lip; ectodermal dysplasia; infrequent association with trisomy 18; partial thickness fusion of central portion of lid margins.


Ankylostomiasis (Hookworm Disease)

General: Causative agents include Necator americanus, Ancylostoma duodenale, Ancylostoma braziliense, and Ancylostoma caninum; final diagnosis depends upon finding eggs in feces.

Ocular: Retinal hemorrhages around optic disk; diplopia; conjunctival xerosis; visual field defects; cataract.

Clinical: Maculopapules; localized erythema; microcytic hypochromic anemia.


Annette Von Droste-Hulshoff Syndrome

General: Premature birth; pseudo strabismus due to macular ectopia in retinopathy of prematurity.

Ocular: Myopia; retinal detachment; negative and positive angle kappa; esotropia; temporal macular ectopia; chorioretinal colobomata; falciform folds; persistent hyaloids artery; abnormal position of blind spot; epicanthus; telecanthus; blepharophimosis; hypertelorism; asymmetrical orbits; exophthalmos; enophthalmos.

Clinical: Face turn; angioma; neoplasia.


Anorexia Nervosa (Apepsia Hystericci)

General: Compulsive neurosis; refusal to eat; occurs in adolescent to young adult females; symptomatic recovery or chronic course.

Ocular: Cataract; central retinal vein occlusion; myopathy of orbicularis oculi (weakness of eye closure).

Clinical: Severe cachexia; loss of hair; nausea; constipation; diarrhea; depression; vigorous activity; weight loss; menstrual disturbance.


Anoxic Overwear Syndrome

General: Caused by a reduction in oxygen supply due to continuously worn hydrogel lenses; allergic or toxic reactions to preservatives used in the cleaning process.

Ocular: Refractive error changes; endothelial cell changes; physical trauma to the anterior surface of the cornea; corneal neovascularization; giant papillary conjunctivitis; contact lens deposits; acute red eye syndrome.

**Anterior Capsule Contraction Syndrome**  
**General:** Associated with diabetes mellitus, myotonic muscular dystrophy, uveitis, shrinkage of anterior capsular opening after anterior continuous curvilinear capsulorrhesis  
**Ocular:** Uveitis, retinitis pigmentosa, high myopia, pseudoexfoliation syndrome  

**Anterior Chamber Cleavage Syndrome (Reese-Ellsworth Syndrome; Peters-Plus Syndrome)**  
**General:** Abnormalities in the embryologic development of the anterior chamber due to failure of normal migration of mesodermal cells across the anterior segment of the eye or failure of later differentiation of the mesodermal elements; various conditions described as congenital: central anterior synechiae, persistent mesenchymal tissue in the chamber angle, posterior embryotoxon, congenital corneal hyaline membrane, posterior marginal dysplasia, prominent Schwalbe line, mesodermal dysgenesis, and internal corneal ulcer seem all to fall in this same category of the anterior chamber cleavage syndrome; condition is present at birth; about 80% are bilateral; autosomal dominant inheritance; may be associated with congenital sensory neuropathy and ichthyosis.  
**Ocular:** Increased intraocular pressure; adhesions between the iris and cornea; persistence of mesenchymal tissue in the chamber angle; usually shallow anterior chamber; iris coloboma and hypoplasia; prominent Schwalbe ring; contiguous hyaloid membrane; corneal opacities of various density with or without edema, usually at the site of iris adhesion; anterior pole cataract; remains of hyaloid artery.  
**Clinical:** Dental anomalies; mental retardation; cleft palate; syndactyly; craniofacial dysostosis; myotonic dystrophy.  

**Anterior Segment Ischemia Syndrome**  
**General:** Occasional complication of strabismus surgery; usually occurs in adult patients who have paretic strabismus after extensive transposition procedures; also may be secondary to giant cell arteritis or develop following trabeculectomy or strabismus surgery.  
**Ocular:** Corneal edema; corneal ulceration; uveitis; iris atrophy; ectopic pupil; posterior synechiae; cataract; hypotony; phthisis bulbi.  

**Anterior Spinal Artery Syndrome (Ventral Medullary Syndrome; Myelomalacia Syndrome; Beck Syndrome; Medullary Syndrome)**  
**General:** Impaired blood supply to the anterior two thirds of the spinal cord, which includes the corticospinal and lateral spinothalamic tracts and anterior horns of the gray matter; caused by thrombosis, aneurysms, or extramedullary tumors; onset is sudden, and symptoms are preceded by pain and paresthesias.  
**Ocular:** Nystagmus.  
**Clinical:** Quadriplegia (usually sudden with cervical lesions) with loss of sense of position; arms may be unimpaired with thoracic location of the lesion; anesthesia for determination of sensations; disturbed intestinal and bladder function with incontinence; disturbed sense for temperature and pain.  
Anthrax

**General:** Disease of wild and domestic animals; transmitted to humans by contact with animals or their products; causative agent is *Bacillus anthracis.*

**Ocular:** Pustules and edema of lids; phlebitis of ophthalmic veins; optic neuritis; optic atrophy; panophthalmitis; itchy, erythematous papule of the eyelids.

**Clinical:** NECrotic cutaneous ulcer.


Tasman IS. *The eye manifestations of internal diseases.* St. Louis: CV Mosby, 1951;315-317.

**Antimongolism Syndrome (Chromosome 21 Partial Deletion Syndrome; Monosomy 21 Partial Syndrome; G-Deletion Syndrome)**

**General:** Partial monosomy of chromosome 21 with absence of the short arm and part of the long arm of this chromosome.

**Ocular:** Antimongoloid slant of lid fissures; blepharochalasis; sclerocornea.

**Clinical:** Hypertony; large ear lobes; prominent nasal bridge; mental retardation; pyloric stenosis; dystrophic nails; retarded growth; heart disease; hemivertebrae; micrognathia.


**Antiphospholipid Antibody Syndrome (Hughes syndrome)**

**General:** Recurrent arterial and venous thrombosis

**Ocular:** Subconjunctival hemorrhage, hyphema, vitreous hemorrhage, central retinal vein and artery occlusions


**Anton Syndrome (Denial-Visual Hallucination Syndrome)**

**General:** Cause unknown, but isolation of diencephalon from occipital lobe would be necessary to result in the features of the syndrome; lesions of the calcarine-thalamic connections or bilateral destruction of the occipital regions have been claimed to cause denial of blindness; the disease is rare and little understood; has been reported in association with blindness from a peripheral lesion such as bifrontal contusions and optic nerve damage.

**Ocular:** Denial of blindness; patients may persistently deny having any loss of visual perception; the objects the patient describes and claims to see are regarded as visual hallucinations; visual field hemianopsia.

**Clinical:** Confabulation; allocheiria (reference of a sensation is made to the opposite side to which the stimulus is applied).


**Apert Syndrome (Acrocephalosyndactylism Syndrome; Acrocrani-o-dysphalangia; Acrodysplasia; Sphenacrocra nio-syndactyly; Absent-Digits-Cranial-Defects Syndrome)**

**General:** Inherited; most often recessive, sometimes dominant; an extreme form of Apert syndrome has been described as Carpenter syndrome, with the latter being familial and transmitted as an autosomal recessive.
Ocular: Shallow orbit; exophthalmos; hypertelorism; ptosis; strabismus; nystagmus; ophthalmpoplegia; hyperopia; exposure keratitis; cataracts; ectopia lentis; medullated nerve fibers; retinal detachment; papilledema with subsequent optic atrophy; keratoconus.

Clinical: Oxycephaly ("tower skull"); syndactyly (symmetrically); synostoses and synarthroses of shoulder and elbows frequent; agenesis of spinal bones and limbs; headaches; hypertelorism; hypoplastic maxilla; acrocephaly; abnormality of sutures.


Arcus Cornea (Arcus Senilis)  82

General: Autosomal dominant; occurs earlier in blacks; frequently associated with abnormal serum lipid levels, but may occur without any predisposing factors; more commonly associated with familial hypercholesterolemia.

Ocular: Arcus of cornea.

Clinical: Can be associated with Tangier disease, Norum disease, and homozygotes type II hyperlipoproteinemia; osteogenesis imperfecta ring resembling arcus seen.


Argininosuccinic Aciduria (Trichorrhexis Nodosa)  83

General: Deficient argininosuccinase (A Sase) deficiency; both sexes affected; prevalent in females; autosomal recessive inheritance.

Ocular: Friable tufted eyelashes and eyebrows; visual field defects; cataract.

Clinical: Clinical findings vary widely; mental retardation; seizures; ataxia; hepatomegaly; friable hair (trichorrhexis nodosa); may have citrullinemia; hyperammonemia; increased argininosuccinic acid (most pronounced in the cerebrospinal fluid).


22
**Argyll Robertson Syndrome (Spinal Miosis)**

**General:** Caused by syphilis or, rarely, epidemic encephalitis; disseminated sclerosis; diabetes; brain tumor; syringomyelia; syringobulbia; chronic alcoholism; injury; encephalitis lethargica, Guillain-Barré syndrome, Lyme disease, multiple sclerosis, polyarteritis nodosa, and sarcoidosis have been associated with this condition.

**Ocular:** No direct or consensual pupil reaction to light but to normal accommodation (except in terminal stages, when pupil is fixed to all stimuli); pupil contraction with eserine but poor dilation with atropine; miosis (generally); irregular pupil; occurs unilaterally and bilaterally; anisocoria or discoria frequent.

**Clinical:** Syphilis of central nervous system; general paresis; tabes dorsalis.


**Arndt-Gottron Syndrome (Scleromyxedema)**

**General:** Etiology unknown; rare cutaneous disease of hyaluronic acid deposits in dermis; variant of lichen myxedematosus (papular mucinosis); progressive disease commonly involving the face, neck, upper trunk, forearms, and hands, producing thickening of the skin with overlying fine papules.

**Ocular:** Corneal opacities of amyloid deposits; thickening of eyelids; lagophthalmos; ectropion; thickened eyebrow or eyelid skin; corneal opacities.

**Clinical:** Exaggerated facial folds impair opening of the mouth; flexion contractures from poor joint mobility; erythema; scaling of skin; phimosis; urethral stenosis.


**Arnold Pick Syndrome (Aphasia-Agnosia-Apraxia Syndrome; Pick Syndrome [2]; Pick Disease of the Brain)**

**General:** Widespread cortical atrophy; manifested between 40 and 70 years; pathogenesis remains unknown; cannot be consistently differentiated from Alzheimer disease on clinical grounds alone.

**Ocular:** Apperceptive blindness (inability of patient to fix upon objects within his or her gaze); visual agnosia (inability to recognize familiar objects by sight); visual field defects due to atrophy in occipital lobe.

**Clinical:** Presenile or progressive dementia; patient is unaware of his or her surroundings; poor insight; loss of words and utterance of stereotyped phrases; aphasia (motor type); apathy and indifference.


**Arnold-Chiari Syndrome (Platybasia Syndrome; Cerebellomedullary Malformation Syndrome; Basilar Impressions)**

**General:** Malformation of the hindbrain; developmental deformity of the occipital bone and upper cervical spine; recognized in children or adults; clinical picture may be indistinguishable from that of Dandy-Walker syndrome in infants.

**Ocular:** Horizontal, vertical, and rotary forms of nystagmus; vertical nystagmus in both up gaze and down gaze is most common; papilledema; esotropia; Duane retraction syndrome (association); oscillopsia.

**Clinical:** Hydrocephalus; cerebellar ataxia; bilateral pyramidal tract signs.


Arterial Occlusive Retinopathy and Encephalopathy Syndrome

**General:** Rare; etiology unknown but may be virally induced, immune-mediated disease; most frequent in women; mechanism could be related to microangiopathy secondary to immunologically mediated vasculitis, although an abnormal coagulation system or microembolisms have been proposed to explain this condition.

**Ocular:** Multiple branch retinal arterial occlusions; rotary nystagmus; retinal hemorrhage; visual field defects; bilateral gaze palsy.

**Clinical:** Encephalopathy; behavior and memory disturbances; hearing loss; paranoid psychosis; neurologic dysfunction; seizures; headache; spasticity and hyperreflexia.


Arteriosclerosis

**General:** Thickening and induration of the arterial wall; prominent in the elderly.

**Ocular:** Increased arterial light reflex, copper/silver wire arteries; arteriovenous crossing changes; arterial caliber variation/irregularity; arterial straightening or tortuosity; intimal hyperplasia, medial atrophy, atherosclerotic fibrous plaques and calcifications of the internal elastic lamina observed in aged human orbital arteries.

**Clinical:** Increased collagen deposition in small- and medium-sized arteries with progressive replacement of the smooth muscle in the vessel walls; arterial wall changes at arteriovenous crossings.


Arteriovenous Fistula (Arteriovenous Aneurysm; Arteriovenous Angioma; Arteriovenous Malformation; Cirsoid Aneurysm; Racemose Hemangioma; Varicose Aneurysm)

**General:** Abnormal communications between arteries and veins that allow arterial blood to enter the vein directly without traversing a capillary network; may be congenital or secondary to penetrating trauma or blunt trauma.

**Ocular:** Uveitis; chemosis and neovascularization of conjunctiva; bullous keratopathy; eyelid edema; ptosis; exophthalmos; iris atrophy; papilledema; retinal hemorrhages; cataract; paresis of third or sixth nerves; glaucoma; upper lid tumor; total choroidal detachment; leaking retinal macroaneurysms; central retinal vein occlusion; iris neovascularization.

**Clinical:** Cerebral hemorrhage; death; substernal pain; dyspnea; varicose veins.


Arthrogryposis Multiplex Congenita

**General:** Heterogeneous group of disorders of multiple proposed etiologies; often one manifestation of a complex of congenital anomalies; probable autosomal recessive transmission; found in Eskimos; affects more males than females; characterized by decreased fetal joint mobility secondary to neuropathic disease, myopathic disease, or some other cause.

**Ocular:** Congenital bilateral cataract; associated with ophthalmoplegia, retinopathy, goniodysgenesis, and infantile glaucoma, as well as Duane retraction syndrome.

**Clinical:** Multiple articular rigidities; hypoplasia of adjacent muscle groups; soft tissue shortening; ducklike waddle; muscle atrophy.


**Arylsulfatase A Deficiency** (Metachromatic Leukodystrophy; Sulfatide Lipidosis Syndrome; Greenfield Disease; Scholz Syndrome; Scholz-Bielschowsky-Henneberg Syndrome; Van Bogaert-Nyssen Disease; Van Bogaert-Nyssen-Peiffer Disease; Familial Progressive Cerebral Sclerosis; Infantile Progressive Cerebral Sclerosis; Infantile Metachromatic Leukodystrophy; Leukodystrophia Cerebri Progressiva Metachromatica Diffusa; Opticochleodentate Degeneration)

**General:** Accumulation of sulfatide caused by deficient activity of arylsulfatase A; autosomal recessive; familial form of metachromatic leukodystrophy; Greenfield disease (late infantile form); van Bogaert-Nyssen-Peiffer syndrome (adult form); affects central and peripheral nervous systems by demyelination and by accumulation of metachromatic material.

**Ocular:** Visual loss in association with optic atrophy; strabismus; macular cherry-red spot; corneal opacification; oculomotor disorders (nystagmus, strabismus); optic nerve and retinal demyelination.

**Clinical:** Motor and mental deterioration with spasticity; paralysis; seizures; dementia; death in early childhood, although attenuated and adult forms of the disease occur; schizophrenia; temporoparietal demyelination; unreactive to visual and auditory stimuli; adult form: moodiness, withdrawal, megalomania, hallucinations, violent reactions, and dementia.


**Ascariasis**

**General:** Roundworm infection caused by *Ascaris lumbricoides*.

**Ocular:** Conjunctivitis; xerosis; periphlebitis; pigmentation of macular lesion; papilledema; uveitis; subluxation of lens; scotoma; secondary glaucoma; vitreous hemorrhages; possible association with phlyctenular eye disease.

**Clinical:** Occasional colicky abdominal pain; slight abdominal distention; pneumonitis; intestinal obstruction.


**Ascher Syndrome (Blepharochalasis with Struma and Double Lip)**

**General:** Rare occurrence; blepharochalasis transmitted as a simple dominant; related to development of thyroid gland; symptoms usually start around puberty.

**Ocular:** "Bulging" of orbital fat; blepharochalasis; protrusion of lacrimal gland; entropion (rare).
Clinical: Goiter; reduplication of upper lip; hypothyroidism; alopecia areata totalis.

Aspergillosis

General: Systemic infection common in poultry farmers, feeders or breeders of pigeons, and persons who work with grains; should be considered in immunocompromised patients.

Ocular: Corneal ulcer; blepharitis; keratitis; endophthalmitis; exophthalmos; retinal hemorrhages; retinal detachment; vitreitis; cataract; conjunctivitis; orbital cellulitis; paresis of extraocular muscles; secondary glaucoma; scleromalacia perforans; endogenous endophthalmitis; anterior chamber mass; invasion of choroid and anterior optic nerve.

Clinical: Pulmonary infections; invasive fungal disease.

Asthma (Hayfever)

General: Asthma characterized by paroxysms of expiratory dyspnea and wheezing, overinflation of the lungs, cough, and rhonchi; causes include allergy to external inhaled allergens, respiratory infections, and psychophysiologic reaction to stress. Hayfever (allergic asthma) characterized by sneezing, rhinorrhea, swelling of nasal mucosa, and itchy eyes; caused by spread of pollens in air or exposure to antigens; seasonal; occurs most frequently in young persons.

Ocular: Lacrimation; allergic conjunctivitis; periorbital xanthogranulomas.

Clinical: Rhinorrhea; sneezing; mucosal swelling with occlusion of airway; insomnia; nasal polyps; wheezing; cough; headache; rhinitis.


Ataxia, Spastic, with Congenital Miosis

General: Autosomal dominant.

Ocular: Congenital miosis; nystagmus; small, nonreacting pupils.

Clinical: Symmetrical ataxia of gait and limb movement; dysarthria; late in walking; slurred speech; increased deep tendon reflexes; extensor plantar reflexes.


Atherosclerosis

General: Etiologic importance of lipid infiltration; cholesterol; patchy nodular form of arteriosclerosis.

Ocular: Central retinal artery obstruction; branch retinal artery obstruction; most common cause is embolization from carotid plaques.

Clinical: Atheroma, which is a nodular, subendothelial lipid deposition in the intima of large- and medium-sized arteries.

Atopic Dermatitis (Atopic Eczema; Besnier Prurigo)

**General:** Highly specific disease resulting from a heredity determined lowered cutaneous threshold to pruritus and characterized by intense itching; elevated total and specific immunoglobulin E.

**Ocular:** Keratoconjunctivitis; keratoconus; cataract; atopic dermatitis of lid; secondary glaucoma; uveitis; possible association with retinal detachment; pannus; blepharoconjunctivitis; corneal scarring; suppurative keratitis.

**Clinical:** In infants it involves the face with dry or oozing erythematous patches; in children and adolescents itching localized in the neck, antecubital spaces, popliteal folds, and ears; seborrheic changes.


Autoimmune Corneal Endotheliopathy

**General:** Etiology unknown; associated with implantation of an intraocular lens (IOL), pars planitis, iritis, secondary herpetic keratitis, and corticosteroid use; rare.

**Ocular:** Stromal edema; migrating line of keratic precipitates; iritis; clouding of the cornea; lymphocytes and macrophages in anterior chamber; linear pigmented endothelial line; evidence supports that herpes simplex virus can be isolated in the aqueous humor of patients with this condition.

**Clinical:** Autoimmune disease, nonspecific.

Autoimmunologically Mediated Syndrome (Lymphocytic Hypophysitis Associated with Dacryoadenitis Syndrome)

**General:** Lymphocytes infiltrate the hypophysis.

**Ocular:** Dacryoadenitis.

**Clinical:** Lymphocytic infiltration of the hypophysis by CD3 cells, T cells, and CD20+ B cells is an autoimmune process that may rarely cause lacrimal gland swelling.

Avitaminosis B2 (Ariboflavinosis; Pellagra)

**General:** Niacin deficiency.

**Ocular:** Conjunctivitis; corneal vascularization; keratitis; pupillary dilation; optic atrophy; optic neuritis; cataract; blepharitis; central scotoma; marked photophobia.

**Clinical:** Occasional cranial nerve palsies; dermatitis; glossitis; gastrointestinal and nervous system dysfunction; mental deterioration; diarrhea; stomatitis.
**Avitaminosis C (Scurvy; Vitamin C Deficiency)**

**General:** Vitamin C deficiency.

**Ocular:** Hemorrhages of lids, anterior chamber, vitreous cavity, retina, subconjunctival space, and orbit (most prominent, with resulting exophthalmos); keratitis, corneal ulcer; cataract.

**Clinical:** Increased capillary fragility with a tendency to hemorrhage in tissues throughout the body; poor wound healing; loose teeth; purpuric rash.


**Avulsed Retinal Vessel Syndrome**

**General:** Visual prognosis good; reports of avulsed retinal vessels not associated with retinal breaks.

**Ocular:** Recurrent vitreous hemorrhages caused by an avulsed retinal vessel, during retinal tear formation; vitreous hemorrhages may recur until interruption of vessel occurs.

Bustros SD, Welch PB. The avulsed retinal vessel syndrome. *Ophthalmology* 1982; 89[Suppl]: 189.


**Axenfeld-Rieger Syndrome (Posterior Embryotoxon; Axenfeld Syndrome)**

**General:** Dominant inheritance; occasionally sporadic; variable in expression.

**Ocular:** Posterior embryotoxon: ringlike opacity of cornea; long trabecula; prominent Schwalbe line; iris adhesions to Schwalbe line and cornea with large abnormal iris processes or broad sheets of tissues of varying size and location; anterior layer of iris may appear hypoplastic; ectopia of the pupil not uncommon; polycoria occurs; ringlike opacity of the deep corneal layers extending several millimeters from the limbus in continuity with the sclera; keratoconus.


**Axenfeld-Schurenberg Syndrome (Cyclic Oculomotor Paralysis)**

**General:** Congenital manifestation; frequently unilateral.

**Ocular:** Cyclic oculomotor paralysis (paralysis alternating with spasm); during periods of paralysis, lid exhibits ptosis and affected eye is abducted; during spasm, lid is raised, deviation of affected eye is either inward or outward, and pupil is fixed and contracted.


Babinski-Nageotte Syndrome (Medullary Tegmental Paralysis)  

**General:** Lesion in pontobulbar transitional region (corpus restiforme, Deiters nucleus, sympathetic fibers); Horner triad is always part of this syndrome; the findings are similar to Cestan-Chenais syndrome and Wallenberg syndrome; rare condition caused by ischemic lesion of the medulla oblongata involving the unilateral and medial areas of the medulla.

**Ocular:** Enophthalmos; ptosis; nystagmus; miosis.

**Clinical:** Contralateral hemiparesis and disturbance of sensibility; ipsilateral cerebellar hemiataxia; perhaps ipsilateral analgesia of the face, vocal cord, and soft palate; adiadochokinesis; lateral pulsion; dysmetria.


Bacillus Cereus

**General:** Highly virulent pathogen; most common contaminant of drug injection paraphernalia; usually enters the body as a result of penetrating trauma with a contaminated metallic foreign object; cause of food poisoning is toxin induced.

**Ocular:** Hypopyon; ring abscess of cornea; panophthalmitis; phthisis bulbi; orbital cellulitis; proptosis; vitreous abscess; necrosis of retina; endophthalmitis; keratitis.

**Clinical:** Fever; leukocytosis; septicemia; meningitis; endocarditis; osteomyelitis; wound infection.


Tuazon CU, et al. Serious infections with Bacillus sp. *JAMA* 1979; 241:1137-1140.

Bacillus Subtilis (Hay Bacillus)

**General:** Gram-positive rod found in air, soil, dust, water, milk, and hay; frequently seen in people who work near hay.

**Ocular:** Conjunctivitis; ring abscess of cornea; corneal ulcer; endophthalmitis; panophthalmitis; dacryocystitis; orbit abscess.

**Clinical:** Fever; leukocytosis.


Bacterial Endocarditis

**General:** Inflammation of the lining on the heart caused by an infective agent.

**Ocular:** Conjunctival petechial hemorrhages; retinal hemorrhages; Roth spots; cotton-wool spots; branch or central retinal arterial occlusion; metastatic endophthalmitis; iridocyclitis; optic disk edema; cranial nerve palsies; diplopia; nystagmus; choroidal abscess; choroidal neovascular membrane; anterior segment necrosis.

**Clinical:** Emboli of the central nervous system; fever; splenomegaly; heart murmur; embolic episodes.


Balint Syndrome (Psychic Paralysis of Visual Fixation Syndrome) 111

General: Bilateral lesion of parietooccipital region; rare occurrence; affected patients are unaware of objects otherwise familiar to them.

Ocular: Psychic paralysis of visual fixation; lack of full voluntary control of eye movements; unstable visual fixation.

Clinical: Tonic and motor phenomena of upper limbs; loss of body coordination (bilateral); optic ataxia; it has been reported to occur in association with human immunodeficiency virus (HIV) encephalitis and with presenile-onset cerebral adrenoleukodystrophy.


Baller-Gerold Syndrome (Craniosynostosis Radial Aplasia) 112

General: Autosomal recessive inheritance.

Ocular: Ocular hypertelorism; epicantthic folds.

Clinical: High nasal bridge; low philtrum; dysplastic ears; radius hypoplastic or absent; ulna short and bowed; carpal bones missing or fused; thumb hypoplastic or absent; craniosynostosis; anal, urogenital, cardiac, central nervous system, and vertebral defects; agenesis of frontal and parietal bones; midline facial angioma; scrotally positioned anus; microcephaly; erythroblastosis of the liver; pancreatic islet cell hypertrophy.


Bamatter Syndrome (Osteoplastic Geroderma; Walt Disney Dwarfism) 113

General: Hereditary X-linked; rare; onset in early childhood; precocious aging; osteoporosis; autosomal recessive inheritance.

Ocular: Glaucoma; microphthalmia; microcornea; corneal opacities.

Clinical: Senile changes in skin; stunted growth; articular hypertrophy; multiple fractures and bone malformations; osteodysplasia; osteoporosis; dwarfism.


Bang Disease (Brucellosis; Malta Fever; Mediterranean Fever; Pig Breeder Disease; Gibraltar Fever; Undulant Fever) 114

General: Transmitted to man from animals or animal products containing bacteria of the genus Brucella; human infection results from ingestion of infected animal tissue and milk products or through skin wounds directly bathed in freshly killed animal tissues.
Ocular: Conjunctivitis; punctate keratitis; optic neuritis; swollen optic nerves; chorioretinitis; extraocular muscle palsies; phlyctenules; dacyroadenitis; papilledema; episcleritis; macular edema; phthisis bulbi; uveitis; vitreous opacities; changes in intraocular pressure (early decrease or late increase).

Clinical: Fever; icterus; weakness; sweats; general malaise; mammary abscess.


**Banti Disease (Chronic Congestive Splenomegaly; Fibrocongestive Splenomegaly; Splenic Anemia; Hepatolienal Fibrosis)**

General: Etiology portal hypertension due to thrombosis, compression, or aneurysm; insidious or sudden onset; most frequently occurs before age 35 years; slowly evolving.

Ocular: Subconjunctival hemorrhage.

Clinical: Pallor; mild jaundice or brown pigmentation of skin; enlarged liver; weakness; melena; flatulence; diarrhea; epistaxis; vomiting of blood.

Banti G. Dell'Anemia Splenica. *Arch Sc Anat Patol (Firenze)* 1883; 2:53-122.


**Baraitser-Winter Syndrome**

General: X-linked mental retardation, macrosomia, macrocephaly, and obesity syndrome.

Ocular: Ptosis; hypertelorism; down-slanting palpebral fissures.

Clinical: May be confused with Noonan syndrome; phenotypic features appear to be variable.


**Bardet-Biedl Syndrome**

General: Polydactyly; obesity; cognitive delay; retinal degeneration; nystagmus.

Ocular: 30% to 65% of patients have clinical nystagmus that may mimic spasmus nutans but either lacks head nodding or is not suppressed by head nodding; retinal degeneration with attenuated retinal vessels and pale optic disks.

Clinical: Nystagmus may be a presenting sign, but patients will have polydactyly, obesity, and motor/cognitive delay.


**Bare Lymphocyte Syndrome**

General: Rare, severe combined immunodeficiency characterized by the lack of expression of human leukocyte antigen (HLA) A, B, and C antigens with severe T and B deficiency.

Ocular: Horizontal nystagmus; candida retinitis.
**Clinical:** Recurrent pulmonary infections; bronchiectasis; gastroenteritis; hepatomegaly; developmental delay; respiratory failure; death.


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### Barlow Syndrome (Mitrval Valve Prolapse)

<table>
<thead>
<tr>
<th>General</th>
<th>Common; usually benign; asymptomatic; predominant in females; vague with psychoneurotic basis.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Retinal branch arterial occlusion; retinal emboli; keratoconus; ophthalmic migraine; amaurosis fugax; bilateral retinal artery occlusion.</td>
</tr>
</tbody>
</table>

**Clinical:** Myxomatous degeneration of the mitral valve; palpitation; chest pain; dyspnea; hyperventilation.


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### Barre-Lieou Syndrome (Posterior Cervical Sympathetic Syndrome)

<table>
<thead>
<tr>
<th>General</th>
<th>Irritation of the vertebral nerve causing circulatory disturbance in the area of the cranial nuclei; fifth and eighth nerves mainly involved; trauma and arthritic changes involving the third and fourth cervical vertebrae or cervical disk pathology may be etiologic factor; course chronic; occurrence in older patients.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Reduced vision (transitory); corneal hypesthesia in association with persistent corneal ulcers confined to the lid fissure.</td>
</tr>
</tbody>
</table>

**Clinical:** Headache; vertigo; mild dizziness; vasomotor disturbances of face and facial pain; laryngeal and pharyngeal paresthesia; chronic cervical arthritis; ear noises are frequent; anxiety; depression; impaired memory; difficulty in thinking.


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### Barrier Deprivation Syndrome (Binkhorst Membrane Deprivation Syndrome; Worst Decompartmentalization of Eye Syndrome)

<table>
<thead>
<tr>
<th>General</th>
<th>Intracapsular cataract extraction; trauma to posterior capsule with extracapsular cataract extraction; more frequent in blue-eyed patients; often bilateral; cause thought to be increased pigment loss, which releases prostaglandins, creating allergic reaction.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Cystoid macular edema; corneal endothelial dystrophy; retinal detachment; leakage in peripheral retina and macula; iris pigment loss; uveitis; vitreous in anterior chamber; retinal holes; band keratopathy; glaucoma; iritis.</td>
</tr>
</tbody>
</table>

**Clinical:** None.


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### Bartsocas-Papas Syndrome

<table>
<thead>
<tr>
<th>General</th>
<th>A rare autosomal recessive variant of popliteal pterygium syndrome (PPS).</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Ptergium.</td>
</tr>
</tbody>
</table>

**Clinical:** Popliteal webbing; cleft lip; cleft palate; lower lip pits; syndactyly; genital and nail abnormalities; equinus feet.


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### Basal Cell Carcinoma

<table>
<thead>
<tr>
<th>General</th>
<th>Most common malignant neoplasm of lids; it can occasionally occur as a primary basal cell cancer of the conjunctiva and in the lacrimal canaliculus.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Neoplasm most common on lower lid and medial canthus; lacrimation.</td>
</tr>
</tbody>
</table>

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Clinical: Tumors of skin and other regions, including sinuses.

Basal Cell Nevus Syndrome (Nevoid Basal Cell Carcinoma Syndrome; Nevoid Basalioma Syndrome; Gorlin Syndrome; Gorlin-Goltz Syndrome; Multiple Basal Cell Nevi Syndrome)  124

General: Autosomal dominant; onset of skin lesions in childhood, usually at puberty.
Ocular: Basal cell carcinomas of eyelids; strabismus; hypertelorism; congenital cataracts; choroidal colobomas; glaucoma; medullated nerve fibers; prominence of supraorbital ridges; corneal leukoma; basalioma of the skin; coloboma of the choroid and optic nerve.

Clinical: Basal cell tumors with facial involvement; shallow pits of the skin of the hands and feet; jaw cysts; rib anomalies; kyphoscoliosis and fusion of vertebrae; medulloblastoma; frontal and temporoparietal bossing; broad nasal root.


Basedow Syndrome (Graves Disease; Hyperthyroidism; Thyrotoxicosis; Exophthalmic Goiter; Parry Disease)  125

General: Diffuse toxic goiter; inherited as a simple autosomal recessive; penetrance greater in females; however, dominant mode of inheritance and variable penetrance are possible; uncommon in either sex before age 15 years.
Ocular: Exophthalmos; swelling of eyelids and discoloration of upper eyelids; lid lag (von Graefe); globe lag (Koeber); lid trembling on gentle closure (Rosenbach sign); reduced blinking (Stellwag); retraction of upper lid; difficulty in evertting upper lid (Gifford sign); convergence weakness (Möobius); impaired fixation on extreme lateral gaze (Suker); possible external ophthalmoplegia (Ballet); Dalrymple sign (staring appearance); tearing; photophobia; epiphora; prolapse of lacrimal gland; neuroretinal edema; tortuous vessels; papilledema and papillitis; anisocoria; keratitis; increased intraocular pressure; increased intraocular pressure on upgaze; decreased visual acuity; enlargement of the extraocular muscles; increased volume of the extraocular muscles; superior rectus muscle enlargement; decreased venous outflow.

Clinical: Tachycardia; anxiety; insomnia; loss of weight; hyperhidrosis; restlessness; myocarditis (toxic); atrial fibrillation.


Bassen-Kornzweig Syndrome (Abetalipoproteinemia; Acanthocytosis; Familial Hypolipoproteinemia)  126

General: Inability to absorb and transport lipids; predominant in males; autosomal recessive inheritance; acanthocytosis, a peculiar burl cell malformation of the red blood cells; the basic defect is thought to be an inability to synthesize the apolipoprotein B peptide of low-density and very-low-density lipoproteins.
Ocular: Ptosis (may be present); nystagmus; progressive external ophthalmoplegia; retinitis pigmentosa (usually atypical); retinopathy develops with age after 10 to 14 years; optic atrophy occasionally; epicantal folds; cataract; optic nerve pallor; hypopigmentation of retina; macular degeneration; dyschromatopsia.
Clinical: Steatorrhea; hypcholesterolemia; neurologic disorder with ataxia (similar to Friedreich ataxia); areflexia; Babinski sign; muscle weakness (facial, lingual; proximal and distal); slurred speech; lordosis; kyphosis.


Batten-Mayou Syndrome (Spielmeyer-Vogt Syndrome; Mayou-Batten Disease; Stock-Spielmeyer-Vogt Syndrome; Cerebroretinal Degeneration; Pigmentary Retinal Lipoid Neuronal Heredodegeneration; Vogt-Spielmeyer Syndrome; Juvenile Ganglioside Lipidosis; Neuronal Cereoid Lipofuscinosis; Myoclonic Variant of Cerebral Lipidosis; Batten Disease; Cerebromacular Dystrophy; Juvenile Amaurotic Family Idiocy; Spielmeyer-Sjögren Syndrome) 127

General: Autosomal recessive; some cases of autosomal dominant; possible disturbance in lipid metabolism; most common in Jewish families; onset between ages 5 and 8 years; mean age at death is 17 years; poor prognosis (see Tay-Sachs Disease; Dollinger-Bielschowsky Syndrome.) The lipopigment storage diseases are divided into four types based on clinical and electron microscopic features: infantile (Hagberg-Santavuori syndrome), late infantile (Jansky-Bielschowsky disease), juvenile (Spielmeyer-Vogt disease), and adult (Kufs disease).

Ocular: Vision initially reduced, progressing to total blindness; fat deposition in the retina with gradual development of pigment disturbances resembling retinitis pigmentosa; progressive primary optic atrophy; granular pigmentary change of macula; there is clinical evidence supporting the idea that the primary lesion of the retina is in the inner layers.

Clinical: Mental disturbances; convulsions (later); apathy; irritability; ataxia; upper and lower motor neuron palsies; rigidity; complete paralysis and dementia in terminal stage; hypertonus; death from intercurrent infection.


Bazzana Syndrome (Angioptastic Ophthalmo-Auricular Syndrome) 128

General: Rare.

Ocular: Visual fields have concentric contraction; retinal vascular tortuosity and irregular contours.

Clinical: Progressive deafness (bilateral), caused by otosclerosis.


BBB Syndrome (Hypertelorism-Hypospadias Syndrome; Opitz Syndrome) 129

General: X-linked inheritance possible; differentiated from G syndrome by facial features and onset in late childhood (see G Syndrome.) This disorder is compatible with normal intelligence and life span. The abnormal gene may be located in the duplicated region 5p13-p12.

Ocular: Epicanthal folds; strabismus; blepharophimosis; telecanthus; widely spaced eyebrows.
**Clinical:** High nasal bridge; hypospadias; cryptorchidism; cleft palate and lip; urinary malformations; mental retardation; osteochondritis dissecans; congenital heart defects; upper urinary tract anomalies.


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**Beal Syndrome**

**General:** Transient unilateral disease; becoming bilateral later, then resolving within 2 weeks.

**Ocular:** Acute follicular conjunctivitis (lymphoid follicles; cobblestoning of conjunctiva with rapid onset).

**Clinical:** No purulent discharge; associated with regional adenitis.


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**Beard Disease (Neurasthenia; Nervous Exhaustion)**

**General:** Predominantly in women who are overworked or emotionally strained; occurs usually in the fourth or fifth decade; onset gradual; episodic recurrence.

**Ocular:** Hippus (visible, rhythmic but irregular pupillary oscillation, deliberate in time, and 2 mm or more excursion; it has no localizing significance).

**Clinical:** Muscle spasms; body aches; autonomic nervous system involvement; tiredness; insomnia; impotence; dyspepsia; phobic neurosis.


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**Bee Sting of the Eye (Bee Sting of the Cornea)**

**General:** Occurs when the toothed lancet of the stinging apparatus penetrates the cornea.

**Ocular:** Conjunctival hemorrhage, chemosis, and hyperemia; corneal abscess; keratitis; lid edema; iris depigmentation; iridoplegia; iritis; lacrimation; apoplectic visual loss; acute disk swelling secondary to acute demyelination.

**Clinical:** Laryngeal edema; anaphylaxis; death; localized tissue edema; fever.


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**Behçet Syndrome (Dermato-Stomatoo-Ophthalmic Syndrome; Oculobuccogenital Syndrome; Gilbert Syndrome)**

**General:** Virus infection?; occurs in adults; chronic disease; complete remission is rare; etiology is unknown.

**Ocular:** Muscle palsies (occasional); nystagmus (occasional); conjunctivitis; hypopyon; iritis; recurrent uveitis; keratoconjunctivitis sicca; keratitis; vitreous hemorrhages; thrombophlebitis retinal veins (occasional); retinal hemorrhages; optic neuritis (occasional); macular edema; optic nerve atrophy; retinitis; secondary glaucoma; retinal vasculitis; disk edema; panophthalmitis; optic neuropathy; skin lesions, posterior uveitis and systemic complications have been associated with loss of vision with this disorder; corneal immune ring opacity.

**Clinical:** Aphthous lesions of mucous membranes of the mouth and genitalia; cerebellar signs; convulsions; paraplegia; skin erythema (multiforme, bullousum); arthritis; urethritis; glossitis; recurrent fever.


Behr Syndrome (Optic Atrophy Ataxia Syndrome)

**General:** Infantile form of heredofamilial optic atrophy and hereditary ataxia; autosomal recessive; rare; temporary progression that after some years leads to a static condition; both sexes equally affected, although transmission of pure hereditary optic atrophy shows marked predominance in males; in most cases, the abnormalities do not progress after childhood.

**Ocular:** Nystagmus; central scotoma; severe progressive temporal atrophy of the optic nerve; bilateral retrobulbar neuritis; horizontal nystagmus.

**Clinical:** Pyramidal tract signs (increased tendon reflexes and positive Babinski sign); ataxia and disturbance of coordination; mental deficiency; vesical sphincter muscle weakness; muscular hypertonia; clubfoot; progressive spastic paraplegia; dysarthria; head nodding.


Bell Palsy (Idiopathic Facial Paralysis)

**General:** Unilateral facial nerve paralysis of sudden onset and gradual recovery involving the nerve as it runs through the fallopian canal; etiology unknown; more common in adults.

**Ocular:** Corneal ulcer; paralysis of seventh nerve; ectropion; lagophthalmos; ptosis; epiphora; decreased visual acuity; diplopia; ocular irritation; exposure keratitis.

**Clinical:** Aching in the ear or mastoid; tingling or numbness of cheek or mouth; alteration of taste; hyperacusis; epiphora; facial weakness; most commonly and frequently affected cranial nerve with herpes zoster is the facial nerve.


Benedikt Syndrome (Tegmental Syndrome)

**General:** Lesion of the inferior nucleus tuber with obstruction of the third nerve; arteriosclerotic occlusion of branches of the basilar artery, trauma and hemorrhages in the midbrain, and neoplasm most common causes.

**Ocular:** Homolateral paralysis of cranial nerve III (oculomotor); involves associated movements of convergence, elevation, and depression of the eyes; loss of reflex to light and accommodation.

**Clinical:** Unilateral hyperkinesis; contralateral hemiparesis, coarse tremor of upper extremity (greatly increased during movement), hemihypesthesia, and absent deep sensibility; ipsilateral ataxia. There is at least one reported case of an HIV-positive patient with Benedikt syndrome who had elevated immunoglobulin G (IgG) toxoplasma IgG titers.


Benign Mucosal Pemphigoid (Chronic Cicatricial Conjunctivitis; Cicatricial Pemphigoid; Essential Shrinkage of the Conjunctiva; Membrane Pemphigus; Ocularpemphigoid)  

**General:** Etiology unknown; involving older age group, especially over 70 years; chronic autoimmune disorder characterized by fibrosis beneath the conjunctival epithelium; associated with the major histocompatibility complex class I alleles, which confer susceptibility to the disease; likely due to a multigene effect and associated with environmental factors; incidence in women is twice as frequent as men, no geographic or racial predilection.

**Ocular:** Conjunctivitis; absence of goblet cells of conjunctiva; conjunctival ulcer; pannus and keratitis; corneal opacity; entropion; trichiasis; cicatization of lacrimal ducts; corneal perforation; symblepharon; dry eyes; bilateral involvement (may be asymmetrical); ocular shrinkage; xerosis; conjunctival and corneal bullae.

**Clinical:** Subepidermal and subepithelial blistering of mucous membranes; blisters may occur in pharyngeal, laryngeal, nasal, anal, and genital mucosa.


**Benjamin-Allen Syndrome**

**General:** Branchial arch syndrome; not hereditary.

**Ocular:** Bilateral dermoids of conjunctiva; marked follicular hyperplasia of conjunctiva.

**Clinical:** Lymphadenopathy; cutaneous nevoid lesions; incomplete alopecia; mental retardation; growth retardation.


**Benson Disease (Asteroid Bodies of the Vitreous; Asteroid Hyalitis; Snowball Opacities of the Vitreous; Scintillatio Albescens)**

**General:** Etiology unknown; occurs in people of advanced age who have been asymptomatic.

**Ocular:** Small, solid, stellate, spherical bodies in an otherwise normal vitreous; creamy, flat white, or shiny when viewed with an ophthalmoscope; may interfere with accurate measurement of axial length.

**Clinical:** Increased prevalence of diabetes mellitus, hypertension, atherosclerosis, and hyperopia.


**Berardinelli-Seip Syndrome (Congenital Generalized Lipodystrophy)**

**General:** Autosomal recessive; disorder of the hypothalamus.

**Ocular:** Punctate corneal infiltrations (lipodystrophia corneae).

**Clinical:** Advanced bone age; dilation of the third ventricle and basal cistern; frequent elevation of growth hormone; severe lipid levels; enlarged liver; diabetes mellitus; hyperpigmentation of axillae and chest wall; phlebomegaly.


**Best Disease (Best Macular Degeneration; Vitelliruptive Macular Dystrophy; Polymorphic Macular Degeneration of Braley; Vitelliform Dystrophy)**

**General:** Up to 7 years of age; a type of heredomacular dystrophy; autosomal dominant with variable expressivity.

**Ocular:** Egg yolk lesion at macula, later absorbed to leave atrophic scar; hemorrhagic or serous exudates beneath pigment epithelium; hyperopia; esotropia; strabismic amblyopia; unusual associations with full-thickness macular hole and extramacular multifocal vitelliform disease have been reported.


**Beta-Glucuronidase Deficiency (Mucopolysaccharidosis VII; MPS VII)**

**General:** Autosomal recessive disorder associated with enzyme deficiency of β-glucuronidase; disorder combines clinical and biochemical features of the Morquio and Sanfilippo syndromes.

**Ocular:** Clouding of the cornea.

**Clinical:** Dwarfism; hepatosplenomegaly; skeletal deformity; mental retardation; hernias; unusual facies; delayed psychomotor development; frequent symptomatic pulmonary infections.


**Bieber Syndrome**

**General:** X-linked recessive inheritance.

**Ocular:** Microphthalmos; corneal pannus; cataracts; uveal hypoplasia; retinal dysplasia; optic nerve hypoplasia; congenital blepharoptosis.

**Clinical:** Microencephaly; mental retardation; agenesis of corpus callosum; hypospadias; cryptorchidism.


**Bielschowsky-Lutz-Cogan Syndrome (Internuclear Ophthalmoplegia)**

**General:** Lesion in the medial longitudinal fasciculus; anterior internuclear ophthalmoplegia consists of paresis of convergence with paresis of homolateral medial rectus muscle during lateral gaze toward opposite side of the lesion; in posterior internuclear ophthalmoplegia, convergence is not affected, while the homolateral medial rectus muscle is paralytic on lateral gaze; the most common causes in young patients include a demyelinating process such as multiple sclerosis, whereas an ischemic process is more common in the elderly; other reported causes of brainstem infarction associated with internuclear ophthalmoplegia include sickle cell trait, priarteritis nodosa, Wernicke encephalopathy, "crack" cocaine smoking.
**Ocular:** Unilateral or bilateral palsy of the medial rectus muscle during conjugate lateral gaze but with or without normal function of this muscle during convergence, depending on the type of internuclear ophthalmoplegia; dissociated nystagmus in the maximal abducted contralateral eye.


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**Biemond Syndrome**

**General:** Simple recessive; hypophyseal infantilism.

**Ocular:** Night blindness in the presence of retinal pigment degeneration; iris coloboma (occasionally); retinal pigmentary degeneration.

**Clinical:** Mental retardation; polydactyly; genital dystrophia (genital organs may have been arrested in their development; absence of secondary sex characteristics); obesity; hypogenitalism; postaxial polydactyly; hydrocephalus; hypoplasias.


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**Bietti Disease (Bietti Marginal Crystalline Dystrophy)**

**General:** Autosomal recessive; metabolic disturbance; histopathologic studies demonstrated advanced panchorioretinal atrophy with crystals and complex lipid inclusions seen in choroidal fibroblasts.

**Ocular:** Marginal corneal crystalline dystrophy with retinitis punctata albescens; panchorioretinal atrophy.

**Clinical:** Asymptomatic.


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**Bing-Neel Syndrome**

**General:** Association of macroglobulinemia and central nervous system symptoms; excessive production of gamma M globulin; over 50 years of age; anoxia secondary to blood sludging from increased viscosity is explanation for peripheral retinal vascular changes.

**Ocular:** Ptosis; paralysis of extraocular muscles; glaucoma; chorioretinitis; dilated and segmented retinal veins; vascular tortuosity; retinal hemorrhages; peripheral microaneurysms; mild papilledema.

**Clinical:** Chronic encephalopathy; peripheral neuropathy; strokes; subarachnoid hemorrhages; weakness; fatigability; weight loss; splenomegaly; anemia.


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**Bipolaris**

**General:** Dematiaceous septate fungus found in dust, soil, and decaying matter; associated with acquired immunodeficiency syndrome (AIDS).

**Ocular:** Gradual progressive visual loss; afferent pupillary defect; proptosis; corneal ulcers; optic disk pallor; orbital cellulitis; ophthalmoplegia; endophthalmitis.

**Clinical:** Pansinusitis; sinusitis; headaches; nosebleeds; mucoid rhinorrhea; allergic rhinitis; nasal stuffiness; allergic fungal sinusitis.


**B-K Mole Syndrome (Familial Atypical Multiple Mole Melanoma Syndrome; DNS; Dysplastic Nevus Syndrome)**

**General:** Autosomal dominant.

**Ocular:** Ocular melanoma; metastasis to the anterior segment of the eye; occurs much less often than metastases to the choroid; iris nevi; choroidal nevi; conjunctival nevi.

**Clinical:** Many large, irregular, variable nevi predominantly occurring on the upper part of the trunk and extremities; atypical melanocytic hyperplasia; lymphocyte infiltration of the dermis and neovascularization; cutaneous melanoma (possible association).


**Blackwater Fever**

**General:** Usually occurs in association with malaria, *Plasmodium falciparum* infection; mortality 20% to 30%; recurrent hemolytic episodes with subsequent malarial infections.

**Ocular:** Scleral icterus; cotton-wool spots; retinal edema; optic disk edema; conjunctival calcium deposits; band keratopathy; cortical blindness; epibulbar hemorrhage of conjunctiva, and episclera; retinal hemorrhages.

**Clinical:** Fever; hemolysis; icterus; hemoglobinuria; malaria; uremia; nausea; vomiting; vertigo; convulsions; coma; acute renal failure; hypertension, azotemia; hypervolemia; metabolic disturbances; hyponatremia; hypercalcemia.


**Blastomycosis**

**General:** Chronic fungal disease caused by *Blastomyces dermatitidis*.

**Ocular:** Hypopyon; mycotic keratitis; corneal ulcer; choroidal granuloma; nodules of iris; cicatrization of eyelid; ectropion; descemetocoele; panophthalmitis; recurrent papillomatous lesion upper lid; granulomatous conjunctivitis.

**Clinical:** Granulomatous lesions of skin, lung, bone, or any part of the body.


Blatt Syndrome (Cranio-Orbito-Ocular Dysraphia)

General: Autosomal dominant; characterized by distichiasis and anisometropia; both sexes affected; present from birth.

Ocular: Hypertelorism; microophthalmos; distichiasis with the meibomian glands usually absent; anisometropia.

Clinical: Meningocele or meningoencephalocele; cranial deformities; malformations of facial bones.


Blau Syndrome

General: Rare, autosomal dominant Iris bombe, uveitis, secondary angle closure glaucoma

Clinical: Skin rash, camptodactyly, early-onset granulomatous arthritis, flexion deformity


Blepharo-Naso-Facial Malformation Syndrome

General: Autosomal dominant.

Ocular: Telecanthus; lateral displacement of lacrimal puncta; lacrimal excretory obstruction.

Clinical: Masklike face; bulky nose; weak facial muscles; torsional dystonia; mental retardation.


Blepharophimosis Syndrome

General: Dominantly inherited tetrad of ptosis, epicanthus inversus, telecanthus, and blepharophimosis.

Ocular: Scarred or contracted in secondary blepharophimosis because of ocular pemphigus or trachoma; ectropion; epicanthus inversus; lacrimal puncta displacement; ptosis; telecanthus; optic nerve coloboma; angle dysgenesis; optic nerve hypoplasia; amblyopia; strabismus.

Clinical: Low-set ears; low nasal bridge.


Blepharoptosis, Myopia, Ectopia Lentis

General: Autosomal dominant.

Ocular: Ptosis; high-grade myopia; ectopia lentis; displacement of crystalline lens of eye; connective tissue defect of sclera, zonules, and levator aponeurosis.

Clinical: None.


Bloch-Sulzberger Syndrome (Incontinentia Pigmenti; Siemens-Bloch-Sulzberger Syndrome)  

**General:** Familial disorder affecting ectoderm; manifestations at birth; female predominance; X-linked dominant phenotype; disturbance of skin pigmentation.

**Ocular:** Orbital mass; retrolental fibroplasia; pseudoglioma; strabismus; blue sclera; cataract; optic nerve atrophy; papillitis; nystagmus; chorioretinitis; anomalies of chamber angle; neovascularization of retina; retinal hemorrhages and edema; microphthalmia; tractional retinal detachment.

**Clinical:** Dental and skeletal anomalies common; neurologic abnormalities; recurrent inflammatory lesions; skin melanin pigmentation on the trunk: (marble cake); occipital lobe infarct; neonatal infarction of the macula.


Blocked Nystagmus Syndrome (Nystagmus Blockage Syndrome; Nystagmus Compensation Syndrome)  

**General:** Von Noorden documented this syndrome with electrooculographic recordings.

**Ocular:** Bilateral or monocular convergence where the adducted eye(s) cannot be abducted to the midline; if monocular, it may alternate; esotropia increases with prolonged fixation; head turn; nystagmus.


Bloom Syndrome (Bloom-Torre-Mackacek Syndrome; Levi-Type Dwarfism; Telangiectasis; Facial Dwarfism)  

**General:** Autosomal recessive inheritance; male preponderance; usually low birth weight following full-term gestation; full-term, abnormally small children.

**Ocular:** Erythema of the lower eyelids.

**Clinical:** Facial rash; erethema of any part of the body; hypersensitivity to light; failure to grow; microcephaly; dolichocephaly; abnormalities of ears, extremities, digits, and nose; facial rash from sensitivity to sunlight; predisposition to neoplasia (especially leukemia) and diabetes mellitus; multiple chromosomal breaks have been observed in deoxyribonucleic acid (DNA) from these patients.


Blue Rubber Bleb Nevus Syndrome (Bean Syndrome)  

**General:** Onset after birth; autosomal dominant.

**Ocular:** Subconjunctival hemangioma with overlying fibrosis; raised hemorrhagic lesion near macula suggestive of a small arteriovenous malformation.

**Clinical:** Vascular lesions; cutaneous lesions found anywhere on the body; profuse sweating may occur over the skin lesions with pain or tenderness on palpation; visceral lesions are common; tender bluish papules on the trunk and extremities; colonic hemangiomas.


Bobble-Head Doll Syndrome

**General:** Caused by massive dilation of the third ventricle; occurs in childhood.

**Ocular:** Pallor of the optic disk or optic atrophy; visual loss.

**Clinical:** Flexion extension movements of the head and neck on the trunk at a rate of two or three per second; pendular movements also may involve the trunk and upper limbs; hydrocephalus; obesity; head bobbing ceases during sleep; mental retardation.


Bogorad Syndrome (Paroxysmal Lacrimation Syndrome; Crocodile Tear Syndrome)

**General:** Subsequent to facial palsy if the lesion is proximal to the geniculate ganglion; during regeneration, fibers supposed to reinnervate the sublingual and submandibular glands are partly interchanged with fibers innervating the lacrimal gland and, hence, gustatory stimulation causes lacrimation.

**Ocular:** Unilateral lacrimation while eating or drinking due to misdirected nerve fiber regeneration.

**Clinical:** Excessive salivation (occasionally); diffuse facial muscle response or facial contracture with lacrimation.


Bonnet-Dechaume-Blanc Syndrome (Cerebroretinal Arteriovenous Aneurysm Syndrome; Neuroretinoangiomatosis Syndrome; Wyburn-Mason Syndrome)

**General:** Dominant inheritance; unilateral or bilateral arteriovenous aneurysm of the midbrain with ipsilateral retinal angioma and skin nevi; severity and extent of symptoms depend on location of cerebral aneurysm and structures it may involve; not regarded as hereditary; incidence is equal in men and women; usually becomes symptomatic at age 30 years.

**Ocular:** Exophthalmos; ptosis; strabismus; nystagmus; hemianopsia due to lesion in optic tract or pulvinar; sluggish pupils; anisocoria; retinal arteriovenous aneurysm; varicosity of retinal veins; arteriovenous angiomas; papilledema; optic atrophy of fellow eye; vitreous hemorrhage; rubeosis iridis; optic neuropathy secondary to compression by vascular malformation; proptosis; partial ophthalmoplegia.

**Clinical:** Arteriovenous angiomas of the thalamus and mesencephalon; facial vascular and pigmented nevi, usually in the trigeminal distribution; psychic disturbances; slow and scanning speech; hydrocephalus; headache; dizziness; hemiplegia; congenital defects of bone, muscle, kidneys, and gastrointestinal tract.


Bornholm Disease (Epidemic Pleurodynia)

**General:** Associated with group B coxsackievirus; epidemic occurrence in summer and early fall; person-to-person contact; incubation 3 to 5 days; affects both sexes; prevalent in children and young adults; recurrent episodes of sudden excruciating pain in abdominal or thoracic regions, increased by movement and respiration.

**Ocular:** Optic neuritis.

**Clinical:** Malaise; sore throat; anorexia; muscle pain; abdominal pain; cutaneous hyperesthesia and paresthesia over affected area; meningitis; myocarditis; hepatitis; orchitis.
Botulism

**General:** Caused by a toxin-producing strain of *Clostridium botulinum*; occurs primarily after the ingestion of contaminated food; the organism can produce a neurotoxin, the effect of which can be life threatening.

**Ocular:** Absent optokinetic nystagmus, absent vertical gaze; marked limitation of horizontal gaze; ptosis; diplopia; decreased tear secretion; mydriasis; paralysis of accommodation; nystagmus; optic atrophy; optic neuritis; extraocular muscle paresis.

**Clinical:** Dizziness; severe respiratory impairment; gastrointestinal disturbances; dysphagia; dysarthria; postural hypotension.


Bourneville Syndrome (Bourneville-Pringle Syndrome; Tuberous Sclerosis; Epiloia)

**General:** Irregular dominant inheritance; more frequent in females; most patients die before age 24 years.

**Ocular:** Vitreous often cloudy; lens opacities; retinal mushroom-like tumor of grayish-white color; yellowish-white plaques with small hemorrhages and cystic changes in retina; papilledema; disk drusen; cerebral astrocytoma; 40% to 50% of patients have normal intelligence.

**Clinical:** Grand mal, petit mal, or jacksonian seizures (manifest first 2 years of life); mental changes from feeblemindedness to imbecility and idiocy; skin changes arranged usually about nose and cheeks (adenoma sebaceum); congenital tumors of kidney (hypernephroma or tubular adenoma) and heart (rhabdomyoma); cerebral astrocytoma.


Boutonneuse Fever (Marseilles Fever)

**General:** Caused by *Rickettsia conorii* and transmitted by ticks.

**Ocular:** Conjunctivitis; central serous retinopathy; retinal detachment; perivasculitis; uveitis; papillitis; keratitis.

**Clinical:** Fever; lymph node enlargement; papular rash.


Bowen Disease (Intraepithelial Epithelioma; Carcinoma In Situ; Dyskeratosis)

**General:** Squamous cell carcinomas *in situ* of the skin or conjunctiva.

**Ocular:** Dysplastic epithelium, intraepithelial epithelioma, or invasive squamous cell carcinoma of conjunctiva or cornea; infiltration of lacrimal system and sclera.


Brachymetapody-Anodontia-Hypotrichosis Albinoidism Syndrome (Anodontia-Hypotrichosis Syndrome) 169

**General:** Autosomal recessive.

**Ocular:** Strabismus; nystagmus; distichiasis; cataracts; high myopia.

**Clinical:** Congenital anodontia; small maxilla; short stature; shortening of metacarpals and metatarsals; little hair growth; albinoidism.


Brain Dysfunction Syndrome (Dyscontrol Syndrome) 170

**General:** Suspected causes include tumors of the limbic system and/or localized atrophy of the brain.

**Ocular:** Visual field defects.

**Clinical:** Manic behavior; physical brutality without motive; sexual assaults; seizures; lack of memory; hallucinations; temporal lobe seizures; depression; speech difficulties.


Branched-Chain Ketoaciduria (Maple Syrup Urine Disease) 171

**General:** Deficiency in the oxidative decarboxylation of the corresponding α-ketoacids; possibly autosomal recessive inheritance; both sexes affected; onset in first week of life.

**Ocular:** Ptosis; epicanthal folds; hypertelorism; prominence of supraorbital ridges; cataract; strabismus; decreased or absent pupillary reaction to light; horizontal nystagmus; optic atrophy; ophthalmoplegia.

**Clinical:** Maple syrup odor of urine; neurologic symptoms; death may follow promptly or the patient may live for a decade during which severe mental retardation is apparent; vomiting; failure to thrive; absence of grasping reflex; generalized rigidity; hypoglycemic crisis; cortical blindness.


Branchial Clefts with Characteristic Facies, Growth Retardation, Imperforate Nasolacrimal Duct, and Premature Aging 172

**General:** Autosomal dominant.

**Ocular:** Strabismus; obstructed nasolacrimal ducts.

**Clinical:** Low birth weight; retarded growth; bilateral bronchial cleft sinuses; broad nasal bridge; protruding upper lip; carp mouth; premature aging; malformed ears; linear skin lesions behind the ears.


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Brill-Symmers Disease (Lymphosarcoma; Giant Follicular Lymphoma)  
**General:** Occurs primarily in older patients.

**Ocular:** Unilateral or bilateral lid swelling; orbital masses with exophthalmos; lacrimal gland infiltration.

**Clinical:** Patients survive 6 to 7 years; special form of lymphoma.


Brittle Cornea Syndrome (Brittle Cornea, Blue Sclera and Red Hair Syndrome; Blue Sclera Syndrome)  
**General:** Autosomal recessive; rare.

**Ocular:** Spontaneous perforation of cornea (brittle cornea); blue sclera; acute hydrops; microcornea; sclerocornea; cornea plana; keratoconus; keratoglobus.

**Clinical:** Red hair; associated with Ehlers-Danlos syndrome, osteogenesis imperfecta, and Marfan syndrome.

Cameron JA. Corneal abnormalities in Ehlers-Danlos syndrome type VI. *Cornea* 1993; 12:54-59.

Brown Syndrome (Superior Oblique Tendon Sheath Syndrome)  
**General:** Etiology unknown; affects both sexes; present from birth; may be congenital or acquired (secondary to trauma, orbital surgery, or injections, or following delivery).

**Ocular:** Bilateral ptosis with associated backward head tilt; widening of palpebral fissure with attempted upward gaze; ocular movements show failure in direction of superior oblique action; may be associated with underaction of the inferior oblique; adduction or abduction restricted or completely abolished; choroidal coloboma.


Brown-Marie Syndrome (Brown-Marie Ataxic Syndrome; Sanger Brown Syndrome; Hereditary Ataxia Syndrome; Marie Hereditary Ataxia)  
**General:** Cause unknown; simple recessive inheritance, although irregular dominant transmission has been observed.

**Ocular:** Nystagmus; strabismus; ophthalmoplegia; anisocoria; Argyll Robertson pupil; retinitis pigmentosa; optic nerve atrophy; retrobulbar optic neuritis.

**Clinical:** Hereditary ataxia; choreiform movements; athetosis; pyramidal tract paresis; speech difficulties; hyperreflexia.


Brown-McLean Syndrome  
**General:** Following cataract surgery Photophobia, posterior staphyloma, peripheral corneal edema with underlying endothelial pigment, iridonesis
Clinical: Foreign body sensation

Brown-Sequard Syndrome

**General:** Caused by lesion (injury, tumor pressure) of spinal cord.

**Ocular:** Nystagmus (if lesion in upper cervical area); sluggish pupillary reaction to light (occasional finding); optic atrophy.

**Clinical:** Homolateral spastic paralysis with (i) loss of ipsilateral deep joint, tendon, and vibratory sensations below level of the lesion; (ii) loss of contralateral pain and temperature sensations; and (iii) sphincteral disturbances. Cases of this syndrome caused by meningomyelitis secondary to syphilis, herpes zoster, and multiple sclerosis have been reported.


Bruch Membrane Drusen

**General:** Autosomal dominant; round or oval lesions in grape like clusters in the posterior polar region; found on the vitreal side of Bruch membrane, secreted by the retinal pigment epithelial cells, apparently secondary to an inborn error of metabolism localized in the retinal pigment epithelium.

**Ocular:** Crystalline retinal degeneration; Doyne honeycomb choroiditis; fleck retina disease; macular edema; macular hemorrhage; pigmentary disturbances with secondary calcifications; central scotomata.

**Clinical:** None.


Bruns Syndrome (postural Change Syndrome)

**General:** Caused by tumors of the third, fourth, or lateral ventricle or by lesions of the midline in the brain.

**Ocular:** Partial ophthalmoplegia (third nerve paralysis) and gaze paralysis; oculomotor paresis associated with postural change of head or body; amaurosis or transient blindness; flashes of light.

**Clinical:** Severe paroxysmal headache; nausea and vomiting; vertigo; irregular respiration; apnea; syncope; tachycardia; free-floating cysts within the fourth ventricle may produce intermittent foramen obstruction and Bruns syndrome; Kramer reported a patient with a free-floating cysticercus cyst with this condition.


Buerger Disease (Thromboangiitis Obliterans)

**General:** Unknown etiology; males who smoke and are under age 35 years; affects small- and medium-sized arteries and veins of the extremities; segmented episodic inflammatory panarteritis with associated thrombosis.

**Ocular:** Exudative retinopathy; occlusion of retinal vessels; retinal hemorrhages; perivasculitis and endovasculitis; blindness; cataract.

**Clinical:** Intermittent claudication; coolness; paresthesia; hyperemia; cyanosis and gangrene may be present in the lower extremities.
Bullous Ichthyosiform Erythroderma (Collodion Baby; Congenital Ichthyosis; Epidermolytic Hyperkeratosis; Ichthyosis; Ichthyosis Vulgaris; Lamellar Ichthyosis; Nonbullous Ichthyosiform Erythroderma; Xeroderma; X-Linked Ichthyosis)

**General:** Autosomal inherited disorder; affects both sexes; normal at birth; onset within first 7 days; X-linked; pathogenesis may be secondary to physicochemical changes of corneal tissues including accumulation of cholesterol sulfate.

**Ocular:** Keratopathy; corneal scarring; keratitis; conjunctivitis; lagophthalmos; photophobia; lid erythema; lacrimation; keratoconus; deep corneal punctate/filiform lesions

**Clinical:** At birth, the skin surface is moist, red, and tender; within several days, thick verrucous scales form.


Burnett Syndrome (Milk Drinker Syndrome; Milk-Alkali Syndrome)

**General:** Characterized by alkalosis, hypercalcemia, and transient renal insufficiency with azotemia; develops during milk-alkali therapy for peptic ulcer; seen in excessive intake of milk or soluble alkali, as in therapy for peptic ulcer.

**Ocular:** Band-shaped keratopathy; conjunctivitis with calcification.

**Clinical:** Nausea, vomiting; headache; irritability; dizziness; depression; confusion.


Schneider AB, Sherwood LM. Calcium homeostasis and the pathogenesis and management of hypercalcemic disorders. *Metabolism* 1974; 23:975

C Syndrome (Opitz Trigonocephaly Syndrome; Trigonocephaly Syndrome)

**General:** Autosomal recessive; consanguinity; early death.

**Ocular:** Hypertelorism; up-slanted palpebral fissures; strabismus.

**Clinical:** Polydactyly; unusual facies; cardiac abnormality; cryptorchidism; Omtra-oral anomalies; abnormally modeled ears; cardiac anomalies; neonatal hypotonia; severe mental retardation; short neck with loose skin.


Caffey Syndrome (Caffey-Silverman Syndrome; Infantile Cortical Hyperostosis) 185

**General:** Cause unknown, possibly collagen disease or viral infection; prevalent in females; onset in early infancy; sudden onset; benign; self-limited.

**Ocular:** Periorbital edema and tenderness; transient proptosis; mild conjunctivitis.

**Clinical:** Tender swelling over regions of cortical hyperostosis (may resemble periostitis); severe anemia; fever; dysphagia; pleurisy.


Caisson Syndrome (Bends Syndrome; Diver’s Palsy; Compressed-Air Illness) 186

**General:** Under high atmospheric pressure (18 lb/in² at least) the blood becomes saturated with nitrogen; sudden decompression (e.g., return to normal atmospheric pressure when divers surface too rapidly) causes the nitrogen to bubble out in gas form, with resulting destruction of tissue spaces; symptoms appear usually within 3 hours after decompression.

**Ocular:** Nystagmus; diplopia; transient blindness; cataract formation with rapidly developing vacuoles and gray opacities; narrowing of the retinal vessels.

**Clinical:** Severe joint pain; dyspnea with sensation of chest constrictions; giddiness; hemiplegia; vertigo; deafness; aphasia; paraplegia; convulsion; pruritus; abdominal pain.


California Syndrome  187

**General:** Functional and/or malingering visual complaints in people with psychosocial problems relating to parental divorce, poor school performance, and attention-getting behavior; seen in adults as well as children; usually secondary gain motive (e.g., disability benefits, Workmen's Compensation, litigation, or income tax).

**Ocular:** Blurred vision; abnormal vision fields; abnormal color vision; voluntary nystagmus; decreased visual acuity at near or far distance.

**Clinical:** Hysteria; malingering; social stresses.


CAMAK Syndrome (Cataract, Microcephaly, Arthrogryposis, Kyphosis Syndrome) 188

**General:** Low birth weight; autosomal recessive.

**Ocular:** Cataracts at birth or within 3 weeks.

**Clinical:** Mental retardation; stiffness of joints, microcephaly; birdlike facies; progressive curvature of the spine.


CAMFAK Syndrome (Cataract, Microcephaly, Failure to Thrive, Kyphoscoliosis Syndrome) 189

**General:** Autosomal recessive; there is evidence supporting that it is a neurologic disease characterized by peripheral and central demyelination similar to that seen in Cockayne syndrome.

**Ocular:** Cataracts.

**Clinical:** Microcephaly; failure to thrive; mental retardation; spasticity; hip dislocation; kyphoscoliosis.


### Canalis Opticus Syndrome 190

**General:** Loss of vision after blunt trauma to the head (mainly forehead) without direct eye injury; thought to occur because of sudden stretching of the fixed, as well as the movable, portions of the optic nerve during movement of the brain at the time of injury.

**Ocular:** Spontaneous unilateral or bilateral, reversible or irreversible amaurosis; absent pupil reaction in cases of complete blindness; spontaneous visual recovery has been reported anecdotally.

**Clinical:** Blunt head injury.


### Candidiasis 191

**General:** Yeastlike opportunistic fungal infection caused by *Candida albicans*.

**Ocular:** Uveitis; hypopyon; conjunctivitis; keratitis; corneal ulcer; blepharitis; endophthalmitis; dacyrocystitis; papillitis; retinal atrophy; Roth spot; vitreous abscess; retrobulbar abscess; retinal detachment; panophthalmitis; chorioretinitis; infectious crystalline keratopathy.

**Clinical:** *C. albicans* normally is present as an intestinal saprophyte in 35% to 75% of the human population; in situations of internal environmental change, however, *Candida* can become pathogenic (e.g., obesity, diabetes mellitus, malignancy, and other debilitating conditions).


### Canine Tooth Syndrome (Class VII Superior Oblique Palsy) 192

**General:** Caused by trauma to the trochlear area, producing a "double Brown syndrome"; secondary to strengthening the superior oblique along with a residual superior oblique palsy, or a combination of local trauma to the trochlea causing restriction to upgaze along with closed head trauma producing a fourth nerve palsy.

**Ocular:** Underaction of the superior oblique and underaction of the inferior oblique on the same side.


### Capgras Syndrome (Illusion of Double Syndrome; L’illusion des Sosies; Phantom Double Syndrome; Nonrecognition-Misidentification Syndrome) 193

**General:** Characterized by misidentification or nonrecognition of a person by the patient who believes that this person appears in double in front of him or her; occurs in paranoid psychosis, and only people familiar or important to the patient appear in double; preponderance in women; agnosia of identification.

**Ocular:** Illusion of double perception with failure of recognition of a known person.
Clinical: General claims include statements that the person seen is an impostor, although the person is well known to the patient; hallucinations; delusions.


Capsular Bag Distension Syndrome

General: Occurs after phacoemulsification as a result of occlusion of the anterior capsularhexis opening by the intraocular lens optic

Ocular: Shallow anterior chamber, pupil peaking, accumulation of turbid fluid in the capsular bag

Capsular Block Syndrome

General: Following cataract surgery

Ocular: Complete sealing by the anterior capsule opening by the optic and displacement of the posterior capsule far behind the posterior optic surface

CAR Syndrome (Cancer-Associated Retinopathy Syndrome)

General: Rare; antiretinal antibodies in blood of cancer patients experiencing concomitant loss of vision; vision loss may be noted before cancer is diagnosed; mechanisms involved in the vision loss experienced by these patients is not understood, but serologic studies indicate they may include a series of autoimmune reactions directed at specific components of the retina.

Ocular: Vision loss usually progressive; retinal degeneration; retinal hole; abnormal visual fields; loss of color vision; retinal detachment; optic atrophy; ringlike scotoma; night blindness; retinal phlebitis.

Clinical: Carcinoma with or without metastasis to any part of the body.

Carcinoid Syndrome

General: Slow-growing neoplasms of enterochromaffin cell; metastatic tumors usually arise from small primary tumors in the ileum.

Ocular: Lacrimation; periorbital edema; choroidal and orbital metastases.

Clinical: Cutaneous flushing; telangiectasia; intestinal hypermotility.

Cardiac Myxomas

General: Myxomas of the heart account for approximately 50% of primary heart tumors; although benign, they can cause serious complications and death by obstruction or embolism; tumor arises from the mural endocardium.
Ocular: Central or branch retinal artery obstruction with ganglion cell edema and a "cherry-red macula"; choroidal and retinal infarct; ischemic optic neuropathy; conjunctival and caruncle pigmentation; eyelid pigmentation; transient loss of vision.

Clinical: Signs and symptoms are a result of emboli that travel to the extremities, brain, liver, spleen, kidney, cerebrum, and, rarely, the coronary arteries.


**Cardiorespiratory Obesity Syndrome (Pickwickian Syndrome)**

General: Chronic pulmonary insufficiency caused by extensive obesity.

Ocular: Venous congestion; spontaneous hyphema; chorioretinal venous congestion with retinal hemorrhages and exudates; papilledema; rubeosis iridis (may be bilateral).

Clinical: Pronounced and greatly excessive obesity; headache; dyspnea; drowsiness; cyanosis; heart failure; muscular twitching; disturbed consciousness.


**Carotid Artery Syndrome (Carotid Vascular Insufficiency Syndrome; Ocular Ischemic Syndrome)**

General: Causes include microemboli, atherosclerotic plaques, arteritis, arterial compression by cicatricial tissue surrounding the vessel, and tumors; male preponderance; onset between ages 50 and 70 years.

Ocular: Lacrimation; homolateral transient, painless visual loss; photopsia; hemianopsia; retinal infarcts; cholesterol plaques may be seen in retinal arteries on funduscopic examination; optic atrophy; hypoxic retinopathy; low-tension glaucoma; anterior uveitis; cataract; visual acuity 20/400 or less; iris neovascularization; angle neovascularization; optic disk pale; retinal hemorrhages; Homer syndrome; amaurosis fugax; retinal artery occlusion; ophthalmoparesis; proptosis; chemosis; conjunctival hyperemia; acute orbital infarction.

Clinical: Transient cerebral ischemia with contralateral weakness of arm and leg; hemisensory disturbances; mental confusion and dysphasia; headache; dizziness; epileptiform seizures; carotid dissection.


**Carotid Artery Syndrome (Cavernous Sinus Fistula Syndrome; Red-Eyed Shunt Syndrome)**

General: Seventy-five percent of cases caused by trauma; others occur spontaneously or are congenital; fistula from carotid artery to cavernous sinus.

Ocular: Progressive, pulsating exophthalmos; distended pulsating superior orbital vein; venous congestion of lids; variable ophthalmoplegia, depending on involvement of cranial nerves III to VI; secondary glaucoma; congestion of conjunctiva with chemosis; corneal ulcerations; eversion of the lower lid; loss of corneal sensation; retinal edema; engorgement of retinal veins; papilledema; optic atrophy; oculuc bruit that may be subjective and/or objective; diplopia; visual decrease; choroidal folds; dilated superior ophthalmic vein.

Clinical: Severe unilateral headache; buzzing noise.


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**Carpenter Syndrome (Acrocephalopolysyndactyly Type II)**

**General:** Hereditary; transmitted as an autosomal recessive trait; severe form of Apert syndrome; normal intelligence has been reported with this syndrome; polysyndactyly is not an absolute requirement for this diagnosis.

**Ocular:** Lateral displacement of inner canthus; epicanthal folds; microcornea; corneal opacities.

**Clinical:** Acrocephalopolysyndactyly; brachydactyly; peculiar facies; obesity; mental retardation; hypogonadism; generalized aminoaciduria; cryptorchidism; hypogenitalism.


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**Cataract, Anterior Polar**

**General:** Autosomal dominant; imperfect separation of lens from surface ectoderm during fifth week of embryologic development; abnormal mass in region of anterior pole and incomplete resorption of blood vessels and mesoderm at anterior pole of embryonic lens; they can be associated with chromosomal abnormalities including 3, 18 chromosomal translocation.

**Ocular:** Small opacities on anterior surface of lens; microphthalmia; cataracts usually do not interfere with vision; corneal astigmatism.

**Clinical:** None.


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**Cataract and Congenital Ichthyosis**

**General:** Autosomal recessive; rare.

**Ocular:** Cortical cataract.

**Clinical:** Ichthyosis.


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**Cataract, Congenital or Juvenile (Cataract, Juvenile, Hutterite Type)**

**General:** Autosomal recessive; seen most frequently in people of Japanese origin; autosomal dominant inheritance also has been reported.
Ocular: Retinitis pigmentosa; Usher syndrome (retinitis pigmentosa and congenital deafness); congenital cataract of the "i" phenotype; microphthalmos; keratoconus.

Clinical: Congenital deafness; galactokinase deficiency; epimerase deficiency.


Cataract, Congenital Total with Posterior Sutural Opacities

206

General: Sex-linked; initial lens changes occur in both men and women with continuation of process in men; women show progression at much later age; it has been suggested that several X-linked cataract syndromes are due to deletions of different sizes in the X chromosome.

Ocular: Y-shaped sutural cataracts; congenital cataracts; nuclear cataract; cortical cataract; posterior subcapsular cataract; asymptomatic posterior y-sutural cataracts; severe visual impairment; bilateral pendular nystagmus; bilateral microcornea; exotropia; keratoconus.

Clinical: Mental retardation.


Cataract, Crystalline Aculeiform or Frosted

207

General: Autosomal dominant.

Ocular: Small crystal-like opacities of lens.

Clinical: None.


Cataract, Crystalline Coralliform

208

General: Autosomal dominant.

Ocular: Cataracts characterized by fine crystals in the axial region of the lens.

Clinical: None.


Cataract, Floriform

209

General: Autosomal dominant; rare.

Ocular: Lens opacity takes the form of annular elements, arranged either independently or grouped together like petals of a flower; lenticous; aniridia.

Clinical: None.

Cataract, Membranous

**General:** Autosomal dominant.

**Ocular:** Total cataract that has undergone regression or resorption.

**Clinical:** None.


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Cataract, Microcornea Syndrome

**General:** Autosomal dominant; prominent in Sicilian families.

**Ocular:** Cataracts; microcornea; myopia.

**Clinical:** None.


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Cataract, Microphthalmia and Nystagmus

**General:** Autosomal recessive.

**Ocular:** Miosis; cataract; nystagmus; microphthalmia.

**Clinical:** None.


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Cataract, Nuclear (Coppock Cataract; Cataract, Discoid)

**General:** Autosomal dominant; epidemiologic evidence suggests that a single major gene can account for the correlation among siblings of nuclear sclerosis.

**Ocular:** Congenital zonular cataract; total nuclear cataract; fetal nucleus with scattered fine diffuse cortical opacities and incomplete cortical riders.

**Clinical:** None.


Cataract, Nuclear Diffuse Nonprogressive

**General:** Autosomal dominant; nonprogressive.

**Ocular:** Opacity of fetal nucleus resembles senile nuclear sclerosis.

**Clinical:** None.


Cataract, Posterior Polar

**General:** Autosomal dominant; onset in childhood; progressive.

**Ocular:** Congenital posterior polar opacity; scattered cortical opacities; choroideremia; myopia.

**Clinical:** None.


Cat's-Eye Syndrome (Schachenmann Syndrome; Schmid-Fraccaro Syndrome; Partial Trisomy G Syndrome)

**General:** Causative factor is one extra chromosome, a G chromosome, which may be from a 13-15 or 21-22 chromosome; although the ocular findings of the syndrome are similar to the D 13-15 trisomy group, the systemic manifestations usually are less severe; this syndrome is associated with a supernumerary bisatellited marker chromosome derived from duplicated regions of 22pter→22q11.2; partial cat's-eye syndrome is characterized by the absence of coloboma.

**Ocular:** Hypotelorism; microphthalmos; antimongoloid slant of palpebral fissures; strabismus; inferior vertical iris coloboma (cat eye); cataract; choroidal coloboma; epicanthal folds.

**Clinical:** Anal atresia; preauricular fistulae (bilateral); umbilical hernia; heart anomalies.


Cebophealia

**General:** Term derived from *Cebus* monkey-like head, defective nose, and eyes close together.

**Ocular:** Hypotelorism; mongoloid obliquity,

**Clinical:** Flat, incomplete nose; full cheeks; medial nostril; no palate or cleft lips.


Central Nervous System Deficiency Syndrome (Garland Syndrome; Spillan-Scott Syndrome)

**General:** Cause unknown; found in prisoners who had long been on a deficient diet; no improvement after normal diet resumed.

**Ocular:** Greatly reduced vision, particularly near vision, increasing over weeks or months but rarely progressing to complete blindness; relative or absolute central or paracentral scotomata; bitemporal pallor of the disks; optic neuropathy.
Clinical: Incomplete bilateral deafness, never proceeding to complete deafness; tinnitus; numbness and tingling in the legs, rarely in the hands; unsteadiness of gait; abnormal tendon reflexes (both hyperactive or absent); peripheral neuropathy.


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**Cerebellar Ataxia, Cataract, Deafness, and Dementia or Psychosis (Heredopathia Ophthalmico-Otencephalica)**

General: Autosomal dominant.

Ocular: Posterior polar cataracts.

Clinical: Tremor, paranoid psychosis; dementia; deafness.

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**Cerebellar Ataxia, Infantile, with Progressive External Ophthalmoplegia**

General: Autosomal recessive; neurologic lesion.

Ocular: Paralysis of all extraocular muscles; ptosis; retinal degeneration; blindness.

Clinical: Spinocerebellar degeneration; ataxia.

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**Cerebellar Degeneration with Slow Movements**

General: Autosomal dominant; described only in Indian families; associated with spinocerebellar degeneration and abnormal eye movements.

Ocular: Paramedian pontine reticular formation (horizontal gaze center); absent rapid movements of both eyes and abnormally slow movements.

Clinical: Brainstem lesion of paramedian pontine reticular formation; progressive mental deterioration.

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**Cerebral Autosomal Dominant Arteriopathy**

General: Autosomal dominant, generalized non-atherosclerotic nonamyloid arteriopathy

Ocular: Scotoma with migraine, cataract, iris atrophy, retinal microinfarction

Clinical: Recurrent stroke, cognitive decline, subcortical vascular dementia


Cerebral Cholesterinosis (Cerebrotendinous Xanthomatosis; CTX) 223

**General:** Autosomal recessive; lipid storage characterized by progressive neurologic dysfunction; large amounts of cholestanol and cholesterol in every tissue in body, particularly in brain and lungs.

**Ocular:** Cataracts; juvenile cataracts.

**Clinical:** Cerebellar ataxia; systemic spinal cord involvement; atherosclerosis; mental retardation; unsteady gait; liver damage; jaundice; chronic diarrhea.


Cerebral Palsy 224

**General:** Group of diverse nonprogressive syndromes resulting from injury to the motor centers of the brain; lesions may occur prenatally, in infancy, or in childhood up to age 5 years or more; constitutes the most common cause of permanent physical handicap in children.

**Ocular:** Strabismus; ptosis; congenital cataract; optic nerve atrophy; papilledema; iris coloboma; nystagmus; uveitis; paresis of extraocular muscles; blepharospasm; leukoma.

**Clinical:** Systemic abnormalities, such as mental retardation, seizures, microcephalus, hydrocephalus, speech delays, and behavioral or emotional disturbances; motor defect; central visual impairment due to cerebral cortex and white matter malformation.


Cerebrofacial-Reno-Arthro-Syndactyla Syndrome 225

**General:** Cause familial?

**Ocular:** Trichiasis; slanted and asymmetrical size of lid fissure; blepharitis; contraction of visual fields; peripheral pigmentary anomalies of the retina.

**Clinical:** Mild oligophrenia; small head for body size; slight facial asymmetry; webbed digits; atrophic unilateral sternocleidomastoid muscle; spontaneous shoulder dislocation; small kidneys with renal dysplasia; chronic interstitial nephritis.


Cerebro-Oculo-Facio-Skeletal Syndrome (COFS Syndrome) 226

**General:** Inherited as autosomal recessive disorder; death within the first 3 years of life; feeding difficulties secondary to incoordination of the swallowing mechanism.

**Ocular:** Microphthalmia; blepharophimosis; cataracts.

**Clinical:** Microcephaly; hypotonia; prominent nasal root; large ear pinnae; flexion contractures at elbows and knees; camptodactyly; osteoporosis; kyphosis; scoliosis; congenital muscular dystrophy.


Cerebroretinal Vasculopathy  227

**General:** Autosomal dominant; frontoparietal lobe pseudotumor and retinal capillary abnormalities; neuropathologic findings are largely confined to the white matter.

**Ocular:** Capillary obliteration; retinal telangiectasia; glaucoma; cystoid degeneration; branch retinal vein occlusion; infectious retinitis; reduction in central visual acuity; progressive retinal ischemia.

**Clinical:** Jacksonian and grand mal seizures; headaches; loss of memory; slow speech; cytomegalic virus septicemia; perivascular inflammation; hepatic fibrosis; intracranial mass lesion; systemic collagen vascular disease; systemic lupus erythematosus; central nervous system disease.


Ceroid Lipofuscinosis  228

**General:** Ceroid lipofuscinoses are disorders characterized by the accumulation of fluorescent lipopigments in a number of body tissues; included in this group are several diseases that were once considered variants of Tay-Sachs disease but are now classified separately; ceroid lipofuscinoses may be divided into infantile, late infantile (Bielschowsky-Jansky), juvenile (Spielmeyer-Vogt), adult (Kufs), and atypical forms (see Dollinger-Bielschowsky Syndrome; Kufs Disease; Infantile Neuronal Ceroid Lipofuscinosis; Batten-Mayou syndrome.)

**Ocular:** Tapetoretinal degeneration; pigmentary macular changes.

**Clinical:** Seizures; ataxia; dementia; cerebellar and extrapyramidal signs; "release" hallucinations.


Cestan-Chenais Syndrome (Cestan [1] Syndrome)  229

**General:** Combination of Babinski-Nageotte and Avellis syndromes; lesion in the lateral portion of medulla oblongata.

**Ocular:** Enophthalmos; ptosis; nystagmus; miosis.

**Clinical:** Pharyngolaryngeal or glossopharyngeal paralysis; cerebellar hemiataxia; disturbance of sensibility; contralateral side of lesion.


Charcot-Marie-Tooth Disease (Progressive Neuritic Muscular Atrophy; Progressive Peroneal Muscular Atrophy)  230

**General:** Dominant inheritance; onset between 5 and 15 years; rare disease; demonstrates autosomal dominant as well as recessive and X-linked recessive inheritance.

**Ocular:** Nystagmus; vision reduced if associated with optic nerve involvement; primary optic atrophy (rare).

**Clinical:** Positive familial history; atrophy of small muscles of hands and feet, slowly progressing to distal and then proximal arm and leg; fibrillar muscle twichings (fasciculations) are common; cramps are common.


**Charcot-Wilbrand Syndrome**

**General:** Lesion of artery of angular gyrus of dominant side; lesion can be bilateral; alexia.

**Ocular:** Visual agnosia; loss of ability to revvisualize images; prosopagnosia.

**Clinical:** Occlusion of a portion of the posterior cerebral artery.


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**Charge Association (Multiple Congenital Anomalies Syndrome; Coloboma, Heart Disease, Atresia, Retarded Growth, Genital Hypoplasia, Ear Malformation Association)**

**General:** Syndrome consisting of four of six major manifestations of ocular coloboma, heart disease, atresia, retarded growth and development, genital hypoplasia, and ear malformations with or without hearing loss.

**Ocular:** Blepharoptosis; iris coloboma; optic nerve coloboma; macular hypoplasia; lacrimal canalicular atresia; nasolacrimal duct obstruction.

**Clinical:** Microcephaly; brachycephaly; malformed ear; bilateral finger contractures; heart disease; genital hypoplasia; heart disease; choanal atresia; retarded growth; hearing loss; facial nerve palsies; mental retardation.


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**Charlin Syndrome (Nasal Nerve Syndrome; Nasociliaris Nerve Syndrome; Nasociliary Syndrome)**

**General:** Neuritis of the nasal branch of the trigeminal nerve; three typical spots of pain according to the nerve distribution: (i) above and outside the nose; (ii) above the inner canthus; and (iii) inferior angle of the medial tarsal ligament (see Sluder Syndrome).

**Ocular:** Severe ocular and orbital pain, mainly upper nasal-orbital angle; slight inflammatory swelling of upper lid (occasional); photophobia; ciliary and conjunctival injection; pseudopurulent conjunctivitis; anterior uveitis; iritis; hypopyon; keratitis; corneal ulcers.

**Clinical:** Rhinorrhea; rhinitis always on same side of the ocular involvement; severe pain of ala nasi.


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**Chediak-Higashi Syndrome (Anomalous Leukocytic Inclusions with Constitutional Stigmata)**

**General:** Occurs in albinoid siblings born of consanguineous parents; tyrosinase-positive type of oculocutaneous albinism associated with a fetal reticuloendothelial incompetence.

**Ocular:** Decreased iris pigmentation; photophobia; narrowness and decreased number of vessels in retina; decreased pigmentation of choroid elevated disk; papilledema; infiltration of immature leukocytes in the uvea, retina, and optic nerve; nystagmus; ocular motor palsies; optic disk edema; oculocutaneous albinism.

**Clinical:** Anemia; neutropenia; thrombocytopenia; recurrent infections; hepatosplenomegaly; lymphadenopathy; oculocutaneous albinism; hyperpigmentation of sun-exposed areas.


**Cherry-Red Spot Myoclonus Syndrome (Type I Sialidosis)**

**General:** Morphologic changes in storage process in lysosomes of retinal ganglion cells, neurons of mesenteric plexus, hepatocytes, and Kupffer cells.

**Ocular:** Cherry-red spot of macula; horizontal nystagmus; white dot and flake lens changes; decreased visual acuity and gray perimacular halo.

**Clinical:** Development of a slowly progressive neurologic illness whose most crippling symptoms are myoclonus, seizures, decreased muscle tone, speech disorder, and hyperactive reflexes.


**Chickenpox (Varicella)**

**General:** Acute exanthematous disease; highly contagious; children between ages of 2 and 8 years.

**Ocular:** Conjunctival ulcer; corneal ulcer; descemetocoele; corneal opacity; keratitis; paresis of third, fourth, and sixth nerves; optic neuritis; papilledema; retinitis; hemorrhagic retinopathy; uveitis; cataract; paralytic mydriasis; phthisis bulbi; unifocal chorioiditis; dendritic keratitis; acute retinal necrosis (in a patient with acquired immunodeficiency syndrome [AIDS]); disciform keratitis.

**Clinical:** Fever; malaise; rash; pruritus.


**Chlamydia (Inclusion Conjunctivitis; Paratrachoma)**

**General:** Organism that infects the epithelium of mucoid surfaces; sexually transmitted; major cause of nongonococcal urethritis in men and cervicitis in women; major cause of neonatal ophthalmia; *Chlamydia trachomatis* is an intracellular bacterium lacking respiratory enzymes that has an affinity for mucosal epithelium; serotypes A through C have been epidemiologically associated with trachoma; serotypes E through K have been associated with genital infection and keratoconjunctivitis in sexually active adults and neonates; other serotypes have been associated with lymphogranuloma venereum and Reiter syndrome.

**Ocular:** Follicular conjunctivitis; corneal opacities; keratitis; corneal ulcer; lid edema; uveitis.

**Clinical:** Pneumonia; gastrointestinal disturbances; genital discharge.


Cholera

**General:** Acute illness that results from colonization of small bowel by *Vibrio cholerae*; rare cases of *V. cholerae* meningitis have been reported, some associated with bacteremia.

**Ocular:** Hyperemia of lids and conjunctivae; subconjunctival hemorrhages; madarosis of lids; xerosis of conjunctiva; lagophthalmos; keratomalacia; retinal ischemia; cataract.

**Clinical:** Diarrhea; shock; vomiting; muscle cramps; cyanosis; scaphoid abdomen; thready pulse; tachycardia; hypotension; tachypnea; acute tubular necrosis; metabolic acidosis; death.


Cholestasis with Gallstone, Ataxia, and Visual Disturbances

**General:** Autosomal recessive; not clear if distinct from Byler disease or another form of intrahepatic cholestasis; retinal neurologic features may be secondary to nutritional abnormalities.

**Ocular:** Retinal lesions; optic atrophy; ptosis.

**Clinical:** Congenital cholestasis; gallstone; cerebellar ataxia; jaundice; hepatitis; pruritus.


Chorea (Acute Chorea; Sydenham Chorea; St. Vitus Dance; Huntington Hereditary Chorea)

**General:** Mendelian dominant trait.

**Ocular:** Lid retraction; spasmodic closures; apraxia of lid opening; disoriented ocular movements; anisocoria; mydriasis; hippus.

**Clinical:** Involuntary purposeless movements; emotional ability; muscle weakness.


Choroideremia (Tapetochoroidal Dystrophy, Progressive; Choroidal Sclerosis)

**General:** Sex-linked; onset at early age; progressive; primary degeneration may be of the retina, retinal pigment epithelium, or choriocapillaris; pigment stippling or granularity also evident in female carriers who possess normal and abnormal cells, through Barr body inactivation of one X chromosome.

**Ocular:** Reduction of central vision; constriction of visual fields; night blindness; choroidal and retinal atrophy.

**Clinical:** None.


Choroidoretinal Degeneration with Retinal Reflex in Heterozygous Women 242

**General:** Sex-linked; choroidoretinal degeneration differentiated by presence in heterozygous women of a tapetal-like retinal reflex; there is probably more than one X-linked locus leading to a retinitis pigmentosa type of picture.

**Ocular:** Retinitis pigmentosa; golden-hued, patchy appearance around macula.

**Clinical:** None.


Choroidoretinal Dystrophy 243

**General:** Sex-linked; similar to retinitis pigmentosa with absence of annular scotoma and little vascular change.

**Ocular:** Early poor central vision; retinitis pigmentosa; night blindness.

**Clinical:** None.


Chromosome 11 Long-Arm Deletion Syndrome 244

**General:** Patients with deletion of the long arm of chromosome 11 exhibit a distinctive countenance; female preponderance.

**Ocular:** Colobomas of the choroid, retina, and iris; retinal reduplication; retinal dysplasia; epicanthus; blepharoptosis; abnormal slanting of the interpalpebral fissures; bilateral uveal colobomas; hypertelorism; avascular retina (bilateral); abnormal pattern of retinal vessels.

**Clinical:** Keeled forehead; small carp-shaped mouth; low-set ears; highly arched palate; long upper lip with absent philtrum; short neck; widely spaced nipples; flexion contractures of the knees and elbows; hypoplastic nails; broad thumbs with low insertion; deeply pigmented skin on buttocks, lower back, and abdomen and in inguinal regions, axillas, and clavicular areas; congenital heart disease.


Chromosome 13q Partial Deletion (Long-Arm Syndrome; 13q Syndrome) 245

**General:** No hereditary factor.

**Ocular:** Microphthalmos; antimongoloid slant of lid fissures; bilateral epicanthus; esotropia; cataract; choroidal coloboma; ptosis; retinoblastoma.

**Clinical:** Genital malformations; meningoceles; short neck; small mouth; mental and physical retardation; small head; short stature; broad nasal bridge; simian crease; microcephaly; high nasal bridge; thumb hypoplasia.


Chromosome 18 Partial Deletion (Long-Arm) Syndrome (Monosomy 18 Partial [Long-Arm] Syndrome; De Grouchy Syndrome)  246

**General:** Deletion of approximately one half of the long arm of chromosome 18.

**Ocular:** Hypertelorism; epicanthal folds; narrow palpebral fissure; nystagmus (horizontal); strabismus; myopia; astigmatism; glaucoma; oval pupils; microcornea; posterior staphyloma; oblique disk; optic nerve staphyloma; optic nerve atrophy; microphthalmia; corneal opacities; iris hypoplasia; corectopia.

**Clinical:** Dwarfism; mental retardation; microcephaly; midface dysplasia; prominent antihelix and antitragus; congenital cardiac disease; abnormal, spindle-shaped fingers; genital defects.


Chromosome 18 Partial Deletion (Short-Arm) Syndrome (Monosomy 18 Partial [Short-Arm] Syndrome)  247

**General:** Deletion of the short arm of chromosome 18 (note similarity of clinical features to those of the cri-du-chat syndrome or B1 deletion syndrome) (see Cri-du-Chat Syndrome).

**Ocular:** Hypertelorism; epicanthal folds; ptosis; mongolian or antimongolian slant; strabismus; eccentric pupil; cataract; corneal opacities; concentric visual field defects.

**Clinical:** Short stature; mental retardation; low-set ears; dysphagia; moon face; oliguria; arhinencephaly; microcephaly; congenital alopecia; flat bridge of nose; pyramidal tract signs; weakness and focal dystonia of the lower extremities.


Chronic Epstein-Barr Virus (Epstein-Barr Virus, Chronic; Chronic Infectious Mononucleosis)  248

**General:** Onset late adolescence or early adulthood; rare.

**Ocular:** Bilateral uveitis; cystoid macular edema; papilledema; cataract; keratitis; peripapillary and macular preretinal membranes; vitreitis; lacrimal gland swelling; conjunctivitis; corneal edema; keratoconjunctivitis; follicular conjunctivitis; subepithelial corneal opacities; retinitis; opthalmoplegia; optic neuritis; endophthalmitis; dacryocystitis; nasolacrimal duct obstruction.

**Clinical:** Recurrent fever; pharyngitis; lymphadenopathy; fatigue; malaise; weight loss; splenomegaly.


Chronic Granulomatous Disease of Childhood  249

**General:** Genetically determined metabolic defect manifested by inability of the leukocytes to operate the hexose monophosphate shunt during phagocytosis.

**Ocular:** Conjunctivitis; keratitis; destructive chorioretinal lesions.

**Clinical:** Eczematous dermatitis; microabscesses of the skin, lymph nodes, and viscera.


Chronic Progressive External Ophthalmoplegia (CPEO; Ophthalmoplegia Plus) 250

General: A general term covering many conditions; onset at any age; familial history; conditions associated with CPEO include myotonic dystrophy, Kearns-Sayre syndrome, Stephens syndrome, and oculopharyngeal dystrophy; disorders that rarely cause external ophthalmoplegia include congenital disorders (abetalipoproteinemia, Refsum disease, extraocular fibrosis syndrome, Möbius syndrome), progressive supranuclear palsy, endocrine exophthalmos, myasthenia gravis, and multiple sclerosis; now considered to be a mitochondrial cytopathy with varied clinical presentation; four distinct disorders of ophthalmic importance are (i) CPEO or Kearns-Sayre syndrome, (ii) myoclonus epilepsy with ragged red fibers (MERRF), (iii) mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS), and (iv) Leber optic neuropathy.

Ocular: Exposure keratopathy; filamentary keratitis; keratoconjunctivitis sicca; corneal scarring; esotropia; exotropia; gaze paralysis; ptosis; levator paralysis; cataract; optic atrophy; diplopia; tapetoretinal degeneration; constriction of visual field; retinitis pigmentosa.

Clinical: Weakness; weight loss; myopathic or Hutchinson facies; cardiac abnormalities; central nervous system abnormalities.


Churg-Strauss Syndrome (Allergic Granulomatosis and Angiitis) 251

General: Severe multisystem vasculitis.

Ocular: Allergic granulomas in the extravascular tissues characterized by a central eosinophilic core consisting of necrotic eosinophilic leukocytes and fibrinoid swelling of collagen fibers; uveoscleritis; papilledema; anterior ischemic optic neuropathy; sensory motor neuropathy; reversible monocular blindness; myositis; episcleritis; amaurosis fugax, central branch retinal artery occlusion; retinal vasculitis; retinal hemorrhage; cranial nerve palsies; orbital inflammatory syndrome; corneal ulcer; conjunctival nodules.

Clinical: Bronchial asthma; fever; eosinophilia; necrotizing of small arteries and veins with an infiltration of vessels and perivascular tissues by eosinophils; systemic vasculitis.


Citrullinemia 252

General: Autosomal recessive; enzyme deficiency called argininosuccinic acid synthetase; mutations causing human citrullinemia are extremely heterogeneous; nonconsanguineous persons studied until 1991 had been found to be compound heterozygotes.

Ocular: Hypotonicity; low intraocular pressure; irregular choroidal and retinal pigment epithelium folding; engorged retinal vessels; pallor of optic disk.

Clinical: Mental retardation; nausea; vomiting; tremors; intermittent hyperammonemia.


### Claude Syndrome (Inferior Nucleus Ruber Syndrome; Rubro-Spinal-Cerebellar-Peduncle Syndrome) 253

**General:** Paramedian mesencephalic lesion starting in midbrain; often occlusion of terminal branches of the paramedian arteries supplying the inferior portion of the nucleus ruber.

**Ocular:** Paralysis of ipsilateral oculomotor and trochlear nerves (III, IV).

**Clinical:** May be associated with motor hemiplegia.


### Clivus Edge Syndrome 254

**General:** Elevated intracranial pressure; root of oculomotor nerve pressed against bone as it enters cavernous sinus; includes subdural hematoma, temporal lobe tumor, and supraclinoid aneurysm.

**Ocular:** Brief miosis, then mydriasis and sluggish reaction of pupil; extraocular muscle paresis.


### Clostridium Perfringens 255

**General:** Gram-positive rod; most important cause of gas gangrene infection.

**Ocular:** Hypopyon; gas bubbles in anterior chamber; endophthalmitis; proptosis; glaucoma; coffee-colored discharge; eyelid edema; severe ocular pain; endophthalmitis after penetrating trauma or metastatic.

**Clinical:** Traumatized ischemic skeletal muscle, abdominal wall, or uterus; hemolytic anemia; shock; death.


### Coarctation of the Aorta 256

**General:** Congenital, local, and developmental; occurs in one of 2,000; not familial; not predictable by sex.

**Ocular:** Straightening of the arteriolar tree; arteriovenous crossing defects; focal caliber changes; cotton-wool spots; retinal edema; pronounced corkscrew tortuosity of the arterioles with little other retinal changes in 50% of patients; retinal vascular anomalies secondary to hemodynamic changes.

**Clinical:** Aorta narrowed to less than 1 cm in the region of the insertion of the ductus arteriosus; dyspnea; headache; epistaxis; palpitations of the heart; thoracic pain; intermittent claudication.


### Coats Disease (Leber Miliary Aneurysm; Retinal Telangiectasia) 257

**General:** Exudative retinitis; rare; more common in males than females; 95% unilateral.

**Ocular:** Leukocoria; telangiectatic retinal vessels; solid gray-yellow retinal detachment; optic atrophy; vitreous hemorrhage; anterior uveitis; glaucoma; intraocular calcification (rare); fibroosseous retinal nodules (atypical); hemorrhagic retinal macrocysts; cystoid macular edema.


Cocaine Intoxication Syndrome

General: Neonates born to cocaine-abusing mothers.

Ocular: Dilation and tortuosity of the iris vasculature; use of intranasal cocaine has been associated with optic neuropathy.

Clinical: Signs of withdrawal; jittery; irritable.


Coccidioidomyelcosis

General: Caused by fungus Coccidioides immitis.

Ocular: Conjunctivitis; choroiditis; uveitis; retinal hemorrhages; vitreal opacity; vitreal floaters; episcleritis; hypopyon; granulomatous lesion of optic nerve head; paralysis of sixth cranial nerve; secondary glaucoma; papilledema; mutton fat keratic precipitates; necrotizing granulomatous conjunctivitis; iridocyclitis.

Clinical: Mild respiratory illness; cavity lung lesion.


Cockayne Syndrome (Dwarfism with Retinal Atrophy and Deafness; Mickey Mouse Syndrome)

General: Autosomal recessive; onset in second year of life; wide spectrum of symptoms and severity of the disease suggest that biochemical and genetic heterogeneity exist.

Ocular: Enophthalmos; cataracts; pigmentary degeneration of retina; optic atrophy; band keratopathy; exotropia; nystagmus; absence of foveal reflex; corneal dystrophy; corneal perforation; anhidrosis; exposure keratitis; decreased blinking.

Clinical: Dwarfism (nanism) with disproportionately long limbs, large hands, and large feet; kyphosis; deformed limbs; thickened skull; intracranial calcifications; mental retardation; prognathism; deafness (often partial); precociously senile appearance; sensitivity to sunlight, with skin pigmentation and scarring; dental caries.


Coenurosis

General: Rare human infestation of the cystic larval stage of the dog tapeworm; usually infestation in the muscle, subcutaneous tissue, eye, nervous system, or brain; three species may be involved: Multiceps taenia, Multiceps serialis, and Multiceps glomeratus.

Ocular: Hypopyon; retinal detachment; retinal edema; anterior uveitis; conjunctivitis; proptosis; miosis; vitreal haze; increased intraocular pressure; coenurus cysts of the conjunctiva and iris.

Clinical: Ataxia; headache; loss of weight; somnolence; stiffness of neck and shoulders.
Cogan (1) Syndrome (Nonsyphilitic Interstitial Keratitis)  

**General:** Cause unknown; perhaps a generalized hypersensitivity reaction; most frequently affects young adults; unclear etiology; several studies suggest an autoimmunemediated process, possibly a vasculitis.

**Ocular:** Blepharospasm; lacrimation; congested conjunctival vessels; little or no reaction in anterior chamber but ciliary injection present; interstitial keratitis (unilateral or bilateral); granular-type infiltrates; patchy distribution in deeper stroma; later vascularization; conjunctivitis; corneal opacity; uveitis; nystagmus.

**Clinical:** Vestibuloauditory symptoms (similar to Ménière syndrome); nausea; vomiting; vertigo; tinnitus (abrupt onset); rapidly progressive deafness; loss of equilibration (see Ménière Syndrome); aortic insufficiency; sensorineural testing; lacunar infarcts.


Cogan DG. Syndrome of non syphilitic interstitial keratitis and vestibuloauditory symptoms. *Arch Ophthalmol* 1945; 33:144.


Cogan (2) Syndrome (Oculomotor Apraxia Syndrome; Wieacker Syndrome)  

**General:** X-linked; oculomotor apraxia and muscle atrophy; prevalent in males; corpus callosum can be hypoplastic.

**Ocular:** Rapid and frequent blinking; conjugate palsy; congenital oculomotor apraxia with patient unable to move eyes voluntarily to one side but with otherwise normal ocular movements; patient fixes objects by head tilt and turning, which causes further ocular deviation via the vestibular reflex; compensation for this overshoot is accomplished by some jerky eye movements with final fixation possible and gradual return of the head to the primary position; may be associated with abnormal electroretinographic responses.

**Clinical:** Slow progression, predominantly distal muscle atrophy; congenital contracture of feet; dyspraxia of face and tongue muscles; mild mental retardation.


Cogan-Guerry Syndrome (Microcystic Corneal Dystrophy; Map-Dot Fingerprint Dystrophy)  

**General:** Etiology obscure; condition benign and asymptomatic; females predominantly affected; ultrastructural studies show discontinuous multilaminar thickened basement membrane under abnormal epithelium; the primary defect appears to be synthesis of abnormal basement membrane and adhesion complexes by the dystrophic epithelium.

**Ocular:** Reduced vision mainly with involvement of center of cornea; very fine wavy lines resembling fingerprints within or very close to corneal epithelium and best seen on biomicroscopy with retroillumination; fine grayish spheres (0.1- to O.5-mm diameter) in superficial corneal epithelium; maplike irregular border-lined slightly grayish area.

**Clinical:** None.


Coloboma of Macula (Agenesis of Macula)  265

**General:** Autosomal dominant; can be caused by intrauterine inflammation, birth hemorrhage; infantile inflammation.

**Ocular:** Defect in central area of fundus; coloboma can be pigmented, nonpigmented, or have abnormal vessels associated or completely absent; visual defect; absolute central scotoma; nystagmus; myopia; destruction of pigment epithelium; microphthalmos; coloboma of optic nerve (rare); keratoconus; paravenous retinochoroidal atrophy.

**Clinical:** Microencephaly.


Coloboma of Macula with Type B Brachydactyly (Apical Dystrophy)  266

**General:** Autosomal dominant; bilateral pigmented macular coloboma and brachydactyly.

**Ocular:** Myopia; retinal detachment; coloboma of retina, choroid, sclera, and macula.

**Clinical:** Cleft palate; flexion deformity of distal interphalangeal joints of little fingers of hand; retarded growth; delayed sexual maturity; recurrent dislocation of left patella; short feet; coxa valga; genu valgum.


Coloboma, Ocular  267

**General:** Autosomal recessive; congenital or secondary to faulty closure of embryonic fissure.

**Ocular:** Optic nerve coloboma; retinochoroidal coloboma; orbital cysts; retinal dysplasia; retinal detachment; iris coloboma.

**Clinical:** White sponge nevus.


Colobomatous, Microphthalmia and Microcornea Syndrome  268

**General:** Autosomal dominant pattern of inheritance with complete penetrance

**Ocular:** Bilateral infernasal coloboma; axial enlargement; myopia; iridocorneal angle abnormalities; elevated IOP


Color Blindness, Blue-Mono-Cone Monochromatic Type  269

**General:** Sex-linked; progressive.

**Ocular:** Total to partial color blindness; macular scar; poor central vision; poor color discrimination; infantile nystagmus; nearly normal retinal appearance in most cases; there is evidence showing alterations in the red and green visual pigment gene cluster.

**Clinical:** None.
Color Blindness, Partial, Deutan Series (Deuteranopia) 270

**General:** Sex-linked; affects males; deuteranopes can have one red pigment gene and one anomalous hybrid red-green gene with a spectral absorbance close to the red pigment gene.

**Ocular:** Red-green color blindness.

**Clinical:** None.


Comedo Cataract 271

**General:** Etiology unknown; possibly in the group of neurectodermal dysplasia syndromes (phakomatoses).

**Ocular:** Bilateral cataract or both unilateral comedo nevus and cataract; therapy is cataract operation; good prognosis.

**Clinical:** Bilateral comedo nevus (dermatosis characterized by groups of dilated, keratin-filled follicular spaces).


Computer User Syndrome 272

**General:** Seen in people who use computers extensively.

**Ocular:** Ocular pain; asthenopia; exyclotorsion; depression of gaze; ocular synkinesis.

**Clinical:** Hand-wrist pronation; ulnar abduction; headaches; fatigue; various types of head and shoulder distress; carpal tunnel syndrome.


Cone Dysfunction Syndrome (Achromatopsia) 273

**General:** Male-linked recessive inheritance; condition is stagnant and nonprogressive; all modes of inheritance have been reported as well as many sporadic cases.

**Ocular:** Nystagmus; vision decreased 20/50 to 20/200 or less with no or reduced color vision; color vision might be affected with or without amblyopia; peripheral field loss if rods and cones are involved; photophobia; general fundus lesions, mainly macular involvement with depigmentation and degenerative changes; decreased central vision; difficulty adjusting from light to dark environment.

**Clinical:** Head movements.


Conic-Rod Dystrophy (CRD)  
**General:** Autosomal dominant; retinal dystrophy of photoreceptors, characterized by abiotrophic degeneration of rods and cones; onset before age 10 years; it has been suggested that a locus for cone-rod dystrophy may be located in the segment 18q21.1-q21.3 and 19q.

**Ocular:** Decreased central vision with progressive constriction of peripheral visual fields; degeneration of rods and cones.

**Clinical:** None.


Congenital Cataract and Hypertrophic Cardiomyopathy Syndrome  
**General:** Autosomal recessive; characterized by congenital cataract, hypertrophic cardiomyopathy, mitochondrial myopathy of voluntary muscles, and exercise-related lactic acidosis.

**Ocular:** Cataract; hyperplastic primary vitreous; aniridia; iris colobomas; microphthalmos; nystagmus; strabismus; myopia; keratoconus.

**Clinical:** Pulmonary stenosis; ventricular septal defects; structurally abnormal mito-chondria.


Congenital Cataracts Facial Dysmorphism Neuropathy Syndrome  
**General:** Autosomal recessive; motor and sensory neuropathy.

**Ocular:** Congenital cataracts, microcorneas, strabismus, pendular nystagmus, bilateral blepharoptosis.

**Clinical:** Patients are recognized in infancy by the presence of congenital cataracts and microcorneas; initially, a predominantly motor neuropathy begins in the lower limbs followed by upper limb involvement; severe disability occurs by the third decade; short stature, moderate nonprogressive cognitive deficits, pyramidal signs, and mild chorea are characteristic.


**Congenital Cataract, Microcornea, Abnormal Irides, Nystagmus, and Congenital Glaucoma Syndrome**  
**General:** Autosomal dominant.

**Ocular:** Microphakia; cataract with two concentric disks, with the anterior being swollen; microphthalmos; microcornea; nystagmus; congenital glaucoma; honey-colored iris with absence of pattern; peripheral anterior synechiae; pupillary abnormality; corneal edema; posterior synechiae; vitreous hemorrhage; shallow anterior chamber; vitreous loss; corneal staphyloma; keratoconus.

**Clinical:** High arched palate; increased webbing of fingers and toes; deafness.


**Congenital Cataract with Oxycephaly (Tower Skull Syndrome)**

**General:** Autosomal dominant; craniostenosis.

**Ocular:** Congenital cataracts; keratoconus.

**Clinical:** Large fontanelles; deformed skull; dwarfish; osteopetrosis.


**Congenital Dyslexia Syndrome (Developmental Dyslexia of Critchley; Congenital Word Blindness of Hermann; Primary Dyslexia; Dyslexia Syndrome; Minimal Brain Dysfunction Syndrome; Attention Deficit Disorder; Congenital Word Blindness)**

**General:** Primary reading disability in children with an average or above-average intelligence; male preponderance; dysfunction of the dominating parietotemporal lobe; Levinson postulates a primary cerebellar-vestibular (inner ear) dysfunction underlying this syndrome resulting in a secondary scrambled sensory input and motor output.

**Ocular:** No obvious connection seems to exist between coordination of ocular functions and dyslexia, although associated ocular findings may exist in reading problems (e.g., abnormal optokinetic nystagmus, metamorphopsia, defective color vision, convergence insufficiency, muscle imbalance, refractive errors); low accommodative converge/accommodation associated with decreased visual acuity and contrast sensitivity.

**Clinical:** General clumsiness; disorientation (time-space, right-left); behavioral changes; lack of integration of visual and auditory stimuli.


**Congenital Epiblepharon Inferior Oblique Insufficiency Syndrome**

**General:** Prognosis is good with treatment; present in infancy; inversion of lash line occurs with epiblepharon and is exaggerated by the inferior oblique insufficiency.

**Ocular:** Narrow interpupillary distance; some ocular prominence; epicanthus; epiblepharon exaggerated in downward gaze; spastic entropion with retroflexion of the eyelashes; epiblepharon becomes less pronounced with growth and development; usually bilateral but in some cases asymmetrical; inferior oblique insufficiency usually unilateral; persistent unilateral keratoconjunctival irritation by the inverted cilia; lacrimation due to conjunctival and corneal irritation.

**Clinical:** Chubby cheeks (occasionally).


**Congenital Heart Disease**

**General:** Represents a wide variety of cardiac diseases or defects.

**Ocular:** Dilated, tortuous conjunctival vessels; tortuous retinal vessels; retinal edema; papilledema; retinal arterial macroaneurysm; Duane retraction syndrome; double elevator palsy.

**Clinical:** Hypoxia; increased cerebrospinal fluid; features vary because of a wide variety of cardiac diseases and defects.


### Congenital Hereditary Retinoschisis (CHR; Juvenile X-Linked Retinoschisis)

**General:** X-linked recessive; bilateral; develops early in life but often stabilized toward the end of the second decade; severity varies widely.

**Ocular:** Bilateral vitreoretinal dystrophy; retinoschisis; vitreous veil; vitreous detachment; vitreous hemorrhage; decreased visual fields; maculopathy; cataract; neovascular glaucoma; vitreoretinopathy; proliferative retinal detachment; Mizuo phenomenon.


### Congenital Lues (Congenital Syphilis)

**General:** Caused by intrauterine transplacental infection of fetus by *Treponema pallidum* (see Syphilis).

**Ocular:** Conjunctivitis; keratitis; dacryocystitis; optic nerve atrophy; periostitis; anisocoria; Argyll Robertson pupil; retinal degeneration; nystagmus; gumma of conjunctiva, eyelids, and orbit; paresis of extraocular muscles; secondary glaucoma; uveitis; iridoschisis.

**Clinical:** Cutaneous and mucous membrane lesions; periostitis; anemia; hepatosplenomegaly; ectodermal defects; central nervous system involvement; gummatous lesions.


### Congenital Retinal Nonattachment

**General:** Autosomal dominant or recessive; acquired prenatally or perinatally; retinal dysplasia typical; x-ray irradiation has been reported to cause retinal nonattachment.

**Ocular:** Vascularized mass behind the lens; malformation of the chamber angle and elongation of the ciliary processes; retinal dysplasia.

**Clinical:** Severe fragility of the bones.


### Congenital Retinal Nonattachment with Mental Retardation, Osteoporosis, and Hypotonia

**General:** Autosomal recessive; well-demarcated entity; affects both males and females.

**Ocular:** Retinoblastoma; retina nonattached.

**Clinical:** Mentally retarded; osteoporosis; hypotonia; ligamentous laxity; dwarfism; microcephaly.


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Congenital Spherocytic Anemia (Congenital Hemolytic Jaundice; Hereditary Spherocytosis)  

**General:** Hereditary deficiency of erythrocyte glucose-6-phosphate after exposure to certain drugs, chemicals, and foods such as fava beans.

**Ocular:** Congenital cataract; ring-shaped pigmented deposits of cornea; tortuosity of retinal vessels; mongoloid palpebral aperture; microphthalmos.

**Clinical:** Leukemia; anemia.


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Congenital Tilted Disk Syndrome  

**General:** Defective closure of embryonic cleft; disk appears tilted downward and nasal; affects both sexes; central vision is not affected; field defect improves with myopic correction.

**Ocular:** Myopia; astigmatism; coloboma; situs inversus; posterior inferonasal staphyloma; superotemporal, bitemporal, or superior altitudinal visual field defects; inferior fundus pallor; central retinal vein occlusion; hypoplastic retina and choroid; macular choroidal neovascularization.

**Clinical:** None.


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Congenital Varicella Syndrome  

**General:** Varicella passed in utero from mother to fetus

**Ocular:** Microphthalmia; microcornea; persistent hyperplastic primary vitreous

**Clinical:** Urinary tract infection; neurogenic bladder


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Congenital Vertical Retraction Syndrome  

Aberrant regeneration of the oculomotor nerve; concurrent protective eyelid closure; congenital alterations in the extraocular muscle, its insertion, and its peripheral innervation; nystagmus retractorius; surgical or traumatic rearrangement of orbital structures may account for retraction.


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Conjunctivitis, Ligneous  

**General:** Autosomal recessive; palpebral conjunctiva becomes the site of dense woody membrane that has global shape; associated with systemic use of tranexamic acid.

**Ocular:** Corneal scarring; dense membrane of the conjunctiva; may occur as a complication following strabismus surgery.

**Clinical:** None.


Conradi Syndrome (Multiple Epiphyseal Dysplasia Congenita; Dysplasia Epiphysealis Congenita; Chondrodystrophia Foetalis Hypoplastic; Calcino sis Universalis; Congenital Calcinifying Chondrodystrophy; Stippled Epiphyses Syndrome; Conradi-Hünermann Syndrome; Chondrodysplasia Punctata)

**General:** Autosomal recessive; manifestations within the first 6 months of life; epiphyseal stippling present at birth; perinatal manifestations include disorganization of the spine, premature echogenicity of femoral epiphyses, and frontal bossing with depressed nasal bridge.

**Ocular:** Hypertelorism; heterochromia iridis (rare); bilateral total congenital cataract appearing at or shortly after birth; primary optic atrophy (rare); bilateral corneal punctate erosions.

**Clinical:** Short limbs (mainly proximal part) resulting in "short-limbed dwarfism"; deformities of hip, knee, and elbow joints by contraction and immobility, and possible transformation of muscles into fibrous tissue as a result; congenital heart defect with calcium deposits in the cardiac valves; skin anomalies (dyskeratosis); mental retardation.


Contact Dermatitis (Dermatitis Venenata)

**General:** Reaction of skin due to contact with foreign material; inflammatory disorder of the skin that may result from immunologic hypersensitivity (allergic contact dermatitis) or cutaneous injury not involving immunologic mechanisms (irritant contact dermatitis) from offending topical agents.

**Ocular:** Keratoconjunctivitis; chemosis; leukoma; corneal ulcer; pruritus of lids.

**Clinical:** Dermatitis; itching, erythema; vesiculation; edema with weeping and crusting.


Convergence Insufficiency Syndrome (Asthenovergence of Stutterheim)

**General:** An exodeviation that is greater at near distances than at far ones; inadequate accommodative and fusional convergence impulses; prevalence is generally considered to be low in children under age 10 years and higher in females.

**Ocular:** Burning, itching, blurred vision; diplopia; difficulty in following moving objects; astigmatism; decreased visual acuity; exotropia; hypermetropia; orthotropia.

**Clinical:** Headache; associated with thyroid eye disease.


Cooley Anemia (Thalassemia; Thalassemia Major; Thalassemia Minor)

**General:** Autosomal dominant in synthesis of the α or β chain of hemoglobin; most prevalent in Mediterranean and Oriental populations.

**Ocular:** Retinal hemorrhages; angioid streaks; macular vascular abnormalities; pigmented chorioretinal scars (black sunbursts); occlusion of peripheral retinal arteries; vitreous hemorrhages.

**Clinical:** Hemolytic anemia; hypochromic anemia.

Cornea Plana

General: Autosomal dominant; may be inherited as autosomal dominant or recessive.

Ocular: Hyperopia; hazy corneal limbus; opacities in corneal parenchyma and marked arcus; posterior embryotoxon; iris and lens abnormalities.

Clinical: Associated with epidermolysis bullosa dystrophica.


Cornea Crystals, Myopathy, and Nephropathy

General: Etiology unknown; may represent an atypical variant of myotonic dystrophy.

Ocular: Retinal pigment epithelial mottling; nystagmus; deep corneal crystals; conjunctival crystals.

Clinical: Weakness and atrophy of pharyngeal, facial, and intrinsic hand muscles; decreased hearing; hypertension; chronic renal disease with decreased glomerular filtration and proteinuria; asymmetrical smile; diminished gag reflex; chronic serous otitis media.


Corneal Dystrophy, Granular Type (Groenouw Type I Corneal Dystrophy)

General: Autosomal dominant; hyaline degeneration with absence of acid mucopolysaccharide deposition; autosomal dominant with complete penetrance; evidence links it with chromosome 5q: Avellino dystrophy is a variant of granular corneal dystrophy with lattice changes.

Ocular: Grayish-white granules in a disk-shaped area of central cornea; hyaline material separates epithelium from Bowman membrane; keratoconus.

Clinical: None.


Corneal Dystrophy, Hereditary Polymorphous Posterior (PCD)

General: Autosomal dominant.

Ocular: Clouding of posterior cornea; reduced number of endothelial cells; thickening and opacities of Descemet membrane; associated with keratoconus; iridocorneal adhesions; glassy membranes; pupillary ectropion (rare).

Clinical: None.


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**Corneal Dystrophy, Lattice Type (Lattice Corneal Dystrophy; LCD; Lattice Dystrophy Type I; Biber-Haab-Dimmer Dystrophy)**

**General:** Autosomal dominant; progression to severe visual impairment by fifth or sixth decade.

**Ocular:** Grayish lines between the centers of cornea and periphery; rounded dots scattered over the cornea; elongated deposits that form reticular pattern in corneal stoma; keratoconus.

**Clinical:** Secondary form of inherited localized amyloidosis inherited as an autosomal dominant trait with low penetrance.


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**Corneal Dystrophy, Macular Type (Groenouw Type II Corneal Dystrophy)**

**General:** Autosomal recessive; onset in first decade, between 5 and 9 years; progressive; acid mucopolysaccharides found in corneal fibroblasts; it has been suggested that the defect may not be limited to the cornea.

**Ocular:** Minute gray, punctate opacities; reduced corneal sensitivity; photophobia; foreign body sensations; recurrent corneal erosions; keratoconus.

**Clinical:** Defect in metabolism of glycoprotein processing.


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**Corneal Dystrophy, Meesmann Epithelial (Meesmann Epithelial Dystrophy of Cornea)**

**General:** Autosomal dominant; rare; onset first year of life; possible that a disturbance of the cytoplasmic ground substance results in cellular homogenization with cyst formation.

**Ocular:** Myriads of fine punctate opacities in epithelium and Bowman membrane of cornea; thickening of the epithelial basement membrane of cornea; keratoconus.

**Clinical:** None.


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**Corneal Hypesthesia, Familial**

**General:** Autosomal dominant; decreased corneal sensation, reflex tearing, blinking, and foreign body sensation.
Ocular: Punctate epithelial erosions; corneal edema; neurotrophic keratitis; corneal ulcers; poor blinking; decreased tearing; decreased corneal sensation.

Clinical: Trigeminal anesthesia; hypoplastic trigeminal nerves and gasserian ganglia.


Corneal Snowflake Dystrophy 303

General: Autosomal dominant; prevalence of green irides.

Ocular: Star-shaped chromatophore-like cells attached to anterior lens capsule; Bitot spots; white flecks on endothelium and Descemet membrane.

Clinical: Lactose intolerance; malabsorption of fat; vitamin A deficiency; dry skin; nevi; freckles.


Corneo-Dermato-Osseous Syndrome (CDO Syndrome; Corneal Dystrophy, Epithelial with Skin and Skeletal Changes) 304

General: Autosomal dominant; similar lesions of palms, soles, and cornea occur III Richner-Hanhart syndrome.

Ocular: Epithelial and stromal corneal changes; photophobia; keratoconus.

Clinical: Palmoplantar hyperkeratosis; brachydactyly, short stature; premature birth; soft teeth.


Cortical Blindness, Retardation, and Postaxial Polydactyly 305

General: Autosomal recessive; combination of cortical blindness, retardation, and postaxial polydactyly.

Ocular: Cortical blindness.

Clinical: Growth and psychomotor development severely retarded; prominent forehead and short nose.


Costen Syndrome (Temporomandibular Joint Syndrome) 306

General: Dental malocclusion; overaction of jaw joint followed by the development of a loose joint due to absorption of the meniscus, condyles, and bone.

Ocular: Headaches; facial pain.

Clinical: Severe neuralgia; "full" ear sensation with reduced hearing; tinnitus; dizziness; associated with vertigo.


Cowden’s Disease

**General:** Autosomal dominant; PTNE gene; tumor suppressor gene

**Ocular:** Cataract; angoid streaks; myopia; multiple trichilemmomas of eyelids

**Clinical:** Malignancies of breast, thyroid, ovary, uterus, colon or bladder; seizures; tremor; mental retardation; cerebellar gangliocytoma


Cranial Nerves, Congenital Paresis

**General:** Autosomal recessive; damage to peripheral nerve and brainstem.

**Ocular:** Paralysis of pupillary sphincter; oculomotor synkinesis; ptosis; exotropia; hypotropia; amblyopia; diplopia.

**Clinical:** Vascular lesion from damage to peripheral nerve and brainstem; generalized developmental delay; seizures; facial paralysis; malformed external ears; abnormalities of motor system of arms and legs.


Cranial Nerves, Recurrent Paresis

**General:** Autosomal recessive; rare, recurrent episodes of Bell palsy and external ophthalmoplegia; lack of iridoplegia distinguishes it from aneurysm.

**Ocular:** External ophthalmoplegia; third nerve palsy.

**Clinical:** Bell palsy; diabetes; polycythemia.


Craniocebral Syndrome (Whiplash Injury)

**General:** Disturbed accommodation is due to a central lesion rather than a peripheral lesion of the ciliary muscle; Homer syndrome observed where a palsy of the cervical sympathetics occurs (see Homer Syndrome).

**Ocular:** General ocular pain; enophthalmos; mild ptosis; reduced ability to accommodate; disturbance in ocular movements; primarily those extraocular muscles innervated by the oculomotor nerve are involved; convergence insufficiency; nystagmus (gaze direction and vestibular, central, peripheral, and mixed type); vestibular impairment in more than 50% of cases; asthenopia; fogging; double vision; myosis; mydriasis; retinal arteriolar pressure may show changes in systolic and diastolic pressure and be more pronounced than changes in the brachial blood pressure; decreased stereoacuity; vitreous detachment.

**Clinical:** Headache; vertigo; dizziness; neck and back pain.


Craniocephalomalacia Syndrome (Cleidocranial Dysostosis Syndrome; Marie-Sainton Syndrome; Scheuthaurer Syndrome; Hulkcantz Anosteoplasia; Mutational Dysostosis Syndrome)

**General:** Autosomal dominant; hypoplastic dysostosis of the skull; shows brachycephaly or platycephaly.
Ocular: Proptosis (unilateral); prominent orbital ridges; greater vertical diameter of the orbit compared with horizontal diameter; hypertelorism; antimongoloid palpebral fissure.

Clinical: Saddle nose; prominent forehead (frontal bossing); hypoplasia of facial bones and clavicles; high arched palate and protruding jaw; oligodontia; pathologic fractures; hyperlaxia of joints; kyphoscoliosis and spina bifida; scoliosis; hemiplegia; spastic paraplegia; mental deficiency; psychosis; incomplete closure of fontanelles; dwarfism; epilepsy; hypoplasia/aplasia of nasal bones; high and narrow orbital opening; absent or diminished paranasal sinuses; small sella; skeletal immaturity due to defect in bone remodeling.


Craniofacial, Deafness, Hand Syndrome

General: Autosomal dominant.

Ocular: Hypertelorism.

Clinical: Flat facial profile; hypoplastic nose with slitlike nares; hearing loss; ulnar deviation of hands.


Craniofrontonasal Dysplasia

General: Sex-linked; unexplained higher prevalence in females.

Ocular: Down-slanting palpebral fissures; hypertelorism.

Clinical: Coronal synostosis; brachycephaly; clefting of the nasal tip; joint anomalies; longitudinally grooved fingernails; other digital anomalies.


Cranimetafysseal Dysplasia Syndrome (Pyle Syndrome; Familial Metafysseal Dysplasia; Bakwin-Krida Syndrome; Leontiasis Ossea)

General: Autosomal recessive; absorption of secondary spongiosa is lacking, with resulting long-bone deformities.

Ocular: Hypertelorism.

Clinical: Splaying of metaphysseal ends of long bones; thick and dense base of bony skull; absent air filling of mastoid process and paranasal sinuses; late dentition; deafness; progressive headache; vomiting; low intelligence; prominent glabella and zygomatic arch; genu valgus deformity.


Cranio-Oro-Digital Syndrome (Otopalatodigital Syndrome; OPD II Syndrome; Facio-Palato-Osseous Syndrome; FPO)

General: Sex-linked; can occur as a sporadic condition.
Ocular: Downward-slanting palpebral fissures (antimongoloid obliquity).

Clinical: Microcephaly; small mouth; midface hypoplasia; cleft palate; flexed, overlapping fingers with syndactyly of digits 3 and 4; syndactyly of toes 2 and 5; bifid uvula; slight deviation of the terminal phalanges of the third fingers; radial deviation of the terminal phalanx of the fourth finger; short first toe and long second toe; short first metacarpal; extra bone in the capitae-hamate complex; small thorax; bowed limbs with absent fibula; mild frontal bossing; conductive hearing impairment; flat facies; broad nasal base; wavy irregular clavicles and ribs; widely spaced eyes; prominent forehead.


**Craniopharyngioma**

**General:** Benign congenital tumors arising from epithelial remnants of Rathke pouch; most common nonglial intracranial tumors in childhood; second most common sellar-parasellar tumor primarily in children or young adults; 35% of cases occur in patients over age 40 years.

**Ocular:** Paresis of third or sixth nerve; optic nerve atrophy; optic neuritis; papilledema; dilation of pupil; diplopia; hemianopsia; nystagmus; scotoma; visual field defects; visual loss.

**Clinical:** Hydrocephalus; infantilism; diabetes insipidus; abnormal sexual development; headaches; acute aseptic meningitis.


**Craniostenosis**

**General:** Skull deformity caused by premature fusion of cranial sutures.

**Ocular:** Optic atrophy; exophthalmos; strabismus; papilledema; nystagmus; ocular colobomas; swollen optic nerves; dissociated eye movements; ptosis; anisometropia; corneal exposure; amblyopia.

**Clinical:** Elevated cerebrospinal fluid; abnormal development of the skull.


**Craniosynostosis-Mental Retardation-Clefting Syndrome**

**General:** Autosomal recessive.

**Ocular:** Choroidal coloboma.

**Clinical:** Craniosynostosis; mental retardation; seizures; dysplastic kidneys; bat ears; cleft lip and palate; beaked nose; small posterior fontanelle.


**Craniotelencephalic Dysplasia**

**General:** Autosomal recessive.
Ocular: Optic nerve hypoplasia.

Clinical: Frontal bone protrusion; encephalocele; craniosynostosis; developmental retardation; agenesis of the corpus callosum; lissencephaly; arhinencephaly.


Cretinism (Hypothyroid Goiter; Hypothyroidism; Juvenile Hypothyroidism; Myxedema) 320

General: Deficient thyroid function.

Ocular: Blepharitis; ptosis; enophthalmos; temporal madarosis; decreased tear secretion; glaucoma; proptosis; optic atrophy; optic neuritis; blue dot cataract; conjunctivitis; scleritis; optic disk hemorrhage and arcuate scotoma associated with glaucoma.

Clinical: Myxedema; larynx and tongue swollen; hoarse speech; dry, yellowish skin; slow pulse; mental retardation; infertility; pericardial effusion; cardiac enlargement; physical development retarded.


Creutzfeldt-Jakob Syndrome (Spastic Pseudosclerosis; Corticostriatospinal Degeneration; Disseminated Encephalopathy; Heidenhaim Syndrome; Presenile Dementia with Spastic Paralysis; Presenile Dementia-Cortical Degeneration Syndrome) 321

General: Heredofamilial occurrence; caused by degenerative changes in cerebral cortex, basal ganglion, and spinal cord; disease is progressive; begins in middle or later age; occurs in both sexes.

Ocular: Cortical blindness; myoclonic conjugate eye movements; paralysis of seventh nerve; ptosis; dyschromatopsia; homonymous hemianopsia; nystagmus; slow vertical saccades; mild demyelination of the optic nerve.

Clinical: Mental deterioration; psychosis; stupor; weakness and stiffness of extremities; slow development of pyramidal signs; loss of reflexes; tremor, rigidity; dysarthria; ataxia; myoclonus; convulsive seizures; cerebellar ataxia decerebrate posture.


Cri-du-Chat Syndrome (Cat-Cry (5p-) Syndrome; Crying Cat Syndrome; BI Deletion Syndrome; Lejeune Syndrome) 322

General: Short arm deletion of a no. 5 chromosome (5p-); increased inheritance risk; 13% have one parent with balanced translocation; female preponderance 2:1 (see Wolf Syndrome).

Ocular: Hypertelorism; epicanthal folds; antimongoloid slanting of palpebral fissures; strabismus; increased tortuosity of retinal vessels.

Clinical: High-pitched, plaintive cry by an infant (reminiscent of a crying cat); mental retardation; broad nasal root; micrognathia or retrognathia; low-set ears; simian crease; congenital heart defect; small larynx and epiglottis.

Criswick-Schepens Syndrome (Familial Exudative Vitreoretinopathy)

**General:** Familial exudative vitreoretinopathy, similar to retrolental fibroplasia; bilateral; slowly progressive; full-term babies; no oxygen therapy; autosomal dominant; may be inherited as X-linked or autosomal dominant condition.

**Ocular:** Posterior vitreous detachment of organized membranes of vitreous; snowflake-like opacities of vitreous; heterotropia of macula; subretinal exudates; retinal detachment; degenerative retinal changes; retinal hemorrhage; retinal folds; enophthalmos; ptosis; intraretinal exudate; vitreous hemorrhage; ambylopia; falciform retinal fold.

**Clinical:** Normal general development; normal birth weight.


Crohn Disease (Granulomatous Ileocolitis)

**General:** Autoimmune or hypersensitivity inflammatory change; slight prevalence in males; Jewish people most frequently affected; onset at any age; more severe in young people; remission; relapses.

**Ocular:** Recurrent conjunctivitis; marginal corneal ulcers; keratitis; blepharitis; dry eye; scleritis; episcleritis; iris atrophy; uveitis; pupil immobility and dilatation; macular edema; macular hemorrhages; extraocular muscles palsy; vitreal haze; retinal vasculitis; subconjunctival nodules; conjunctival ulcer; pannus; acute dacryoadenitis; orbital pseudotumor.

**Clinical:** Inflammatory bowel disease; abdominal distention; tenderness of abdomen; mass in right lower quadrant of abdomen; diarrhea; abdominal cramps; bloating; flatulence; weight loss; nervousness; tension; depression; pyoderma gangrenosum.


Crome Syndrome

**General:** Fatal; death usually at 4 to 8 months; autosomal recessive.

**Ocular:** Congenital cataracts.

**Clinical:** Epileptic seizures; mental retardation; small stature; renal tubular necrosis; encephalopathy.


Cronkhite-Canada Syndrome

**General:** Hypoproteinemia; hypocalcemia
Ocular: Nuclear and posterior subcapsular cataract

Clinical: Gastrointestinal polyposis; hyperpigmentation of skin; hair loss; nail atrophy

Goto A. Cronkhite-Canada syndrome: observation of the 180 cases reported in Japan. Nippon Rinsho 1991; 49: 221-226


Crouzon Syndrome (Dysostosis Craniofacialis; Oxycephaly; Craniofacial Dysostosis; Parrot-Head Syndrome; Möbius-Crouzon Syndrome; Hereditary Craniofacial Dysostosis)

General: Autosomal dominant; manifestations present at birth.

Ocular: Bilateral exophthalmos; hypertelorism (wide interpupillary distance); obliquity of palpebral fissures with outer canthus slanting downward; nystagmus; exotropia; upper field defects due to pressure upon the optic nerve on its lower part; bluish sclera; exposure keratitis in extreme exophthalmos; cataract; papilledema; secondary optic atrophy; corneal dystrophy; ptosis; strabismus; keratoconus.

Clinical: Prognathism; maxillary hypoplasia with short upper lip; synostosis of coronal and lambda sutures; parrot-beaked nose (psittachosrhina); widening temporal fossae; headaches.


Crowded Disc Syndrome (Bilateral choroidal folds and optic neuropathy)

General: Elevated intracranial ruled out

Ocular: Bilateral choroidal folds; optic disk congestion; optic atrophy; hyperopia; shortened axial length

Clinical: Elevated intracranial ruled out


CRST Syndrome (Thibierge-Weissenbach Syndrome; Calcinosi, Raynaud Phenomenon, Sclerodactyly, and Telangiectasia)

General: Scleroderma variant; possible autosomal dominant transmission; resembles Rendu-Osler-Weber disease; well-established association with primary biliary cirrhosis; prevalent in females; average age of onset 45 years; reported only in whites.

Ocular: Conjunctival hyperemia; keratitis; profuse tear lysozyme; gritty, burning sensations; bilateral optic neuropathy.

Clinical: Dermal and subcutaneous calcinosis; Raynaud phenomenon; sclerodactyly; telangiectasia; calcinosis cutis; multiple intracranial aneurysms.


Cryoglobulinemia

General: Hematologic disorder in which a group of proteins precipitate from serum on exposure to cold but redissolve when warmed. Frequently occurs in diseases of reticuloendothelial systems, multiple myeloma, and Hodgkin disease.
Ocular: Congestive retinopathy; vitreous hemorrhage; retinal detachment; rubeosis iridis; neovascular glaucoma; optic disk edema; conjunctival vascular congestion.

Clinical: Widespread intravascular coagulation in small vessels throughout body.


**Cryptococcosis (Torulosis)**

**General:** A pulmonary infection caused by *Cryptococcus neoformans,* a saprophyte found in weathered pigeon droppings, soil, and unpasteurized cow’s milk; infection acquired through respiratory system and usually manifests as meningoencephalitis; higher incidence in patients with AIDS.

**Ocular:** Blurred or poor vision; diplopia; uveitis; papilledema; retinal detachment; retinal hemorrhage and exudates; secondary glaucoma; vitreous reaction; retinitis; proptosis; a mass over the optic nerve head; disease process can be bilateral or unilateral; cranial nerve VI palsy; visual loss; conjunctivitis.

**Clinical:** Severe headache; dizziness; ataxia; vomiting; tinnitus; memory disturbances; jacksonian convulsions; fever usually is absent; occurs frequently in patients with leukemia or lymphoma.


**Cryptophthalmia Syndrome (Cryptophthalmos Syndactyly Syndrome; Fraser Syndrome)**

**General:** Autosomal recessive.

**Ocular:** Microphthalmia; epibulbar dermoid; cryptophthalmos; enophthalmia; eyebrows partially or completely missing; skin from forehead completely covers one or both eyes, but the globes can be palpated beneath the skin; in unilateral cases, the fellow eye may present lid coloboma; buphthalmos; conjunctival sac partially or totally obliterated; absence of trabeculae, Schlemm canal, and ciliary muscles; cornea is differentiated from the sclera; lens anomalies from complete absence to hypoplasia, dislocation, and calcification.

**Clinical:** Syndactyly (finger, toes) (about 40%); coloboma of alae nasi and nostrils; urogenital abnormalities, including pseudohemaphroditism and renal hypoplasia; abnormal, bizarre hairline; narrow external auditory meatus and malformation of ossicles; cleft lip and palate may occur; atresia or hypoplasia of larynx in some cases; hoarse voice; dysplastic pinna; meatal stenosis; glottic web and subglottic stenosis.


**Curly Hair-Ankyloblepharon-Nail Dysplasia Syndrome (CHANDS)**

**General:** Autosomal recessive with pseudodominance.

**Ocular:** Congenital ankyloblepharon (fused eyelids).

**Clinical:** Curly hair; hypoplastic nails.

Curtius Syndrome (Ectodermal Dysplasia with Ocular Malformations) 334

**General:** Ectodermal dysplasia of the skin with ocular involvement; occasional combinations such as hypoplasia of nails and hair, and malformation of cheeks, temples, and breasts.

**Ocular:** Hypertelorism; sparse eyelashes (hypotrichosis); nystagmus; decreased tear secretion; congenital cataract; tapetoretinal degeneration; coloboma.

**Clinical:** Hidrotic ectodermal dysplasia; ichthyosis vulgaris; acrofacial syndactylic dysostosis; hypodontia.


Cushing (1) Syndrome (Adrenocortical Syndrome; Hyperadrenalism Syndrome; Pituitary Basophilism; Suprarenal Syndrome) 335

**General:** Excessive secretion of adrenal cortical hormones due to primary or secondary adrenal hyperplasia or induced by adrenal or extraadrenal neoplastic tissue; common in females of childbearing age.

**Ocular:** Proptosis (rare); ocular muscle palsies; uncharacteristic visual field changes (not necessarily resembling bitemporal hemianopsia); optic nerve and/or chiasm compression either unilateral or bilateral; posterior subcapsular cataract; central serous retinopathy; Lisch nodules.

**Clinical:** Hirsutism; obesity; "buffalo hump"; hypertension; diabetes; skin pigmentation; osteoporosis; abdominal striae; polycythemia and lymphopenia; weakness; nervousness; irritability; dysmenorrhea.


Cushing (2) Syndrome (Angle Tumor Syndrome; Cerebellopontine Angle Syndrome; Pontocerebellar Angle Tumor Syndrome; Acoustic Neuroma Syndrome) 336

**General:** Tumor involving cranial nerves V, VI, VII, and VIII and brainstem; occurs between ages 30 and 45 years.

**Ocular:** Paresis orbicularis muscle (VII); paresis external rectus muscle (VI); mixed nystagmus with head tilt; palsies of extraocular muscles are accounted for by increased intracranial pressure if the aqueduct of Sylvius is closed by the growing tumor; decreased corneal reflex V (homolateral and early sign); bilateral papilledema (increased intracranial pressure).

**Clinical:** Deafness (homolateral); labyrinth function disturbed or lost; tinnitus; hyperesthesia of the face; homolateral facial nerve paresis (total paralysis rare); hoarseness; difficulties in swallowing; unilateral limb ataxia; gait ataxia; nuchal headache; emesis; facial pain, numbness and paresis; progressive unilateral hearing loss.


Cushing (3) Syndrome (Chiasmal Syndrome) 337

**General:** Suprasellar meningioma, aneurysm in the anterior part of the circle of Willis, and craniopharyngioma are the three most common lesions; usually occurs in adult patients.

**Ocular:** Bitemporal hemianopsia (progressive); in early stages, optic disk may appear normal or only slightly pale; later, sharp, border-lined white optic disk.

**Clinical:** Cranioopharyngiomas; pituitary adenoma; aneurysm; chiasmal glioma; nasopharyngeal carcinoma; germinoma; glioma; choristoma; chordoma; hemangioma; leukemia; lymphoma; metastatic tumor; arteriovenous malformation; sarcoidosis; pituitary abscess; sphenoid sinus mucocele; arachnoid cyst.


### Cutis Marmorata Syndrome (Marble Skin Syndrome)

**General:** Prominent in children; etiology unknown; possibly a mild form of livedo reticularis; in severe cases, vessel dilation and skin changes may be permanent; occurs with exposure to cold and subsides in a warm environment; extremities and pectoral region most frequently affected.

**Ocular:** Congenital glaucoma; corneal and scleral thinning; staphyloma; sclerotic appearance to trabecular meshwork; corneal edema; cataracts; optic nerve atrophy; heterochromia iridium; iris anterior layer dysplasia; intraoperative suprachoroidal hemorrhage (rare).

**Clinical:** Bluish-red mottling of skin; spasmodic narrowing of arterioles with dilation of vessels; ulceration and scaling of skin; congenital hypothyroidism.


### Cystic Fibrosis Syndrome (Fibrocystic Disease of Pancreas)

**General:** Autosomal recessive; Caucasians; lungs, pancreas, and salivary glands are mainly involved.

**Ocular:** Ischemic retinopathy caused by carbon dioxide retention and chronic respiratory insufficiency; vein congestion and capillary dilation around the optic nerve; retinal hemorrhages; macular degeneration; papilledema; optic atrophy; xerosis of conjunctiva; optic neuritis; abnormal pupillary responses; decreased contrast sensitivity.

**Clinical:** Failure to gain weight properly; recurrent pulmonary infections; salty skin; pancreatic insufficiency with malabsorption; abdominal cramps; diarrhea; increased appetite; dyspnea; chronic cough; production of viscous tenacious sputum; fever; retarded growth; delayed puberty; distended abdomen; hyperresonant chest; depressed diaphragm; clubbing of fingers.


### Cysticercosis

**General:** Caused by *Taenia solium*.

**Ocular:** Tenonitis; endophthalmitis; optic atrophy; papilledema; retinal detachment; retinal hemorrhages and exudates; vitreal hemorrhages; hypopyon; uveitis; paresis of extraocular muscles; periretinal proliferation; cysts may be present almost anywhere in or around the eye; orbital lesion; extraocular myositis; subretinal cysticercosis; acute preseptal cellulitis.

**Clinical:** Dead larvae may cause muscle pain; weakness; fever; eosinophilia; calcification of tissues.


Cystinuria  341

**General:** Caused by abnormal protein metabolism; recessive or incomplete recessive.

**Ocular:** Pigmentary retinopathy; gyrate atrophy.

**Clinical:** Impaired intestinal absorption and renal tubular reabsorption of cystine, lysine, arginine, and ornithine leads to excretion of these dibasic amino acids into urine; associated with renal dwarfism, pyramidal and extrapyramidal symptoms, and deaf mutism.


Cytomegalic Inclusion Disease (Cytomegalovirus; Congenital Cytomegalic Inclusion Disease)  342

**General:** Cytomegalovirus passes transplacentally from an asymptomatic mother to fetus.

**Ocular:** Uveitis; cataract; optic atrophy; inclusion bodies in the aqueous humor; severe conjunctivitis; corneal opacities; microphthalmos; strabismus; dactylopathy; chorioretinitis; cytomegalovirus retinitis (most common cause of acquired viral retinitis, primarily because of the AIDS virus); glaucoma.

**Clinical:** Cerebral calcifications; microcephaly; mental retardation; inclusion bodies in the urine; spastic diplegia; seizures; cerebellar hypoplasia; intraventricular hemorrhage; hydrocephalus.


Danbolt-Closs Syndrome (Acrodermatitis Enteropathica; Brandt Syndrome)  343

**General:** Etiology unknown; autosomal recessive; occurs in both sexes with onset in early infancy; characterized by intermittent simultaneous occurrence of diarrhea and dermatitis with failure to thrive.

**Ocular:** Loss of eyebrows; blepharitis; ectropion; loss of eyelashes; photophobia; conjunctivitis; scattered superficial corneal opacities; keratitis; lacrimal punctal stenosis; corneal superficial punctate lesions, nebulous subepithelial opacities and linear epithelial erosions.

**Clinical:** Symmetrical skin eruptions on hands, feet, elbows, knees, and buttocks usually dry up to an erythematousquamous type; glossitis and stomatitis; alopecia; paronychia with nail dystrophy; gastrointestinal disturbances; diarrhea (intermittent).

Cherry PM, Falcon MG. Punctual stenosis-caused by idoxuridine or acrodermatitis enteropathica? *Arch Ophthalmol* 1976; 94:1632.


Dandy-Walker Syndrome (Atresia of the Foramen of Magendie)  

**General:** Manifested in infants; malformation and stenosis of the foramina of Luschka and Magendie; dilation of fourth ventricle.

**Ocular:** Ptosis; sixth nerve paralysis; papilledema.

**Clinical:** Hydrocephalus (varies in severity) with enlargement of the skull and thinning of the bone predominantly in occipital region; loss of tendon reflexes; basilar impression; scoliosis; hydromelia.


Darier-White Syndrome (Keratosis Follicularis; Dyskeratosis Follicularis Syndrome; Psorospermosis)  

**General:** Unknown etiology; defect in the synthesis, organization, and maturation of tonofilament-desmosome complex; irregular dominant inheritance; both sexes equally affected, with onset in childhood; chronic but relatively benign and more aggravated in the summer.

**Ocular:** Conjunctival keratosis; bilateral corneal subepithelial infiltrations and sometimes corneal ulceration; cataract formation (rare).

**Clinical:** Confluent flesh-colored keratotic papules on head, neck, back, abdomen, and groin; small stature; mild mental retardation; hair loss; genital hypoplasia; oral-mucosal lesions; hypertrophic flexural involvement; acral signs.


Dawson Disease (Dawson Encephalitis; Subacute Sclerosing Panencephalitis; Inclusion-Body Encephalitis)  

**General:** Sclerosing panencephalitis classified as a degenerative, progressive neurologic disorder caused by a measles virus infection of the central nervous system.

**Ocular:** Nystagmus; ptosis; papilledema; optic neuritis; macular pigmentation and degeneration; focal retinitis; ocular motor palsies; optic atrophy; preretinal vitreous membrane; exophthalmos; visual agnosia; chorioretinitis; retinal vasculitis; macular chorioretinitis.

**Clinical:** Chronic inflammation of brain with neuronal degeneration; gliosis; eosinophilic inclusion bodies in brain tissue; decline in intellect; behavioral changes; slurred speech; drooling; motor abnormalities; disorientation; seizures; death.


Kovacs B, Vastag O. Fluoroangiographic picture of the acute stage of the retinal lesion in subacute sclerosing panencephalitis. *Ophthalmologica* 1978; 177:264-269.


DeBarsy Syndrome

**General:** Rare progeroid syndrome associated with characteristic ocular, facial, skeletal, dermatologic, and neurologic abnormalities.

**Ocular:** Congenital corneal opacification (loss of Bowman layer); cataracts.

**Clinical:** Short stature, pectus excavatum, skeletal dysplasia with short legs, multiple joint dislocations, especially involving the hands; skin redundancy (as seen in cutis laxa); midface hypoplasia; thin transparent skin with prominent superficial veins; frontal bossing and aged, progeroid facies; early death; hypotonia; mental retardation; brisk deep tendon reflexes.


Deerfly Fever (Francis Disease; Rabbit Fever; Tularemia; Deerfly Tularemia)

**General:** Acute infectious disease caused by *Francisella (Pasteurella) tularensis.*

**Ocular:** Chemosis; conjunctivitis; corneal ulcer; endophthalmitis; dacryocystitis; optic atrophy; iris prolapse; chalazion; corneal opacity; pannus.

**Clinical:** Local ulcerative lesion; suppuration of regional lymph nodes; fever; prostration; myalgia; severe headache; pneumonia.


Degos Syndrome (Malignant Atrophic Papulosis; Degos-Delort-Tricot Syndrome; Cutaneointestinal Syndrome; Kolmeier-Degos Syndrome)

**General:** Rare cutaneovisceral disease; male preponderance; death occurs within a few months after diffuse eruption of skin lesions; multiple cerebral infarcts and/or thrombosis of small arteries; lymphocytic-mediated necrotizing vasculitis with mucin deposits in the dermis.

**Ocular:** Atrophic skin of eyelids; intermittent diplopia; conjunctiva may be atrophic; telangiectasias of conjunctiva with microaneurysms; peripheral choroiditis; papilledema has occurred with progressive central nervous system involvement; necrotic papules of lids, bulbar conjunctivae, and episcleral tissues.

**Clinical:** Porcelain-white skin lesions (asymptomatic and diffuse); anorexia and/or weight loss; gastrointestinal involvement; peritonitis; intermittent paresthesias with early central nervous system involvement; signs of progressive cerebral and cerebellar atrophy; peripheral telangiectatic rim.

Kohlmeier W Multiple Hautnekrosen bie Thromboangitis Obliterans. *Arch Dermatol Syphil* 1941; 181:783-792.

Dejean Syndrome (Orbital Floor Syndrome)

**General:** Usually secondary to a traumatic lesion involving the floor of the orbit.

**Ocular:** Enophthalmos; exophthalmos; lid hematoma; diplopia due to displacement of the globe or restricted function of the inferior rectus and/or inferior oblique muscles; orbital emphysema.

**Clinical:** Severe pain in superior maxillary region; numbness in area of first and second branches of trigeminal nerve; nausea and vomiting.

Dejerine-Klumpke Syndrome (Lower Radicular Syndrome; Klumpke Syndrome; Klumpke Paralysis)  

**General:** Lesion involving the inferior roots of the brachial plexus with nerves derived from the eighth cervical and first thoracic root.

**Ocular:** Enophthalmos; ptosis; narrowed palpebral fissure; miosis.

**Clinical:** Paralysis and atrophy of the small muscles of forearm and hand (flexor carpi ulnaris, flexor digitorum, interossei, thenar, hypothenar); decreased sensation or increased sensibility on the inner side of the forearm.


Dejerine-Roussy Syndrome (Retrolenticular Syndrome; Dejerine-Roussy Syndrome; Posterior Thalamic Syndrome; Thalamic Hyperesthetic Anesthesia Syndrome; Thalamic Syndrome)  

**General:** Posterior thalamic lesion.

**Ocular:** Hemianopsia when the thalamogeniculate artery is thrombosed near its origin from the posterior cerebral artery, because there is involvement of the medial aspect of the lateral geniculate body; if the posterior cerebral artery is thrombosed, complete hemianopsia with macular sparing results; clinical transient acute esotropia; possible association between thalamic lesions and mononuclear supranuclear palsy; unilateral blepharospasm.

**Clinical:** Sensory disturbances: contralateral; hemiataxia: contralateral; hemiplegia (transient): contralateral; choreoathetoid movements: contralateral; spontaneous pain: contralateral.


Deletion of Chromosome 12q 15- q23  

**General:** Interstitial deletion of chromosome 12; autosomal dominant or autosomal recessive

**Ocular:** Corneal plana

**Clinical:** Deletion of the KERA gene


De Lange Syndrome (I) (Congenital Muscular Hypertrophy Cerebral Syndrome; Brachmann-De Lange Syndrome)  

**General:** Etiology not known; autosomal recessive inheritance.

**Ocular:** Antimongoloid slant of palpebral fissures; mild exophthalmos; hypertrichosis of eyebrows; long eyelashes; telecanthus; ptosis; blepharophimosis; nystagmus on lateral gaze; constant coarse nystagmus; strabismus; alternating exotropia; high myopia; anisocoria; chronic conjunctivitis; blue sclera; pallor of optic disk.

**Clinical:** Mental retardation; growth retardation; extrapyramidal motor disturbances; multiple skeletal abnormalities with congenital muscular hypertrophy; long philtrum; thin lips; crescent-shaped mouth.


Demodicosis

**General:** Demodex folliculorum and Demodex brevis infestation; exact role in causing blepharitis is unclear; most patients are asymptomatic.

**Ocular:** Blepharitis; follicular distention and hyperplasia; lid hyperemia; lid hyperkeratinization; madarosis; meibomian gland destruction; mite colonies of eyelashes and eyebrows.

**Clinical:** Pruritus.


De Morsier Syndrome (Septooptic Dysplasia)

**General:** Absence of the septum pellucidum; agenesis of corpus callosum; enlargement of ventricles; infundibulum primary site of structural derangement in patients with optic nerve hypoplasia.

**Ocular:** Optic disk hypoplasia; bitemporal hemianopia; poor vision; nystagmus.

**Clinical:** Growth retardation; pituitary insufficiency; diabetes insipidus; normal cognitive development; intact neurologic status; normal language development; late appropriate behavior; subtle visual alterations, problems; association between optic nerve hypoplasia and cerebral hemispheric abnormalities, especially schizencephaly, due to migration anomalies.


Dengue Fever

**General:** Endemic over the tropics and subtropics; caused by four distinct serogroups of dengue viruses, types 1,2,3, and 4, group B arboviruses; transmitted solely by mosquitoes of the genus Aedes.

**Ocular:** Lid edema; conjunctivitis; ocular and retrobulbar pain accentuated by ocular movement; dacryoadenitis; keratitis; corneal ulcer; iritis; retinal or vitreous hemorrhages; ocular motor paresis; optic atrophy.

**Clinical:** Hemorrhagic fever, severe headache; backache; joint pain; rigors; insomnia; anorexia; loss of taste; epistaxis; rashes; maculopapular rash; myalgia; human infection with of four serotypes of Dengue virus causing two diseases; classic Dengue fever; Dengue hemorrhagic fever (50% mortality).


Dental-Ocular-Cutaneous Syndrome 358

**General:** Abnormal tooth roots; distinctive features separate this syndrome from oculodentodigital or faciodentodigital syndromes.

**Ocular:** Entropion lower eyelids; glaucoma (juvenile type).

**Clinical:** Unusual upper lip with lack of "cupid's bow" and thickening and widening of the philtrum; syndactyly; cutaneous hyperpigmentation overlying the interphalangeal joints; clinodactyly; single conical roots in all primary teeth and permanent first molars; scant body hair; horizontal ridging of fingernails.


Dermatitis Herpetiformis (Duhring-Brocq Disease) 359

**General:** Malignant; atypical; does not respond well to sulfone or sulfapyridine therapy; uncommon; autoimmune blistering dermatosis; pruritic eruption involving the scalp, buttocks, lower back, and extensor surface of arms; autoantibody is generally of immunoglobulin A class causing deposition at the dermal-epidermal junction.

**Ocular:** Bullae of conjunctiva, skin, and mucous membranes; blisters are intraepithelial (acantholysis) and usually do not leave scars; epithelium desquamates in patches; corneal and conjunctival vascularization; symblepharon; cataract.

**Clinical:** Vesicles; erythema; pruritus; burning; eruption classically involves extensor surface of the knees, elbows, buttocks, sacrum, scalp, and scalp.


Dermatophytosis (Epidermophytosis; Epidermomycosis; Rubrophytia; Tinea; Trichophytosis) 360

**General:** Superficial infection of the skin; ringworm fungi; most frequently seen in children during hot, humid weather.

**Ocular:** Conjunctivitis; corneal ulcer; madarosis; scaly rash; folliculitis; blepharitis; lid edema.

**Clinical:** Scalp, facial, and lid ringworm lesions.


Dermoid (Dermoid Choristoma; Dermoid Cyst; Dermolipoma; Lipodermoid) 361

**General:** Benign tumors composed of epidermal tissue, dermal adnexal structures, skin appendages, hair follicles, sebaceous gland, and sweat glands; slowly growing.

**Ocular:** Dermoid of conjunctiva, cornea, and lids; keratitis; extraocular muscle paralysis; exophthalmos; astigmatism; visual loss; orbital lesions causing diplia and proptosis; may be connected with the lacrimal canaliculum.

**Clinical:** Subcutaneous dermoids of the skin; aplasia cutis congenita possibly associated with strabismus has been reported.


Desert Lung and Cataract Syndrome 362

**General:** Nonoccupational pneumoconiosis; common in desert areas; excessive exposure to atmospheric dust; inhalant of fine, sandy dust.
Ocular: Cataracts (posterior subcapsular most prevalent); corneal opacities.

Clinic: Miliary infiltrates; thickening of bronchial walls; mild restrictive changes with pulmonary function.


Devic Syndrome (Ophthalmonecephalomyelopathy; Optic Myelitis; Neuromyelitis Optica)

General: Etiology unknown; frequent between the ages of 20 and 50 years; mortality rate up to 50%; associated with chickenpox.

Ocular: Ptosis is rare; ocular muscle palsy (rare); abducens and oculomotor palsy; paralysis of conjugate gaze; blindness; onset usually very sudden in one eye, followed soon by blindness in the other eye; miosis; bilateral optic neuritis (unilateral involvement is rare); optic atrophy; pupillary dysfunction.

Clinical: Prodromal signs: headache; sore throat; fever and malaise; ascending myelitis with resulting pain, which may be severe; numbness; weakness; paralysis.


Diabetes Mellitus (Willis Disease)

General: Metabolic disorder that affects carbohydrate and lipid metabolism; familial, usually occurs over age 40 years; frequently occurs in people who are overweight.

Ocular: Ectropion uveae; rubeosis iridis; cataract; optic nerve atrophy; microaneurysms of retina; lipemia retinalis; dilation of the retinal veins; cotton-wool spots; hard yellow exudates; irregular sheathing of retinal veins; macular edema; retinal neovascularization; vitreal hemorrhages; hypotony; glaucoma; asteroid hyalosis; paresis of third or sixth nerve; anterior ischemic optic neuropathy; decreased corneal sensation.

Clinical: Hyperglycemia with resulting glycosuria and polyuria; imperfect combustion of fats leading to acidosis, dyspnea, lipemia, ketonuria, and coma; atherosclerosis; pruritus; lowered resistance to pyogenic infections; proteinuria predicts vision loss, indicates more advanced diabetic retinopathy; glycosylated hemoglobin also is a risk factor for retinopathy and macular edema.


Dialinas-Amalric Syndrome (Amalric-Dialinas Syndrome; Deaf Mutism-Retinal Degeneration Syndrome)

General: Retinal pigmentary disturbances and deafness as outstanding findings but without severe general systemic disorders as seen in the syndromes of Hallgren, Cockayne, Alport, Laurence-Moon-Bardet-Biedl (see Hallgren Syndrome; Cockayne Syndrome; Alport Syndrome; Laurence-Moon-Bardet-Biedl Syndrome).

Ocular: No night blindness but heterochromia iridis; atypical retinitis pigmentosa with small, scattered, fine-pigmented deposits in the macular region with some accumulations and accompanied by small white and yellow spots.
Clinical: Deaf mutism.

### Diamond Blackfan Syndrome

**General:** Rare, congenital hematologic disorder characterized by isolated erythroid hypoplasia (hypoplastic anemia).

**Ocular:** Strabismus, hypertelorism, microphthalmos, and infantile glaucoma.

**Clinical:** May have musculoskeletal abnormalities.


### Diencephalic Syndrome (Diencephalic Epilepsy Syndrome; Autonomic Epilepsy Syndrome; Penfield Syndrome; Anterior Diencephalic Autonomic Epilepsy Syndrome)

**General:** Occurs in males at ages 6 to 7 years; caused by hypothalamic dysfunction and a localized epileptic stimulus originating in the dorsal nucleus of the thalamus.

**Ocular:** Proptosis (occasionally); excessive lacrimation; pupillary abnormalities.

**Clinical:** Abdominal pain; headache; irritability; rapid pulse; elevated blood pressure; salivation; hiccup; chills; dyspnea (Cheyne-Stokes); seizures (possible); sudden onset of vasodilation of the skin (cervical sympathetic).


### Diffuse Keratoses Syndrome

**General:** Ophthalmic complications and manifestations following a group of dermatologic conditions for which the etiology is unknown, including scleroderma, CRST syndrome, Rendu-Osler-Weber syndrome, progressive systemic sclerosis with calcinosis, Spanlang- Tappeiner syndrome, and Savin syndrome (see Rendu-Osler-Weber Syndrome; Progressive Systemic Sclerosis; Spanlang- Tappeiner Syndrome; Savin Syndrome; CRST Syndrome; Scleroderma).

**Ocular:** Dermatologic and cicatricial skin changes; hydrophphthalmos; corneal changes with nodular thickening in the stroma, which worsens in fall and improve during spring (particularly Savin syndrome); retinal phlebitis.

**Clinical:** Ichthyosis (usually congenital); microcephalus (CRST); facial dermatoses; deafness; urticarial (allergic) manifestations; subcutaneous calcinous plaques, mainly on hands; sclerodactyly.


### Diffuse Unilateral Subacute Neuroretinitis Syndrome (DUSN; Unilateral Wipeout Syndrome; Wipeout Syndrome)

**General:** Caused by a nematode that is not *Toxocara canis*, that is, at least two nematodes of different sizes; usually occurs in children or young adults; nematode may remain viable in eye for 3 years or longer.

**Ocular:** Vitreitis; papillitis; gray-white lesions of retina; optic atrophy; retinal vessel narrowing; diffuse pigment epithelial degeneration; endophthalmitis; nematode in fundus; the pathognomonic finding in DUSN is the presence of a motile intraocular nematode.

**Clinical:** Weight loss; lack of appetite; cough; fever; pulmonary infiltration; hepatomegaly; leukocytosis; persistent eosinophilia.
Dimmer Syndrome (Keratitis Nummularis)  
**General:** Onset after minor ocular trauma.

**Ocular:** Photophobia; ocular pain; excessive lacrimation; discoid infiltration of superficial layers of cornea without adjacent conjunctivitis.


Diphtheria  
**General:** Acute infectious disease caused by *Corynebacterium diphtheriae*; severity is dependent upon the amount of exotoxin absorbed prior to initiation of specific therapy.

**Ocular:** Conjunctivitis; xerophthalmia; keratitis; corneal ulcer; blepharitis; cellulitis of lid; meibomianitis; ptosis; dacrocystitis; cataract; central retinal artery occlusion; optic neuritis; accommodative spasm or paralysis; convergence paralysis; divergence paralysis; paralysis of third, fourth, or sixth nerve; paralysis of accommodation (in children); ocular motor nerve paresis; choroiditis; cranial neuropathies involving the trigeminal, vagus, and hypoglossal cranial nerves; myocarditis.

**Clinical:** Local inflammatory lesion, with effect on heart, kidneys, and nervous system.


Dirofilariasis  
**General:** Caused by parasite nematode *Dirofilaria*.

**Ocular:** Chemosis; tenonitis; proptosis; granuloma of conjunctiva, eyelid, orbit, and sclera; periocular soft tissue mass.

**Clinical:** Pulmonary symptoms; coughing; cardiac infection; "coin" lesions in the lung; pruritus, most commonly in subepithelial tissues of eyelids, fingers, cheeks, breast, abdomen, and conjunctivae (rarely); allergic reactions; hemoptysis.


Disseminated Intravascular Coagulation  
**General:** Intravascular coagulation in small vessels throughout the body.

**Ocular:** Serous retinal detachment; choroidal, retinal, and vitreous hemorrhage, and hyphema; recurrent choroidal hemorrhagic detachment.

**Clinical:** Hypocoagulable, hemorrhagic state.

Disseminated Lupus Erythematosus (Systemic Lupus Erythematosus; Lupus Erythematosus; Kaposi-Libman-Sack Syndrome) 374

**General:** Possible etiology includes viral infections and genetic predisposition; immunologic abnormalities.

**Ocular:** Keratitis; keratoconjunctivitis sicca; corneal ulcer; optic nerve atrophy; optic neuritis; papilledema; arteritis; central retinal vein occlusion; retinal detachment; microaneurysm; scleritis; uveitis; ptosis; conjunctivitis; paralysis of third nerve; homonymous hemianopsia; multifocal microinfarcts; mydriasis; nystagmus; proptosis; orbital myositis; pseudoretinitis pigmentosa; photophobia.

**Clinical:** Polyarthritis; morning stiffness; fever; malaise; fatigue; polyserositis; renal disease; central nervous system disease; anemia; leukopenia; maculopapular rash in a "butterfly" distribution over malar region; alopecia.


Disseminated Sclerosis (Multiple Sclerosis) 375

**General:** Disseminated demyelination affecting white matter of the brain, spinal cord, and optic nerves; etiology unknown.

**Ocular:** Nystagmus; ptosis; myokymia; optic atrophy; papillitis; optic neuritis; anisocoria; Argyll Robertson pupil; Marcus Gunn pupil; hipoacusis; decreased or absent papillary reaction to light; peripheral field defects; gaze palsy; paralysis of third or sixth nerve; uveitis; occlusion; Uhthoff symptom (reduction of visual acuity with exercise or ocular hyperthermia); pars planitis; retinal venous sheathing; retinitis; granulomatous uveitis.

**Clinical:** Incoordination; paresthesia; spasticity; tic douloureux; urinary frequency and infections; progressive disability; paralysis; death.


Distichiasis (Distichiasis with Congenital Anomalies of the Heart and Peripheral Vasculature) 376

**General:** Autosomal dominant.

**Ocular:** Double rows of eyelashes; congenital ectropion; absence of meibomian glands; replacement of dense collagenous tissue of the tarsal plates by loose areolar tissue.

**Clinical:** Congenital heart defects; ventricular septal defects; stress-induced asystole; visible varicosities; chronic venous disease of the legs; sinus bradycardia.


Diverticulosis of Bowel, Hernia, Retinal Detachment 377

**General:** Autosomal recessive.

**Ocular:** Severe myopia; esotropia; retinal detachment.

**Clinical:** Femoral or inguinal hernias; diverticula of bowel or bladder.


Dollinger-Bielschowsky Syndrome (Jansky-Bielschowsky Syndrome; Infantile Amaurotic Familial Idiocy [Late]; Infantile Ganglioside Lipidosis [Late]; Bielschowsky-Jansky Disease)  378

**General:** Late infantile form of neuronal ceroid lipofuscinosis; onset age 2 to 5 years; autosomal recessive; other neuronal ceroid lipofuscinoses include infantile (Santavuori-Haltia), juvenile (Spiel-Meyer-Sjögren), and adult (Kufs-Hallervorden).

**Ocular:** Optic nerve atrophy; macular pigmentation.

**Clinical:** Cerebroretinal degeneration; cerebellar ataxia; defective hearing; convulsions; spasticity; contractures; progressive mental deterioration.


Dominant Optic Atrophy Syndrome (Dominant Optic Atrophy, Deafness, Ptosis, Ophthalmoplegia, Dystaxia, and Myopathy)  379

**General:** Autosomal dominant disorder; ptosis, ophthalmoplegia, dystaxia, and nonspecific myopathy occur in midlife; optic atrophy and hearing loss occur in early life; autosomal dominant inheritance.

**Ocular:** Ptosis; ophthalmoplegia; progressive optic atrophy; abnormal electroretinogra-phy; diplopia; ocular myopathy; nystagmus; focal temporal excavation of optic disk; dyschromatopsia (blue-yellow); myopia; temporal pallor of the optic nerve.

**Clinical:** Sensorineural hearing loss; myopathy; dystaxia.


Donohue Syndrome (Leprechaunism)  380

**General:** Etiology unknown; possibly autosomal recessive; prevalent in females; present at birth; possible correlation between Donohue syndrome and insulin receptor gene defects.

**Ocular:** Hypertelorism.

**Clinical:** Failure to thrive; mental retardation; sexual precocity; hirsutism; broad nose; hypertrophic nipples; hypertrophy of external genitals; intrauterine growth retardation; extreme insulin resistance hyperkeratosis; precocious tooth eruption.


Dorsal Midbrain Syndrome  381

**General:** Caused by lesions of the posterior commissure located in the dorsal midbrain, pineal tumors, shunt malfunction, or hydrocephalus; less common causes include midbrain hemorrhage or infection, hypoxia, multiple sclerosis, trauma, lipid storage disease, Wilson disease, Whipple disease, syphilis, and tuberculosis.

**Ocular:** Loss of upward gaze; lid retraction; light-near dissociation; impaired convergence and divergence; convergence-retraction nystagmus.

**Clinical:** Patients tend to adopt abnormal head postures to fixate or maintain binocularity.


**Double Whammy Syndrome (Voluntary Propulsion of the Eyes)**

**General:** Ability to displace the globe forward actively while retracting the upper and lower lids behind the equator of the eyeball.

**Ocular:** Voluntary dislocation of either eye separately or of both simultaneously.


**Down Syndrome (Mongolism; Trisomy G; Trisomy 21 Syndrome; Mongoloid Idiocy)**

**General:** Trisomy of chromosome 21.

**Ocular:** Hypertelorism; epicanthus; blepharitis; ectropion; nystagmus; esotropia; high myopia (30%); hyperopia; color blindness; yellow spots on the iris; hypoplasia of the iris; blepharoconjunctivitis; lens opacities (50%); keratoconus (may be acute); corneal hydrops; corneal ectasia; corneal edema; leukoma; lateral displacement of canaliculi and puncta; megaloblepharon; euryblepharon; decreased accommodation; Leber congenital amaurosis.

**Clinical:** Mental retardation; skeletal abnormalities; overextension of joints; deformed and low-set ears; short fifth finger; transverse palmar crease; fissured tongue; heart anomalies.


**Doyne Honeycomb Choroiditis (Dominant Orbruch Membrane Drusen; Hutchinson-Tays Central Guttate Choroiditis; Holthouse-Batten Superficial Choroiditis; Malattia-Leventinese Syndrome)**

**General:** Autosomal dominant; represents early manifestation of senile macular degeneration; both sexes affected; onset in advanced age; patients present with drusen at an early age (second to third year of life) with near-normal visual acuity in childhood.

**Ocular:** Drusen with multiple yellow lesions becoming calcified and presenting crystalline appearance.

**Clinical:** None.


**Dracontiasis (Dracunculosis; Guinea Worm Infection; Draconculiasis)**

**General:** Caused by *Dracunculus medinensis*; affects connective and subcutaneous tissues.

**Ocular:** Conjunctivitis; proptosis; nematode present in the conjunctiva, eyelid, globe, and orbit.

**Clinical:** Itching; urticaria; small blisters; tetany; septicemia; arthritis; paraplegia; constrictive pericarditis; urogenital involvement.

Dragged-Fovea Diplopia Syndrome

**General:** Binocular double vision with macular conditions such as epiretinal membrane, choroidal neovascular membranes, localized retinal detachment or paramacular scars dragging the fovea.

**Ocular:** Maculopathy: metamorphopsia: vascular wrinkling; central diplopia in the presence of peripheral fusion.

**Clinical:** Lights on-off test with 20/70 to 20/100 is diagnostic.


Drummond Syndrome (Idiopathic Hypercalcemia; Blue Diaper Syndrome)

**General:** Autosomal recessive; manifests itself in infancy; defective intestinal transport of tryptophan, which oxidizes to indigo blue and stains the diaper blue.

**Ocular:** Sclerosis of optic foramina (occasionally); prominent epicanthal folds; nystagmus; strabismus; peripheral retinal atrophy; papilledema; optic atrophy; microcornea; hypoplasia of the optic nerve; abnormal eye movements.

**Clinical:** Dwarfism; osteosclerosis; craniofacial anomalies; depressed bridge of nose; "elfin-like" face; mental retardation; anorexia; vomiting; constipation.


Duane Syndrome (Retraction Syndrome; Stilling Syndrome; Turk-Stilling Syndrome)

**General:** Autosomal dominant; more frequent in females; manifestations in infancy; was thought to be secondary to fibrosis of the lateral rectus muscle or abnormal check ligaments; now established to be due to congenital aberrant innervation affecting third and seventh cranial nerves.

**Ocular:** Narrowing of palpebral fissure on adduction, widening on abduction; primary global retraction; deficiency of medial and lateral recti motility; limitation of abduction in affected eye usually is complete; retraction of the globe with attempted adduction varies from 1 to 10 mm; convergence insufficiency; heterochromia irides; left eye is more frequently involved.

**Clinical:** Associated Klippel-Feil syndrome; malformation of face, ears, and teeth.


Duane A. Congenital deficiency of abduction, associated with impairment of adduction, retraction movements, contractions of the palpebral fissure and oblique movements of the eye. *Arch Ophthalmol* 1905; 34:133.


Dubin-Johnson Syndrome (Chronic Idiopathic Jaundice; Rotor Syndrome)

**General:** Onset in infancy, but may be present at birth; autosomal dominant; liver cells are unable to excrete conjugated bilirubin; manifestations similar to Rotor syndrome, except that in the latter there is no melanin pigment present in the liver cells.

**Ocular:** Jaundice of sclera and conjunctiva in infancy.

**Clinical:** Abdominal pain right hypochondrium; nausea; vomiting; diarrhea; anorexia; weakness; hepatomegaly.


Dubowitz Syndrome (Dwarfism-Eczema-Peculiar Facies)

**General:** Affects both sexes; congenital; may be autosomal recessive inheritance.

**Ocular:** Hypertelorism; lateral telecanthus; palpebral ptosis; short palpebral tissues.
Clinical: Eczema; sparse hair; cleft palate; microcephaly; low birth weight; mild mental retardation; characteristic face; short stature; spontaneous keloids; intrauterine growth retardation.


Duck-Bill Lips and Ptosis

General: Autosomal dominant.

Ocular: Ptosis; strabismus; hypertelorism.

Clinical: Short philtrum; duck-bill lips; low-set ears; broad forehead; slightly anteverted nose and flat nasal bridge; slightly wide-spaced teeth and high-arched palate; slightly receding chin; slightly wide-set nipples; two phalanges in both fifth fingers; impaired speech.


Duplication 14q Syndrome

General: Chromosomal 14q duplication syndrome.

Ocular: Hypertelorism; sparse eyelashes and eyebrows; slanted palpebral fissures; ocular colobomata.

Clinical: Postnatal growth retardation; mental retardation; hypotonia; microcephaly; nasal dysmorphism; tented lip; micrognathia; posteriorly rotated ears; minor skeletal anomalies.


Dyschondroplasia Syndrome (Ollier Syndrome; Enchondromatosis)

General: Chondrodysplasia in which ossification in the epiphyseal region is delayed or absent, with resulting continuation of excessive hypertrophic cartilage formation; dyschondroplasia associated with hemangiomas is referred to as Maffucci syndrome (see Maffucci Syndrome).

Ocular: Narrowing of the optic foramen and supraorbital fissure; ophthalmoplegia; optic atrophy, retinal pigmentation.

Clinical: Joint deformities with functional disturbances; coxa vara or valga; scoliosis; facial asymmetry; unilateral bone involvement with resulting shortening of the extremity; intracranial gliomas; intracavernous chondrosarcoma; clival chondroma.


Eales Disease (Periphlebitis)

**General:** Common; young adults.

**Ocular:** Sheathing of peripheral veins; hemorrhage in new vessels and later retinal detachment; retinal vascular tortuosity; microaneurysms of retina; postneovascularization of vitreous; internuclear ophthalmoplegia.

**Clinical:** Epilepsy and hemiplegia have been reported; chronic encephalitis; ulcerative colitis; central nervous infarction.


East-West Syndrome

**General:** Caused when an intraocular lens is placed so that the edge of the optic, a positioning hole, and the components of the loop-optic junction are well within the papillary aperture.

**Ocular:** Glare; halos; monocular diplopia; irregular pupil.

**Clinical:** None.


Eaton-Lambert Syndrome (Myasthenic Syndrome; Ocular Myoclonus Syndrome; Myoclonic Syndrome)

**General:** Males; over 40 years of age; intrathoracic tumor; myasthenia-like condition less likely to have ocular manifestations; positive association with small cell cancer of the lung; underlying autoimmune basis.

**Ocular:** Decreased amplitude of version in all directions of gaze; ocular myoclonus; corneal abrasion; decreased corneal sensitivity; conjunctival injection; miotic pupil.

**Clinical:** Weakness; fatigue; peripheral paresthesia; dryness of mouth.


Eclampsia and Preeclampsia (Toxemia of Pregnancy; Preeclampsia)

**General:** Disorders of cells in glomeruli of kidneys that occur during gestation or shortly after delivery.

**Ocular:** Cortical blindness; nystagmus; mydriasis; absolute pupillary paralysis; ptosis; choroidal detachment; retinal detachment; cotton wool exudates; optic atrophy; retinal hemorrhages; petechial hemorrhages and focal edema in the occipital cortex.

**Clinical:** Hypertension; edema; proteinuria; convulsions; coma; death; cardiac failure; weight gain.

### Ectopia Lentis with Ectopia of Pupil (Ectopia Lentis et Pupillae)

**General:** Autosomal recessive.

**Ocular:** Lens and pupil displaced in opposite directions; bilateral cataracts; acute intermittent intraocular pressure crises; persistent pupillary membrane; poor pupillary dilation.

**Clinical:** None.


### Ectrodactyly, Ectodermal Dysplasia, Clefting Syndrome (EEC Syndrome)

**General:** Autosomal dominant; low penetrance; variable expressivity.

**Ocular:** Dacryocystitis; photophobia; corneal ulceration; blepharophimosis; atresia or absence of lacrimal puncta; strabismus; decreased visual acuity.

**Clinical:** Cleft lip; cleft palate; abnormalities of the urinary tract, hands, feet, and nail hypoplasia, granulomatous perlèche; scalp dermatitis.


### Ehlers-Danlos Syndrome (Fibrodysplasia Elastica Generalisata; Cutis Hyperelastica; Meeker-Ehlers-Danlos Syndrome; Indian Rubber Man Syndrome; Cutis Laxa)

**General:** Present at birth; autosomal dominant; two groups: (i) cutaneous and (ii) articular; syndrome is one of three primary disorders of elastic tissue (other two are pseudoxanthoma elasticum [Grönblad-Strandberg syndrome] and senile elastosis); inherited disorder of collagen biosynthesis.

**Ocular:** Hyperelasticity of palpebral skin; easy eversion of upper lid; ptosis; epicanthal folds; hypotony of extraocular muscles; strabismus; microcornea; thinning of cornea with keratoconus; thinning of sclera (blue sclera); subluxation of lens; angioid streaks; choriotirenal hemorrhages; retinitis proliferans with secondary detachment; macular degeneration; myopia; ruptured globe after minor trauma; limbus-to-limbus corneal thinning; acute hydrops; cornea plana; keratoglobus.

**Clinical:** Cutaneous manifestations include thin, atrophic, fragile skin, cutaneous hyperelasticity, and pseudomolluscoid tumors; articular manifestations include excessive articular laxity and luxations; hypermobile joints.


Cameron JA. Corneal abnormalities in Ehlers-Danlos syndrome type VI. *Cornea* 1993; 12:54-59.


18p- Syndrome (18p Deletion Syndrome)

**General:** Chromosome 18p deletion syndrome.

**Ocular:** Hypertelorism, epicanthus, horizontal palpebral fissures.

**Clinical:** Microcephaly, round face, broad-based nose, "carp mouth," microretrognathic; pterygium colli; dysplastic and low-set ears; clinodactyly; failure to grow; muscular hypotony; mental retardation; hypoplastic male genitalia.


18q- Syndrome (18q Deletion Syndrome)

**General:** Chromosome 18q deletion syndrome.

**Ocular:** Macular "fibrosis"; Optic disk abnormalities with tractional retinal detachment, retinal degeneration, and tilting of the optic disk.

**Clinical:** Microcephaly; short stature; hypotonia; hypothyroidism; diabetes mellitus; short neck; sensorineural hearing loss; sensorimotor axonal neuropathy; mild-to-moderate mental retardation; chronic arthritis; seizures.


Eldridge Syndrome

**General:** Autosomal recessive inheritance; also may involve an enzyme deficiency.

**Ocular:** Myopia, onset 4 to 6 years of age; increased retinal transluency; temporal crescents; mild electroretinographic abnormalities.

**Clinical:** Severe myopia; sensorineural hearing loss; low intelligence; mild renal disease.


Electrical Injury

**General:** Electric current passes through the body; voltage ranging from 100 to 200 million volts may cause electrical burns.

**Ocular:** Choroidal atrophy; corneal perforation; necrosis of cornea or lids; blefarospasm; anterior or posterior subcapsular cataracts and vacuoles; optic neuritis; optic nerve atrophy; retinal edema; retinal hemorrhage; pigmented degeneration; retinal holes; anterior uveitis; hyphema; hypotony; glaucoma; night blindness; nystagmus; paralysis of extraocular muscles; visual field defects; dilation of retinal veins.

**Clinical:** Skin burns; injury to cardiovascular, central nervous, and musculoskeletal systems; tissue necrosis; vascular injury.


11q- Syndrome

**General:** Chromosome 11q deletion syndrome.

**Ocular:** Telecanthus/hypertelorism; rarely, congenital glaucoma, cyclopia.

**Clinical:** Psychomotor retardation, trigonocephaly, broad depressed nasal bridge, micrognathia, low-set abnormal ears, cardiac anomalies, hand and foot anomalies, renal agenesis, anal atresia, supratentorial white matter abnormality on computed tomography or magnetic resonance imaging; microphallus; holoprosencephaly; female preponderance.
Ellis-Van Creveld Syndrome (Chondroectodermal Dysplasia)

**General:** Autosomal recessive inheritance; occurs in the Amish; associated with de novo chromosomal abnormality: deletion of 12 (p11.21p12.2).

**Ocular:** Esotropia; iris coloboma; congenital cataract.

**Clinical:** Bilateral polydactyly; short and plump limbs; genu valgum; talipes (equinovarus, calcaneovalgus); thoracic constriction; fusion of middle part of upper lip to maxillary gingival margin; dental anomalies: number, shape, spacing; congenital heart defect in about 50% of patients; dystrophic fingernails; genital anomalies; mild mental retardation; short stature; hypoplastic hair and skin; oligodontia; small thoracic cage; hypoplastic pelvis; cone-shaped epiphyses of hands.


Elschnig Syndrome (Elschnig Syndrome II)

**General:** Present from birth; etiology unknown.

**Ocular:** Elongation of lid fissure with downward displacement of the lateral angle; ectropion of lower lid.

**Clinical:** Cleft lip and palate may occur.


Elschnig Syndrome I (Meibomian Conjunctivitis)

**General:** Chronic inflammations; characteristic foamy secretion; benign.

**Ocular:** Conjunctivitis; foamy secretion; ocular irritation; photophobia; minimal visual impairment.

**Clinical:** Hyperplasia of tarsal glands.


Empty Sella Syndrome

**General:** Further progression of ocular findings and symptoms after treatment of pituitary tumors.

**Ocular:** Reduced visual acuity and possible blindness; hemianopsia; quadrantanopsia; irregular field defects; central scotoma; pale optic disks; central retinal vein occlusion.

**Clinical:** Acromegalic features and other general systemic manifestations depend on the type of the primary tumor the patient had and are not part of the "empty sella syndrome" that is responsible for progression of ocular pathologic condition.


Encephalitis, Acute

**General:** In approximately 0.1% to 0.2% of patients having rubeola (measles), an acute encephalitis is seen within 1 week after the onset of the rash; a case of immunosuppressive encephalitis can present with focal seizures leading to progressive obtundation.

**Ocular:** Papillitis; optic atrophy; ocular motor palsies; nystagmus; optic neuritis or neuroretinitis.
Clinical: Rise in temperature; drowsiness; irritability; meningismus; vomiting and headache; stupor; convulsions; coma.


Endocarditis 411

General: Common in people with arteriosclerosis; subacute form leads to pyemia; bacterial endocarditis without heart murmur seen in intravenous drug users.

Ocular: Retinal hemorrhages; conjunctival petechiae; choroiditis; Roth spots; spastic mydriasis; optic neuritis; central retinal artery occlusion; choroidal abscess, subretinal neovascularization.

Clinical: Heart murmur; fever; intracranial aneurysm; cerebral hemorrhage; subarachnoid hemorrhage.


Engelmann Syndrome (Osteopathia Hyperostotica [Scleroticans] Multiplex Infantilis; Diaphyseal Dysplasia; Camurati-Engelmann Disease; Hereditary Multiple Diaphyseal Sclerosis; Juvenile Paget Disease) 412

General: Etiology unknown; progressive resorption and deposits of bone with thickening of periosteum and changes of cortex as evident by diagnostic x-ray studies in the intermediate portion of the long bones.

Ocular: Exophthalmos; hypertelorism (secondary); ptosis; lagophthalmos; lateral rectus palsy; convergence insufficiency; epiphora; cataract; tortuous retinal vessels; papilledema; optic atrophy.

Clinical: Pain in extremities; poorly developed musculature; waddling gait; delayed ambulation; scaly skin; delayed dentition; deafness; hypogonadism; pain in both legs; aching in the forearms; episodic temporofrontal and occipital headache.


Enterobiasis (Oxyuriasis; Pinworm; Seatworm) 413

General: Intestinal infection caused by Enterobius vermicularis; worm's head attached to cecal mucosa, appendix, or parts of bowel; worms travel anal canal and deposit eggs on perianal skin; eggs infective for 10 to 20 days; airborne transmission; common in children; extraintestinal pinworm infection has been reported.

Ocular: Palpebral edema; blepharitis; keratoconjunctivitis; macular edema.

Clinical: Pruritus; eczema; pyogenic infection; vaginal discharge; chronic granulomatous salpingitis; endometritis.


Epiblepharon 414

General: Autosomal dominant; occurs predominantly in Chinese individuals.

Ocular: Epicanthus; epiblepharon of upper and lower lids.
Epicanthus

General: Autosomal dominant.

Ocular: Epicanthus; epiblepharon of upper and lower lids; ptosis.

Clinical: None.

Epidemic Keratoconjunctivitis

General: Highly communicable; adenovirus types 8 and 19; usually bilateral; epidemic keratoconjunctivitis has been reported worldwide associated with 11 virus serotypes, with serotypes 8, 11, and 19 being the most common responsible ones.

Ocular: Follicular or membranous conjunctivitis; chemosis; subconjunctival hemorrhages; corneal opacity; punctate epithelial keratitis; corneal ulcer; blepharospasm; lid edema; serous discharge; uveitis; epiphora.

Clinical: Submaxillary and cervical lymphadenopathy.

Epidermal Nevus Syndrome (Ichthyosis Hystrix)

General: One or a combination of the following epidermal nevi described as nevus unius lateris, ichthyosis hystrix, linear nevus sebaceous, or congenital acanthosis nigricans; autosomal dominant.

Ocular: Blepharoptosis and fibroma on bulbar conjunctiva; antimongoloid eyelid fissures; eyelid colobomata; horizontal and rotary nystagmus; esotropia; conjunctival tumors; corneal opacities; corectopia and colobomata of the iris.

Clinical: Somatic anomalies involving the skeletal and central nervous system; anomalies of bone formation; atrophy; ankylosis; vitamin D-resistant rickets; bone cysts; mental retardation; cortical atrophy; hydrocephalus; focal and grand mal epilepsy; cerebrovascular tumors; cortical blindness.

Epilepsy, Light-Sensitive

General: Autosomal dominant.

Ocular: Photic stimulation of 15 to 30 flashes per second, inducing epileptic seizure.

Clinical: Spastic paraparesis; mental retardation; light sensitivity.
Epiphyseal Dysplasia of Femoral Heads, Myopia, Deafness

**General:** Autosomal recessive.

**Ocular:** Severe myopia.

**Clinical:** Femoral epiphyseal dysplasia; deafness.


Epiphyseal Dysplasia, Microcephaly, and Nystagmus

**General:** Autosomal recessive.

**Ocular:** Nystagmus; retinitis pigmentosa.

**Clinical:** Epiphyseal dysplasia; microcephaly; short stature.


Epiphyseal Dysplasia, Multiple, with Myopia and Conductive Deafness

**General:** Autosomal dominant; onset early adulthood of severe osteoarthritis of hips; deficiency of distal tibial ossification seen in children results in sloping end of tibia in adulthood.

**Ocular:** Progressive myopia; retinal thinning; cataracts.

**Clinical:** Osteoarthritis of hips; short stature; brachydactyly, hyperextensibility of fingers; widening of joint spaces; conductive deafness.


Episkopi Blindness

**General:** Sex-linked; confined to male members of Greek Cypriot family group, most of whom live in Episkopi.

**Ocular:** Microphthalmia; corneal opacities; transverse corneal band; iritis; cataract; retrolental opacities; retinitis pigmentosa; Leber optic atrophy; amaurosis.

**Clinical:** None.


Epithelial Erosion Syndrome (Metaherpetic Keratitis; Kaufman Syndrome; Franceschetti Dystrophy; Posttraumatic Keratitis)

**General:** Most likely caused by herpes simplex virus; previous corneal trauma or autosomal dominant.
Ocular: Recurrent erosions of the corneal epithelium, usually seen within weeks or months after herpes simplex infection of the cornea; "loose" epithelium is removed from the underlying Bowman membrane mechanically by lid blinking; defects are irregular in shape and stain positively with fluorescein dye; underlying corneal stroma usually shows some edema; pain upon opening eyes in morning.

Clinical: Mild fever; occasionally herpetic skin lesions.


Erb-Goldflam Syndrome (Erb II Syndrome; Hoppe-Goldflam Disease; Pseudoparalytic Syndrome; Myasthenia Gravis)

General: Occurs at any age; more frequent between ages 20 and 40 years; more females affected than males; progressive; spontaneous; symptoms improve or resolve with rest in early stages of disease (see Myasthenia Gravis, Neonatal or Infantile); caused by autoantibodies against the acetylcholine receptor at the neuromuscular junction, leading to abnormal fatigability and weakness of skeletal muscle.

Ocular: Transient diplopia; ptosis of upper eyelids.

Clinical: Excessive fatigability of musculature; symptoms appear and increase as day progresses; expressionless face; sagging jaw; difficulty in chewing and talking; nasal regurgitation.


Erosion Syndrome

General: Caused by imperfect adherence of corneal epithelium due to abnormalities in basement membrane; abnormalities may be inherent, induced by trauma, or both; disease not vision threatening.

Ocular: Disabling episodes of pain; stabbing pain in the eye like that from a foreign body, most frequent on awakening; lesser forms of irritation; blurred or vision; drying associated with a stinging in the eyes; irregular astigmatism; corneal findings include anterior membrane dystrophies (epithelial and subepithelial dot, map, or fingerprint-type changes).

Clinical: None.


Erythema Nodosum (Dermatitis Contusiformis)

General: Young females; hypersensitive reaction secondary to viral, bacterial, and fungal infections; duration 2 to 4 weeks; recurrences possible.

Ocular: Subcutaneous nodules involving lids; keratitis; uveitis.

Clinical: Painful nodules on surface of thighs, arms, and face; fever; malaise; red lesions that progress to bruiselike and disappear in a few days to 3 weeks; cervical lymphadenopathy; exquisitely tender, erythematous nodules distributed symmetrically on the extensor surfaces of the lower extremities.

### Escherichia Coli

**General:** Gram-negative rod found in the gastrointestinal tract; urinary tract is the usual portal of entry.

**Ocular:** Uveitis; hyphema; hypopyon; gas bubbles in anterior chamber; purulent conjunctivitis; keratitis; corneal edema; panophthalmitis; endophthalmitis; glaucoma.

**Clinical:** Diarrhea; gastroenteritis; dehydration.


### Espildora-Luque Syndrome (Ophthalmic Sylvian Syndrome)

**General:** Embolism of the ophthalmic artery with refectory spasm of the middle cerebral artery.

**Ocular:** Unilateral blindness (caused by ophthalmic artery embolism).

**Clinical:** Temporary hemiplegia contralateral side of amaurosis (caused by reflex spasm of the middle cerebral artery).


### Ethan Syndrome, Primary

**General:** Congenital, esotropia, head turn, and nystagmus coexistent with nystagmus compensation and nystagmus blockage syndrome (see Nystagmus Compensation Syndrome; Nystagmus Blockage Syndrome).

**Ocular:** Esotropia; esophoria; nystagmus; ambylopia; orthophoria.

**Clinical:** Head turn and chin elevation to compensate for nystagmus.


### Ethan Syndrome, Secondary

**General:** Classic nystagmus blockage syndrome, but after strabismus surgery development of head turn with straight eyes and appearance of nystagmus compensation syndrome (see Nystagmus Blockage Syndrome; Nystagmus Compensation Syndrome).

**Ocular:** Esotropia; nystagmus; orthophoria; ambylopia; nystagmus increased in abduction.

**Clinical:** Abnormal head position.


### Ewing Sarcoma (Ewing Syndrome)

**General:** Highly metastatic round cell tumor of bone; most commonly involves long or trunk bones; metastasizes at high rate; usually occurs between ages 10 and 25 years; seen more frequently in males than in females.

**Ocular:** Exophthalmos; orbital hemorrhages; orbital necrosis; commonly found as the second malignancy in patients with hereditary retinoblastoma.

**Clinical:** Lytic bone destruction; pain; edema; slight fever.


### Exfoliation Syndrome (Capsular Exfoliation Syndrome)

**General:** Only in men older than 60 years.
**Ocular:** Iridodonesis; rubeosis iridis; cataract; phacodonesis; dislocated lens; corneal dystrophy; choroidal sclerosis; primary optic atrophy; lens capsule exfoliation; lower endothelial cell density.

**Clinical:** None.


**Extreme Hydrocephalus Syndrome (Kleblattschädel Syndrome; Cloverleaf Skull Syndrome; Hydrocephalus; Chondrodystrophicus Congenita)**

**General:** Secondary obstruction of cerebrospinal fluid circulation caused by some primary disease such as maternal rubella, Rh incompatibility, or hydramnion; Arnold-Chiari syndrome has similar associated findings; almost all affected children are born dead.

**Ocular:** Exophthalmos with downward placement and downward rotation of the globe; propulsion of globe; upper lid retraction and lower lids covering almost half of the downwardly rotated cornea; nystagmus; strabismus; exposure keratitis; optic nerve atrophy.

**Clinical:** Extreme hydrocephalus; low-set ears; thin and spastic extremities with digital anomalies; convulsions; spina bifida.


**Eyebrow Whorl**

**General:** Autosomal dominant.

**Ocular:** Whorl in the hair of the eyebrow; myopia, telecanthus, hypertelorism.

**Clinical:** Deafness, proteinuria.


**Fabry Disease (Angiokeratoma Corporis Diffusum Syndrome; Diffuse Angiokeratosis; Fabry-Anderson Syndrome; Glycosphingolipid Lipidosis; Glycosphingolipidosis)**

**General:** Lipoid storage disorder; X-linked recessive inheritance; lack of α-galactosidase A enzyme.

**Ocular:** Swelling of eyelids; varicosities of palpebral and bulbar conjunctiva; corneal dystrophy; corneal opacities; increased tortuosity of retinal vessels and aneurismal dilatations; cornea verticillata; cataract; central retinal artery occlusion; internuclear paralysis of extraocular muscles; papilledema; tortuosity and caliber irregularity of conjunctival vessels; characteristic cream-colored whorllike opacity in deep part of corneal epithelium; posterior cataract; occasional edema of optic disk and retina.

**Clinical:** Angiokeratoma of the skin with small, grouped papular lesions mainly over the scrotum, thighs, buttocks, sacral area, umbilical area, and lips; elevated blood pressure; disturbance in sweat secretion; pain in arms and legs; enlarged heart; albuminuria.


### Facio-Oculo-Acoustico-Renal Syndrome

**General:** Autosomal recessive.

**Ocular:** Congenital myopia; undeveloped filtration angle; persistent pupillary remnant membrane; hypertelorism; dysplasia carthorum; antimongoloid obliquity of palpebral fissure.

**Clinical:** Large head; sensorineural hearing loss; proteinuria; epiphyseal dysplasia of the femoral heads.


### Facio-Scapulo-Humeral Muscular Dystrophy (FSH Muscular Dystrophy)

**General:** Autosomal dominant disorder; onset varies from infancy to old age; severity varies from scarcely detectable to incapacitating; recessive inheritance has been reported.

**Ocular:** Retinal telangiectasis; macular lesion; macular edema; retinal sea fans.

**Clinical:** Deafness; wasting of shoulder girdle, upper deltoid, pectoralis, biceps, and triceps; difficulty whistling, drinking through straws, and playing wind instruments; foot dragging; mental retardation.


### Falciform Detachment

**General:** Autosomal dominant or recessive; perinatally acquired; characterized by ocular signs only; falciform detachment and congenital total detachment may alternate in affected siblings; falciform detachment and folds; retina projects as a wedge-shaped fold from the posterior pole of eye into the vitreous, occasionally as far anterior as the lens; less typical fold flattens and tapers out in the midperiphery of the retina.

**Ocular:** Falciform folds; retinal detachment; retrolental fibroplasia.

**Clinical:** None.


### Falciform Detachment with Microphthalmia and Microcephaly

**General:** Rare; both sexes affected; etiology unknown.
Ocular: Microphthalmia; congenital cataract; corneal opacities; falciform folds; glaucoma; buphthalmos; congenital detachment of retina; vitreous hemorrhages; persistent hyperplastic primary vitreous; retinal neovascularization; retinal dysplasia.

Clinical: Microcephaly; vestigial cerebellum; cleft palate; micrognathia; hydrocephalus.


Masuda Y. Two cases of ablation falciforms congenita and two other cases of ocular congenital anomalies, which appeared in a pedigree with consanguineous marriages. *J Clin Ophthalmol Tokyo* 1962; 16:325.


**Falciform Folds with Obesity, Nontoxic Goiter, Hypogenitalism, and Cryptorchidism**

**General:** Etiology unknown; isolated rare cases.

**Ocular:** Falciform folds; vitreous opacity.

**Clinical:** Obesity; nontoxic goiter; hypogenitalism; cryptorchidism.


**Familial Histiocytic Dermatoarthritis Syndrome**

**General:** Autosomal dominant; manifestations in childhood or adolescence; progressive.

**Ocular:** Glaucoma; bilateral uveitis; complicated cataract.

**Clinical:** Multiple histiocytic cutaneous nodules (face, ears, upper and lower extremities); subcutaneous plaques apparent on palpation and thickened, lichenified skin; arthropathy with symmetrical destructive arthritis mainly of hands and wrists but also observed on feet and elbows; possible hearing loss; cardiac and skeletal muscle failure; severe synovitis with arthritis mutilans.


**Familial Hypogonadism Syndrome**

**General:** Defect in testosterone biosynthesis.

**Ocular:** Progressive visual loss to complete blindness beginning shortly after birth; cataract; retinal degeneration.

**Clinical:** Partial deafness (neural type); obesity; shortness of stature; normal virilization.


**Familial Juvenile Nephronophthisis (Medullary Cystic Disease)**

**General:** Number of closely related renal disorders are associated with tapetoretinal degeneration; cause unknown.
**Ocular:** Retinitis pigmentosa; night blindness; progressive constriction of peripheral fields; retinal arterioles narrowed; yellow pigment deposits present throughout retina; macular degeneration.

**Clinical:** Renal disorders; polydipsia; polyuria.


**Familial Mediterranean Fever**

**General:** Recessive inherited polyserositis; progressive, fatal complications are renal failure and amyloidosis.

**Ocular:** Episcleritis; uveitis; colloid bodies; optic neuritis.

**Clinical:** Peritonitis; pleuritis; arthritis; fever; skin rash; renal failure; amyloidosis; recurrent attacks of fever and polyserositis of unknown origin.


**Fanconi Syndrome (Toni-Fanconi Syndrome; Amino Diabetes; Hypochloremic-Glycosuric Osteonephropathy Syndrome; De Toni-Fanconi Syndrome)**

**General:** Autosomal recessive inheritance; hematologic manifestations mainly in young patients; in adults the syndrome resembles milkman syndrome with disorder of calcium and phosphorus metabolism; chronic organic acidosis in Fanconi syndrome due to an inborn error of protein metabolism.

**Ocular:** Massive retinal hemorrhage may be present secondary to blood dyscrasias; bilateral anterior uveitis.

**Clinical:** Ecchymoses and mucus membrane hemorrhages; skin hyperpigmentation; osteomalacia; pseudo fractures; deformities of radius and absence of thumbs; hypophosphatemia.


Fanconi G. Die Nicht Diabetischen Glykosurien und Hyperglykamien des Alteren Kindes. *Jb Kinderheilkd* 1931; 133: 257.


**Fanconi-Turler Syndrome (Familial Ataxic Diplegia; Ataxic Diplegia)**

**General:** Aberration of the third cranial nerve (supranuclear type); ataxic diplegia is cerebellar ataxia with spastic pareses mainly of lower extremities; affects both sexes; onset at birth.

**Ocular:** Nystagmus; uncoordinated eye movements; dysmetria.

**Clinical:** Cerebellar ataxia; mental deficiency; spastic pareses.


**Farber Syndrome (Farber Lipogranulomatosis; Disseminated Lipogranulomatosis)**

**General:** Autosomal recessive inheritance; onset shortly after birth; rare; ceramidase deficiency.

**Ocular:** Parafoveal edema with mild cherry-red spot; grayness of the macula; diffuse fine pigmenitary changes in the fundus.

**Clinical:** Progressive hoarseness; swelling of extremities; nodular and granulomatous infiltrations of periarticular and subcutaneous tissue; mild lymphadenopathy; fever attacks; dysphonia and dyspnea; irritability; ceramidase deficiency associated with storage of ceramide in body tissues.

Fat Adherence Syndrome 448

**General:** Presence of a scar or adhesion that originates in extraconal fat and extends through Tenon capsule to attach to the muscle insertion or sclera; seen following retinal surgery.

**Ocular:** Persistent acquired restrictive strabismus after retinal surgery; diplopia.

**Clinical:** None.


Favre-Racouchot Syndrome (Nodular Elastoidosis) 449

**General:** Reaction to sun; permanent, slowly progressing condition; occurs in people chronically exposed to sun; usually apparent in fourth or fifth decade.

**Ocular:** Yellowish thickening of skin, with comedones and follicular cysts in and around orbit.

**Clinical:** Elastotic degeneration of skin; raised yellow patches and numerous comedones; orifices enlarged; citrine skin; cutis rhomboidalis nuchae; elastoma Dubreuilh.


Ritchie EB, Williams HN. Degenerative collagenous plaques of the hands. *Arch Dermatol* 1966; 93:202-203.

Feer Syndrome (Swift-Feer Syndrome; Infantile Acrodynia; Acrodynia; Pink Disease) 450

**General:** Etiology unknown, possibly allergic reaction to mercury or infection; onset in early childhood; both sexes equally affected.

**Ocular:** Proptosis; lacrimation; pronounced photophobia; severe conjunctival itching; conjunctival injection with occasional marked signs of inflammation; severe keratitis; mild optic neuritis; mydriasis.

**Clinical:** Restlessness; irritability; continuous profuse sweating; muscle hypotony; tachycardia; exanthema of palms and soles with exfoliation of large skin flaps; stomatitis; sleeplessness; cyanosis of fingers, toes, and nose; loss of teeth; rectal prolapse; muscle hypotonia; hypertension; hypertrichosis; gangrene of fingers.


Felty Syndrome (Chauffard-Still Syndrome; Primary Splenic Neutropenia with Arthritis; Rheumatoid Arthritis with Hypersplenism; Still-Chauffard Syndrome; Uveitis-Rheumatoid Arthritis Syndrome) 451

**General:** Etiology not fully understood, possibly infection or allergy; onset in middle-aged patients or children; prognosis poor; collagen disorder; occasionally can occur without articular disease.

**Ocular:** Decreased tear formation; scleromalacia perforans; keratoconjunctivitis; chronic anterior uveitis; scleritis; vitreous opacities; macular edema; choroidal inflammation; papillitis; keratitic precipitates; band-shaped keratopathy.

**Clinical:** Rheumatoid arthritis; splenomegaly; leukopenia; anemia (mild); oral lesion with ulcers and atrophy.

Crosby WH. What to treat in Felty's syndrome. *JAMA* 1973; 225:1114.


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Fetal Alcohol Syndrome

**General:** Dysgenesis in children born to alcoholic mothers; both sexes affected; onset from birth.

**Ocular:** Antimongoloid slant of lid fissures; lateral displacement of inner canthi; ptosis; epicanthus; strabismus; myopia; optic nerve hypoplasia; diffuse corneal clouding; iridocorneal abnormalities with central corneal edema; lens opacification; motility disorders.

**Clinical:** Growth retardation; delayed development (physical and intellectual); maxillary hypoplasia; micrognathia; large, low-set ears; abnormal motor function; irritability; microcephaly; cerebral nervous system dysfunctions; abnormal philtrum; flattened nasal bridge; cardiovascular defects; thin upper lip.

Tenbrinck MS, Buchin SY. Fetal alcohol syndrome. *JAMA* 1975; 232:1144.

Fetal Anticonvulsant Syndrome

**General:** Congenital after maternal ingestion of sodium valproate, carbamazepine and phenytoin.

**Ocular:** Myopia; astigmatism; strabismus; anisometropia; optic nerve hypoplasia

**Clinical:** Craniofacial dysmorphisms vary with type of abnormality


Fetal Hydantoin Syndrome

**General:** Syndrome due to an epoxide hydrolase 1, microsomal, arene oxide detoxification defect.

**Ocular:** Nystagmus.

**Clinical:** In vitro testing for the defect correlates most highly with congenital heart disease, cleft lip/palate, microcephaly, and major genitourinary, eye, and limb defects; hypersensitivity to phenytoin has occurred.


Fibrinoid Syndrome

**General:** From 2 to 14 days postvitrectomy, white-gray criss-cross layers of fibrin appear on the surface of the retina and immediately behind the plane of the iris; occurs only in patients with diabetes mellitus and usually in those requiring insulin; seen more frequently in people who have been diabetic for 15 years or more.

**Ocular:** Fibrin material interlaced on the surface of the retina and behind the iris; retinal detachment; neovascular glaucoma; rubeosis irides.

**Clinical:** Diabetes mellitus.

Fibrosarcoma

**General:** Malignant tumor of fibrous connective tissue; most commonly seen in persons 30 to 70 years old; frequent metastases to lung; true fibrosarcoma has a tendency to occur in children (better prognosis); most fibrosarcomas now would be classified as malignant fibrous histiocytomas.

**Ocular:** Paralysis of extraocular muscles; proptosis; orbital edema; erosion of orbital bony walls; increased intraorbital pressure; metastases to choroid/orbit.

**Clinical:** Tumors of mesenchymal soft tissues of the extremities, especially in the knee region; progressive pain; edema; tumors of the sinuses; tumors of the lungs.


Fish Odor Syndrome (Trimethylaminuria)

**General:** Metabolic syndrome characterized by a strong body odor of rotting fish; trimethylamine levels elevated; it appears that the enzyme flavin-containing monoxygenase is defective in this disorder; possibly autosomal recessive pattern of inheritance.

**Ocular:** Hypertelorism, cortical blindness.

**Clinical:** Hydrocephalus; mental retardation; unusual facies; short stature; skeletal abnormalities; cryptorchidism; hyperextensible skin.

Shelley ED, Shelley WB. The fish odor syndrome. *JAMA* 1984; 251:253-256.

Fisher Syndrome (Ophthalmoplegia Ataxia Areflexia Syndrome; Miller-Fisher Syndrome)

**General:** Acute idiopathic polyneuritis; prognosis good; complete recovery over several weeks (variant of Guillain-Barré Syndrome; see Guillain-Barré Syndrome).

**Ocular:** Moderate ptosis; complete external and almost complete internal ophthalmoplegia; diplopia; sluggish pupil reaction to light; may present without total ophthalmoplegia.

**Clinical:** Dizziness; severe ataxia; loss of tendon reflexes; chest pains; difficulties in chewing; diminished or absent sense of vibration; upper respiratory tract infection preceding this syndrome.


Fish-Eye Disease (Corneal Opacities- Dyslipoproteinemia)

**General:** Etiology unknown; rare; described in Swedish family; currently considered a unique dyslipoproteinemia.

**Ocular:** Visual impairment; marked corneal opacities.

**Clinical:** Very-low-density triglycerides and cholesterol raised.


Fleck Retina of Kandori Syndrome (Kandori Syndrome)

**General:** Possibly hereditary; onset young age; focal disturbance of the retinal pigment epithelium (RPE); affects both sexes; toxic causes also considered.

**Ocular:** Relatively large, irregular, yellowish flecks, sharply border-lined without pigmentation underneath retinal vessels and usually in the midperiphery; poor dark adaptation; normal photopic electroretinographic response; delay in generation of the scotopic response.
Clinical: None. 

**Floppy Eyelid Syndrome**

General: Origin unknown; more common in males; overweight; X-chromosome-linked inheritance pattern or possible hormonal influence; has been postulated that the degenerative changes in the tarsus may result from the combination of local pressure-induced lid ischemia and systemic hypoventilation.

Ocular: Easily everted, floppy upper eyelid and papillary conjunctivitis of the upper palpebral conjunctiva; upper eyelid everts during sleep, resulting in irritation, papillary conjunctivitis, and conjunctival keratinization; most distinct feature is rubbery, malleable upper tarsus; keratoconus; punctate keratopathy; blepharoptosis; lash ptosis.

Clinical: Obesity; sleep apnea.


**Floppy Iris Syndrome**

General: Flomax (tamsulosin) is causative agent during cataract. Flomax is commonly prescribed for benign prostate hypertrophy.

Ocular: Floppy iris

Clinical: Benign prostate hypertrophy; noted during cataract surgery

Gurbaxani A, Packard R: Intracameral phenylephrine to prevent floppy iris syndrome during cataract surgery in patients on tamsulosin 

**Flynn-Aird Syndrome**

General: May be basic hereditary enzyme deficiency, probably autosomal dominant; no sex predilection apparent.

Ocular: Severe myopia; bilateral cataracts; retinitis pigmentosa; total blindness; onset of visual difficulties in first or second decade of life.

Clinical: Hearing loss; joint stiffness; muscular wasting; kyphoscoliosis.


**Foix Syndrome (Cavernous Sinus Syndrome; Hypophysyal-Sphenoidal Syndrome; Cavernous Sinus Neuralgia Syndrome; Godtfredsen Syndrome; Cavernous Sinus-Nasopharyngeal Tumor Syndrome; Cavernous Sinus Thrombosis)**

General: Causes include tumor of lateral sinus wall or sphenoid bone, intracranial aneurysm, cavernous and lateral sinus thrombosis, or lesions; multiple myeloma; may result from infarctions or cancer or be idiopathic.

Ocular: Proptosis; severe ocular and periorbital pain; lid edema; paresis or paralysis of cranial nerves III, IV,V, and VI ; corneal anesthesia; optic atrophy.
Clinical: Postauricular edema; trigeminal neuralgia; deviation of the tongue toward paralyzed side; patients usually have prominent manifestations of sepsis and paranasal sinus; local skin infections are the most common cause.

Folling Syndrome (Phenylketonuria; Phenylpyruvic Oligophrenia; Ikiotia Phenylketonuria Syndrome) 465
General: Rare; autosomal recessive; phenylalanine cannot be converted to tyrosine; poor prognosis without early diet therapy; both sexes affected.
Ocular: Blue sclera; severe photophobia; corneal opacities; cataracts (controversial); partial ocular albinism; macular atrophy.
Clinical: Phenylketonuria; oligophrenia; partial albinism; muscle hypertonicity; hyper-reflexia of tendons; epilepsy; microcephaly; mousy odor of habitus; fair skin.

Foot-in-the-Wound Syndrome 466
General: Occurs when the anterior chamber intraocular lens haptic is within the wound.
Ocular: Haptic in the wound.
Clinical: None.

Foramen Lacerum Syndrome (Aneurysm of Internal Carotid Artery Syndrome) 467
General: Most commonly caused by congenital aneurysm involving the intradural portion of the carotid artery.
Ocular: Periorbital pain; ptosis; oculomotor paralysis with ptosis, diplopia, and internal opthalmoplegia; cranial nerves IV and VI may be involved; homonymous hemianopia (occasionally); loss of pupillary reflexes for light and accommodation; papilledema; optic atrophy.
Clinical: Meningism; mental disturbances; unilateral frontal or orbital headache; migraine attacks.

Forsius-Eriksson Syndrome (Aland Disease) 468
General: Associated with the natives of the Aland Islands; sex-linked inheritance; consanguinity versus mutant gene; affects males only; it has been considered a variety of incomplete congenital stationary night blindness.
Ocular: Microphthalmos; irregular latent nystagmus; myopia; astigmatism; dyschromatopsia; tapetoretinal degeneration; primary foveal hypoplasia or dysplasia; nystagmus.
Clinical: Prematurity; impaired hearing; mental retardation; epilepsy.
Foster Kennedy Syndrome (Basal-Frontal Syndrome; Gowers-Paton-Kennedy Syndrome) 469

General: Caused by tumor in base of frontal lobe or sphenoidal meningioma.

Ocular: Central scotoma may be present on side of optic atrophy; enlarged blind spot and peripheral contraction of field (opposite eye); homolateral descending optic atrophy due to compression of the ipsilateral optic nerve at the optic foramen; contralateral papilledema due to increased intracranial pressure; ipsilateral proptosis.

Clinical: Anosmia; headache; dizziness; vomiting; memory loss; psychic changes; also may be caused by an olfactory groove tumor (usually a meningioma) or pituitary adenoma.


4a Syndrome (Adrenocortical insufficiency associated with achalasia, alacrima, autonomic and other neurological abnormalities) 470

General: Associated with autonomic and other neurologic abnormalities

Ocular: Alacrima

Clinical: Adreno-cortical insufficiency; achalasia


4q- Syndrome (4q Deletion Syndrome) 471

General: Chromosome 4q deletion syndrome.

Ocular: Hypertelorism, epicanthal folds.

Clinical: Depressed nasal bridge; short nasal septum with upturned nose, cleft lip and palate; micrognathia; low-set malformed ears; short neck; distally placed nipples; sacral dimple; hypospadias; dysplastic nails; overriding toes; simian creases; hypoplasia of gall bladder; cardiac defects; mental retardation.


Foveal Hypoplasia and Presenile Cataract Syndrome (O'Donnell-Pappas Syndrome) 472

General: Autosomal dominant.

Ocular: Foveal hypoplasia; nystagmus; presenile cataract; peripheral corneal pannus.

Clinical: None.


Foville Syndrome (Foville Peduncular Syndrome) 473

General: Pontine area tumor, hemorrhage, tuberculoma, multiple sclerosis, or unilateral obstruction of paramedian branches may cause clinical manifestations.
Ocular: Paralysis of cranial nerve VI; paralysis of conjugate movement to the side of the lesion; abduction or horizontal gaze deficit.

Clinical: Peripheral facial palsy; contralateral hemiplegia; headache; ipsilateral: facial weakness, loss of taste, facial analgesia, Homer syndrome, and deafness.

Fragile X Syndrome

General: X-linked recessive; primarily affects males.

Ocular: Strabismus; nystagmus; high myopia; adult-onset glaucoma; blepharospasm; congenital optic atrophy; hyperopia; astigmatism; cataract; ptosis; corneal dystrophy.

Clinical: Mental retardation; dysmorphism; epilepsy; macroorchidism.

Franceschetti Disease (Fundus Flavimaculatus)

General: Affects both sexes; onset between ages 10 and 25 years; autosomal recessive; genetic linkage analysis has assigned the disease locus to chromosome 1p21-p13.

Ocular: Irregular yellowish deposit in and around the macula lutea forming a garland; impaired central vision with intact peripheral retinal function; bilateral retinal dystrophy; progressive subretinal fibrosis; chorioretinal punched-out spots in the posterior pole and midperiphery of the retina.

Clinical: None.

Franceschetti Syndrome (Franceschetti-Zwahlen-Klein Syndrome; Treacher Collins Syndrome; Mandibulofacial Dysostosis; Mandibulofacial Syndrome; Eyelid-Malar-Mandible Syndrome; Oculovertebral Syndrome; Berry Syndrome; Franceschetti-Zwahlen Syndrome; Zwahlen Syndrome; Bilateral Facial Agenesia; Berry-Franceschetti-Klein Syndrome; Franceschetti-Klein Syndrome; Franceschetti Syndrome (II); Treacher Collins-Franceschetti Syndrome; Weyers-Thier Syndrome)

General: Irregular dominant inheritance; Weyers-Thier syndrome has similar features, except it is a unilateral variant; prevalent in Caucasians.

Ocular: Microphthalmia; oblique position of eyes with lateral downward slope of palpebral fissures; temporal lower lid coloboma; lack of cilia on middle third of lower lid; iris coloboma; underdeveloped orbicularis oculi muscle; cataract; optic disk hypoplasia.

Clinical: Fishlike face with sunken cheek bones, receding chin, and large, wide mouth; absent or malformed external ears with auricular appendages; high palate and possible harelip; hypoplastic zygomatic arch with absence of normal malar eminences; prolonged hairline on the cheek; deafness; micrognathia; glossophtosis; cleft palate.


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**Franceschetti Syndrome (I)**

**General:** Inborn disease occurring at birth; etiology unknown.

**Ocular:** Deep punctate dystrophy of cornea with ichthyosis dystrophia (punctiformis profunda corneae with congenital ichthyosis).

**Clinical:** Dry, scaly skin; follicular hyperkeratotic lesions; atopic dermatitis; pruritus.


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**Franceschetti-Thier Syndrome**

**General:** Autosomal recessive inheritance.

**Ocular:** Corneal dystrophy.

**Clinical:** Multiple lipomas; mental retardation.


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**Francois (1) Dystrophy (Francois-Neetens Syndrome; Central Cloudy Dystrophy; Cloudy Central Corneal Dystrophy)**

**General:** Autosomal dominant; etiology unknown; not progressive; isolated keratocytes contain elevated amounts of glycosaminoglycans and lipids.

**Ocular:** Bilateral dystrophy of central third of cornea; snowflake patches covering the pupil; lesions show no definite structure or limits; more dense near Descemet membrane and becoming less toward the anterior surface toward the periphery; associated with central cloudy dystrophy; keratoconus; limbal dermoid; pseudoxanthoma elasticum; lenticular opacities; reduced corneal sensation.


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**Francois (2) Dystrophy (Francois-Evens Syndrome; Speckled Corneal Dystrophy)**

**General:** Etiology unknown; congenital; nonprogressive; autosomal dominant, but sporadic cases have been reported.

**Ocular:** Corneal dystrophy characterized by minute punctate opacities found in all layers of the cornea; varies in size, form, and degree of opacity but is identical in both eyes; anterior limiting membrane is always intact.


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**Francois Syndrome (2) (Dystrophia Dermachondrocornealis Familiaris)**

**General:** Autosomal recessive.
**Ocular:** Central superficial corneal dystrophy with subepithelial opacities.

**Clinical:** Distal osteochondral dystrophy of the extremities; cutaneous xanthomas.


**Frankl-Hochwart Syndrome (Pineal-Neurologic-Ophthalmic Syndrome) 482**

**General:** Pineal tumor, usually in early adulthood; poor prognosis.

**Ocular:** Limitation of upward gaze; concentric field constriction; papilledema; lack of pupillary reaction; nystagmus.

**Clinical:** Bilateral deafness; ataxia; weakness; headache; vomiting; polydipsia; polyphagia; convulsions; facial paralysis; tremor; Romberg sign; hypertonia; tendon hyperreflexia; Babinski sign.


**Freeman-Sheldon Syndrome (Cranio-Carpo-Tarsal Dysplasia; Whistling Face Syndrome) 483**

**General:** Rare; autosomal dominant and recessive inheritance as well as sporadic cases (genetic heterogeneity).

**Ocular:** Eyes deeply sunken (enophthalmos); hypertelorism; blepharophimosis; ptosis; antimongoloid slanting of lid fissures; esotropia.

**Clinical:** Small nose with narrow nostrils and long philtrum; alae nasi often bent, simulating colobomas; nasolabial folds present only near the nose; microstomia; high-arched palate and small mandible; flexion contractures of fingers; excessive bulging of central part of cheeks when whistling.


**Frenkel Syndrome (Ocular Contusion Syndrome; Anterior Segment Traumatic Syndrome) 484**

**General:** Minor blunt trauma to the anterior segment of the globe.

**Ocular:** Sluggish pupil reaction; traumatic mydriasis; iris dialysis; heavy pigment deposits on the vitreous surface; subluxation of the lens; transient posterior cortical lens opacities; permanent anterior or posterior capsular opacities; coronary opacities; late anterior cortical rosette; late total traumatic cataract; Vossius ring following hyphema; peripheral pigment disturbance resembling atypical retinitis pigmentosa; macular edema; retinal detachment.

**Clinical:** None.


**Friedreich Ataxia (Spinocerebellar Ataxia) 485**

**General:** Etiology unknown, either autosomal recessive or dominant; progressive; incapacitating by age 20 years; death from secondary diseases or cardiac failure; prevalent in males.

**Ocular:** Nystagmus; optic atrophy; there is a form of Friedreich ataxia associated with congenital glaucoma.
**Clinical:** Kyphoscoliosis; tremor; dysmetria; asynergia; slow atactic speech; paresthesias; Babinski sign; headache; retarded growth; mental retardation; polyuria; polydipsia; deformity of feet (onset in first year of life); clumsy gait and difficult to turn arms, head, and trunk; deafness.


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**Fröhlich Syndrome (Dystrophia Adiposogenitalis)**

**General:** Caused by chromophobe adenoma of pituitary, Rathke pouch tumors; craniopharyngiomas; suprasellar tumors; encephalitis; trauma; more frequent in Jewish families; manifestations occur in childhood, often during puberty.

**Ocular:** Bitemporal hemianopsia; impaired scotopic vision; papilledema; optic nerve atrophy (with increased intracranial pressure).

**Clinical:** Adiposity; genital hypoplasia; in females, menstruation fails to appear or may cease in postpubertal period; in males, voice remains high-pitched, undescended testes, absent facial hair, and feminine pubic line; possible retarded growth; polyuria; polydipsia.


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**Frontometaphyseal Dysplasia (FMD)**

**General:** Sex-linked; rare; bony dysplasia.

**Ocular:** Strabismus; supraorbital deformity; hyperopia; hypertelorism; prominent supraorbital ridges.

**Clinical:** Agenesis of frontal sinuses; underdevelopment of mandible; metaphysis of tubular bones; deafness; hirsutism; teeth abnormalities; bony overgrowth at base of nose resulting in nasal obstruction and mouth breathing; facial nerve paralysis; normal intelligence.


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**Frontonasal Dysplasia Syndrome (Median Cleft Face Syndrome)**

**General:** Congenital disorder without genetic background; condition may present a variety of facial malformations, depending on the stage of embryonic development at which interference occurs.

**Ocular:** Hypertelorism; anophthalmia or microphthalmia; significant refractive errors; strabismus; nystagmus; eyelid ptosis; optic nerve hypoplasia; optic nerve colobomas; cataract; corneal dermoid; inflammatory retinopathy.

**Clinical:** Broad nasal root may be associated with median nasal groove and cleft of nose and/or upper lip; cleft of ala nasi (unilateral or bilateral); V-shaped hair prolongation into forehead.


Fuchs (1) Syndrome (Heterochromic Cyclitis Syndrome)  489

**General:** Etiology unknown; mild infective cyclitis is the most likely cause; etiology remains unclear, although it is likely to be autoimmune; positive epidemiologic association with ocular toxoplasmosis has been investigated.

**Ocular:** Secondary glaucoma; unilateral hypochromic heterochromia; painless cyclitis with absence of synechiae and little or no ciliary injection; secondary cataract; vitreous opacities; small white discrete keratic precipitates with fine filaments between the precipitates; corneal epithelium may be slightly edematous; peripheral choroiditis occasionally; keratoconus.

**Clinical:** Occasional dysraphia of the cervical cord.


Fuchs-Lyell Syndrome (Debré-Lamy-Lyell Syndrome; Toxic Epidermal Necrolysis)  490

**General:** Allergic reaction with severe manifestations; similar to Fuchs-Salzmann-Terrien syndrome (see Fuchs-Salzmann-Terrien Syndrome); may result as a reaction to *Staphylococcus aureus* toxin in children or associated with certain medications, including penicillin, sulfa, nonsteroidal antiinflammatory agents, and allopurinol.

**Ocular:** Obstruction of nasolacrimal duct; cicatricial changes in conjunctiva and cornea; conjunctivitis; symblepharon; corneal ulceration and possible perforation.

**Clinical:** Inflammation of mucous membrane with ulcerations; general epidermolysis; cicatricial changes, especially of orifices.


Fuchs-Salzmann-Terrien Syndrome  491

**General:** Features similar to those of Fuchs-Lyell syndrome; both are based on drug allergies (antibiotics, sulfonamides, arsenic preparations) (see Fuchs-Lyell Syndrome).

**Ocular:** Features of Salzmann nodular dystrophy; superficial punctate keratitis; marginal degeneration of the cornea; intraocular hemorrhages; choroidal hemorrhages.

**Clinical:** Allergic cutaneous lesions with erythema and degrees of exfoliative dermatitis.


Fusobacterium  492

**General:** Gram-negative; normal inhabitant of mouth and respiratory, intestinal, and urogenital tracts; usually secondary to an underlying disease, surgical procedure, or therapy that impairs the defense of the host; non-spore-forming, nonmotile Gram-negative anaerobic bacilli.

**Ocular:** Conjunctivitis; dacryocystitis; orbital abscess; orbital cellulitis; corneal ulcer; tenonitis; lid edema; panophthalmitis; gangrene of conjunctiva; cavernous sinus thrombosis; cranial nerve palsy.

**Clinical:** Brain abscesses; pneumonia; liver abscess; endocarditis; sepsis; tissue necrosis.


G Syndrome (Hypertelorism Esophageal Abnormality and Hypospadias; Hypospadias-Dysphagia Syndrome)  493

**General:** Neuromuscular defect; autosomal dominant; prevalent in males; males more severely affected (see BBB Syndrome).

**Ocular:** Retinitis pigmentosa; hypertelorism; narrow palpebral fissures; epicanthal folds; telecanthus.

**Clinical:** Defect of esophagus; hoarseness; hypospadias; cryptorchidism; imperforate anus; defect of lingual frenulum; deafness; mild mental retardation; dysphagia; anosmia; swallowing difficulties; nasal bridge broad and flat; stridor; aspiration; prominent forehead; cleft lip and palate; laryngotracheal esophageal clefts.


Gaisbock Syndrome (Emotional Polycythemia; Stress Erythrocytosis)  494

**General:** Prevalent in men, heavy smokers; associated with emotional tension and stress.

**Ocular:** Afferent pupillary defect; conjunctivitis; central retinal vein occlusion; cystoid macular edema; peripapillary retinal hemorrhage; glaucoma.

**Clinical:** Obesity; hypertension; stress; vascular disease; plethora.


Galactosyl Ceramide Lipidosis (Krabbe [1] Syndrome; Infantile Globoid Cell Leukodystrophy; Krabbe Disease; Globoid Cell Leukodystrophy)  495

**General:** Defect in metabolism of galactocerebroside; genetically determined demyelinating disease that is fatal in early childhood; both sexes affected; onset usually in first year of life; ambiguous onset; autosomal recessive; onset at age 4 to 6 months, although some late-onset cases have been reported; diagnosis is made after identification of “globoid cells” in brain tissue.

**Ocular:** Photophobia; cortical blindness; optic atrophy; nystagmus.

**Clinical:** Hypersensitivity to external stimuli; rigidity; vomiting; seizures; episodic fever; mental retardation; death.


Gangliosidosis GMI Type 1 (Generalized Gangliosidosis [Infantile]; Norman-Landing Syndrome; Pseudo-Hurler Lipidosis)  496

**General:** Absence of A, B, and C isoenzymes of β-galactosidase visceral tissue and mucopolysaccharides in visceral tissues; both sexes affected; autosomal recessive; onset from birth; death from age 6 months to 2 years; defect has been localized to chromosome 3 (3p12-3p13).
Ocular: Macular cherry-red spots; optic disk pallor; nystagmus; esotropia; corneal clouding; retinal artery tortuosity and narrowing; retinitis pigmentosa; macular cherry-red spot found in 50% of patients with this disorder.

Clinical: Cerebral degeneration combined with visceromegaly and skeletal dysplasia; mental and motor retardation; seizures; deafness; spastic quadriplegia; feeding difficulties; recurrent bronchopneumonia; broad nose; frontal bossing; prominent maxilla; hepatosplenomegaly.


Gangliosidosis GM1 Type 2 (Juvenile Gangliosidosis) 497

General: Absence of Band C isoenzymes of β-galactosidase results in neural and visceral deposition of gangliosides and visceral deposition of mucopolysaccharides; autosomal recessive; defective hexosaminidase A; defect localized to chromosome 15 (15q22-15q25.1).

Ocular: Optic atrophy; pigmented retinopathy; strabismus; macular cherry-red spot; late optic atrophy.

Clinical: Cerebral degeneration; skeletal changes; visceromegaly; psychomotor deterioration; death between ages 3 and 4 years; abnormal acousticomotor reaction; hypotonia; variable hepatosplenomegaly; abnormal hexosaminidases A and B; defect localized to chromosome 5(5q13).


Ganser Syndrome (Pseudodementia; Nonsense Syndrome; Prison Psychosis Syndrome) 498

General: Found in prisoners and patients with schizophrenia; disparity between the person’s complaints and mental alertness.

Ocular: Patient pretends that he or she cannot see or read; no objective findings on examination of visual function.

Clinical: Patient pretends not to know how to do simple things previously familiar to him or her (not to know his or her age, how to spell or read, etc.); mild degree of mental deficiency; amnesia; analgesia; confusion; lethargy; apathetic indifference; headache.


Gansslen Syndrome (Familial Hemolytic Icterus; Hematologic-Metabolic Bone Disorder) 499

General: Autosomal dominant inheritance; occurs mainly in Caucasians.

Ocular: Hypertelorism; microphthalmos; epicanthus; narrowing of palpebral fissure; lid hemorrhages; myopia; dyschromatopias; hypochromic heterochromia; scleral icterus; conjunctival hemorrhages; retinal pallor and edema in advanced stages; dilated retinal arteries and veins; round retinal hemorrhages in deeper retinal layers; retinal exudates and macular star.

Clinical: Splenomegaly; hemolytic crises; dental deformities; brachydactyly; polydactyly; congenital hip luxation; oxycephaly; deformities of the outer ear and otosclerosis.


GAPO Syndrome (Growth Retardation, Alopecia, Pseudoanodontia, Optic Atrophy Syndrome) 500

General: Autosomal recessive.

Ocular: Progressive optic atrophy; glaucoma; keratoconus.

Clinical: Growth retardation; alopecia; pseudoanodontia; frontal bossing; high forehead; midfacial hypoplasia; wide-open anterior fontanelle; retarded bone age; premature aged appearance; hypogonadism; hepatomegaly; muscular body build.


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**Good Acuity Plus Photosensitivity (GAPP). Track Related Iridocyclitis and Scleritis (TRISC), Transient Light Sensitivity (TLS)**

**General:** Use of Intralase to perform LASIK

**Ocular:** Photophobia; glare; uveitis; iridocyclitis; scleritis

**Clinical:** Associated with refractive surgery and the use of Intralase technology; starts 6 to 8 weeks postoperatively and resolves by four to five months.

Binder P. New ocular syndrome surfaces with Intralase. *Ophthal News* 2004; Aug 12-14

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**Garcin Syndrome (Half-Base Syndrome; Schmincke Tumor-Unilateral Cranial Paralysis)**

**General:** Causes include tumors of nasopharynx, rapidly progressing growth of a sarcoma of base of skull, meningitis, and cranial polyneuritis; cranial nerves VIII to XII are most frequently involved.

**Ocular:** Ptosis (unilateral); unilateral external ophthalmoplegia; papilledema.

**Clinical:** Difficulties in swallowing; impairment of speech; hearing defect; respiratory difficulties; sensory disturbances; hoarseness.


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**Gardner Syndrome**

**General:** Autosomal dominant; both sexes affected; average onset age 20 years.

**Ocular:** Exophthalmos; congenital hypertrophy of RPE; multiple lesions of the eye; bilateral occurrence; orbital osteoma; highly pleomorphic pigmentation; unilateral or bilateral retinal lesions; pilomatrixoma-like epidermal cysts; presence of pigmented fundus lesions appears to cluster within families.

**Clinical:** Intestinal polyps; dermoid tumors; neurofibrosis osteomatosis; colon cancer; supernumerary teeth.


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**Gastrocutaneous Syndrome**

**General:** Combination of symptoms including peptic ulcer, hiatal hernia, multiple lentigines, cafe-au-lait spots, hypertelorism, myopia, acute or chronic; occurs in people with stress who smoke and use ulcerogenic drugs.

**Ocular:** Hypertelorism; myopia; nystagmus.

**Clinical:** Upper abdomen pain 1 to 2 hours after eating; pain at night, when gastric secretion is at its peak; nausea; vomiting; excessive salivation.
Gaucher Syndrome (Glucocerebroside Storage Disease; Glucosyl Ceramide Lipidosis; Cerebroside Lipidosis)

**General:** Storage of glucocerebroside in the reticuloendothelial system; autosomal recessive; occurs frequently in Jewish families; onset at any age; onset usually sudden in the infantile form; disease belongs to group of lipid storage disturbances such as ganglioside (Tay-Sachs), sphingomyelin (Niemann-Pick), and ceramide trihexoside (Fabry) (see Tay-Sachs Syndrome; Niemann-Pick Syndrome; Fabry Syndrome); caused by glucosylceramide β-glucosidase (glucocerebrosidase) deficiency; psychomotor deterioration apparent before age 6 months.

**Ocular:** Strabismus; brown-yellowish, wedge-shaped pinguecula; corneal clouding; oculomotor paralysis; gaze palsies.

**Clinical:** Infantile form: generalized hypertonia, opisthotonus, dysphagia, vomiting, laryngeal spasm, dyspnea; chronic form: hepatosplenomegaly, lymphadenopathy, mild-to-moderate anemia, yellowish-brown patchy skin pigmentation.


Gelineau Syndrome (Narcoleptic Syndrome)

**General:** Etiology not well understood; causes include subthalamic lesions, multiple sclerosis, and tumor of third ventricle; onset in adolescence or early adulthood; male preponderance (6:1); most characteristic sign is sudden attack of sleep that cannot be resisted and may last from only a few minutes to half an hour.

**Ocular:** Transient blurred vision; inability to read between attacks; bilateral diplopia; spontaneous eyelid closing; flickering vision.

**Clinical:** Diurnal attacks of short episodes of uncontrollable sleep (usually several times a day); cataplexy with decreased or absent muscle tone and paralysis caused by an upset emotional state; hallucinations.


General Fibrosis Syndrome (Congenital Enophthalmos with Ocular Muscle Fibrosis and Ptosis; Congenital Fibrosis of the Inferior Rectus with Ptosis; Strabismus Fixus; Vertical Retraction Syndrome (Congenital Fibrosis Syndrome )

**General:** Present from birth; familial history; apparent autosomal dominant transmission; sex-linked recessive transmission also reported.

**Ocular:** Ptosis; enophthalmos; disk hypoplasia; astigmatism; esotropia; exotropia; hypotropia; nystagmus; visual loss; positive forced duction test; may be associated with Marcus Gunn jaw-winking and synergistic divergence in attempted right gaze.

**Clinical:** None.


Gerlier Disease (Kubisagari; Le Tourniquet; Paralytic Vertigo)  
**General:** Cranial nerve dysfunction suggests a basal lesion such as epidemic paralyzed vertigo; infective agent suspected, small Gram-negative coccus from spinal fluid; contact with cows or horses in warm summer months in contaminated stables; pathology unknown; possible brainstem lesion.

**Ocular:** During attack: hyperemia of fundus oculi, fundus normal, and eyesight normal; between attacks: ptosis, dimness of vision, vertigo, and diplopia.

**Clinical:** Attack of palsies precipitated by severe exertion, bright light, warmth, hunger, or looking at a moving object (optokinetic irritation); attacks last approximately 10 minutes and may follow each other at very short intervals; mild form not incapacitating; severe form incapacitating; pains in back of neck; nodding of head during attack; hyperreflexia at intervals; attack of temporary palsy muscles, levator palpebrae superior, muscles of back of neck, extension limbs, face, pharynx, and larynx.


German Syndrome (Fetal Trimethadione; Tridione)  
**General:** Both sexes affected; prenatal onset; occurs when trimethadione or paramethadione is taken during pregnancy.

**Ocular:** Eyebrows up-slanted.

**Clinical:** Mild brachycephaly; short upturned nose; prominent forehead; cleft lip and palate; micrognathia; ear abnormalities; tetralogy of Fallot; genital abnormalities; hypospadias; hypertrophic clitoris; abnormalities of skin, gastrointestinal, renal, and skeletal systems.


Gerstmann Syndrome (Dominant Hemisphere Syndrome; Left Angular Gyrus Syndrome)  
**General:** Most frequently caused by tumors of dominant hemisphere in which the angular gyrus is involved; may follow an ischemic posterior cerebellar artery (PCA) lesion affecting the inferior parietal lobule and resulting in jargon aphasia, dyscalculia, agraphia right-left confusion, constructional dyspraxia, and alexia.

**Ocular:** Homonymous hemianopsia; visual agnosia for colors.

**Clinical:** Agraphia (inability to write); finger agnosia; anosognosia; aciculae (difficulties in or inability to perform serial number projects); confusion (mainly between left and right).


Giant Fornix Syndrome  
**General:** Enlarged upper fornix with buildup of mucopurulent debri and persistant discharge.

**Ocular:** Bacterial conjunctivitis; blepharospasm; enlarged fornix; secondary ptosis

**Clinical:** Recurrent chronic conjunctivitis


Giant Papillary Conjunctivitis Syndrome  
**General:** Commonly associated with contact lenses (hard and soft), foreign bodies, and ocular prosthesis; immunologic in origin.

**Ocular:** Ocular irritation; itching of the eye; decreased visual acuity; increased mucous production; papillary changes of the upper tarsal conjunctiva; contact lens coatings; may appear after a lens change from one style to another or by replacement of
the previous design; aging of a lens, particularly of a soft contact lens, may be associated; usually bilateral, although it can be markedly asymmetrical.


**Giardiasis**

**General:** Multiflagellate protozoan; parasite of human duodenum; encystation occurs in transit through colon.

**Ocular:** Uveitis; retinal hemorrhages; central serous retinopathy; palpebral edema.

**Clinical:** Nausea; flatulence; epigastric pain; abdominal cramps; diarrhea; weight loss; malabsorption.


**Gillum-Anderson Syndrome**

**General:** Genetic defect responsible for weakness in the orbital connective tissue; proposed connective tissue defect of sclera, zonules, and levator aponeurosis.

**Ocular:** Dislocated lenses; high myopia; bilateral ptosis typical for levator disinsertions; ectopia lentis.


**Gitelman Syndrome**

**General:** Autosomal recessive, renal tubulopathy characterized by hypokalemia, hypomagnesemia, metabolic alkalosis, hypocalciuria, and hypernatremia.

**Ocular:** Sclerochoroidal calcification.

**Clinical:** Patients may have arthralgias, seizures, episodes of tetany, muscular weakness, or paresthesia; also may be asymptomatic.


**Glander Syndrome**

**General:** Serious infection caused by *Malleomyces mallei*; no naturally acquired infections in United States since 1938; transmission from equine animals to man; caused by traumatic inoculations or inhalations; either acute, gangrenous, or chronic ulcerative; fatal usually in 7 to 10 days.

**Ocular:** Conjunctivitis; dacryocystitis; ulcerating granulomatous orbital lesions; photophobia; lacrimation.

**Clinical:** Systemic erythematous pustules; inhalation infection; fever; rigors; generalized myalgia lymphangitis; fatigue, headaches; pleuritic chest pain; diarrhea; lymphadenopathy; splenomegaly; mild leukocytosis; multiple subcutaneous and intramuscular abscesses (often arms and legs); visceral involvement, including pulmonary, pleural, skeletal, hepatic, splenic, meningeal, and intracranial.
Glaucoma, Congenital

**General:** Autosomal recessive; occurs more frequently in males; can occur isolated or associated with other systemic or ocular malformations (dysgenesis of iris, angle and peripheral cornea).

**Ocular:** Buphthalmos; corneal haze; glaucoma; epiphora.

**Clinical:** None.


Glaucoma with Elevated Episcleral Venous Pressures

**General:** Autosomal dominant.

**Ocular:** Open-angle glaucoma; elevated episcleral venous pressure; dilated episcleral veins.

**Clinical:** None.


Glaucoma, Goniodysgenesis (Dysgenic Glaucoma; Steroid Glaucoma)

**General:** Autosomal dominant; use of topical dexamethasone under the two-allele system; three genotypes; high, intermediate, and low; more prominent in blacks; mechanism may be related to morphologic changes in the trabecular meshwork.

**Ocular:** Glaucoma; cataracts.

**Clinical:** None.


Glaucoma, Hereditary Juvenile

**General:** Autosomal dominant, present at birth; there has been linkage with chromosome 1q21q23; age at diagnosis is 5 to 30 years; positive family history; male-to-female ratio of 2:1.

**Ocular:** Dysgenesis of the iris and iridocorneal angle; glaucoma; hypoplasia of iris; dark-colored irides; myopia; smooth iris; prominent iris processes; grayish-pale color of trabecular meshwork.

**Clinical:** None.


**Glaucoma, Recessive Juvenile**

**General:** Autosomal recessive; rare; normal parents and consanguineous marriages; asymptomatic and insidious onset.

**Ocular:** Buphthalmos; aching of eyes; colored halos around lights; elevated intraocular pressure; corneal epithelial edema; severe cupping and atrophy of optic nerve; constriction of visual fields.

**Clinical:** Headaches.


**Glucagonoma Syndrome**

**General:** Alpha-cell islet tumor of pancreas with retrobulbar neuritis first sign and necrolytic migratory erythema early signs.

**Ocular:** Central scotoma; retrobulbar neuritis.

**Clinical:** Necrolytic migratory erythema; diabetes; hypoaminoacidemia and hyperglucagonemia secondary to alpha-cell islet tumor of pancreas; anemia; glossitis; weight loss; angular stomatitis; onycholysis; diarrhea; monochromic monocytic anemia; recurrent venous thrombosis (associated with alpha-cell tumor).


**Goldberg Disease**

**General:** Unclassified syndrome with features of mucopolysaccharidoses, sphingolipidoses, and mucolipidoses; deficiency of neuraminidase; located in chromosome 20q 13.1.

**Ocular:** Macular cherry-red spot; corneal clouding; cerebromacular degeneration.

**Clinical:** Dwarfism; gargoyl facies; mental retardation; seizures; hearing disorder.


**Goldenhar Syndrome (Oculo-Auriculo-Vertebral Dysplasia; Goldenhar-Gorlin Syndrome)**

**General:** Most cases have been sporadic, but cases of autosomal dominant and recessive inheritance have been reported; male preponderance (60%); present at birth.

**Ocular:** Anophthalmia; colobomata of choroid, iris, and eyelid; antimongolian slant of lid fissure; epibulbar dermoid or lipodermoids of conjunctiva, cornea, and orbit; tilted optic disk; nerve hypoplasia; microphthalmia; macular heterotopia; tortuous retinal vessels.

**Clinical:** Frontal bulging of the skull; receding chin; malar hypoplasia; micrognathia and macrostomia; auricular appendices (single or multiple); multiple vertebral anomalies; preauricular fistulas; mental retardation.


Goldscheider Syndrome (Weber-Cockayne Syndrome; Epidermolysis Bullosa; Dominant Epidermolysis Bullosa Dystrophiea Albopapuloidea)

**General:** Rare; Weber-Cockayne syndrome, inherited as an autosomal dominant trait, is actually a milder form without scar formation, whereas Goldscheider syndrome, inherited either autosomal dominant or recessive, shows dystrophic changes with scarring; consanguinity frequent.

**Ocular:** Blepharitis; shrinkage of conjunctiva; pseudomembrane formation with symblepharon; conjunctivitis; bullous keratitis and subepithelial blisters lead to erosions with subsequent ulcerations and corneal opacities or even perforation; sclera may be similarly involved; lagophthalmos, cicatricial lacrimal stenosis; retinal detachment; cataract; pannus.

**Clinical:** Vesicular and bullous skin lesions and similar lesions of mucous membranes occur spontaneously or after mild trauma; keloid scars and contraction after healing are common in the dystrophic forms, whereas in the mild form the lesions heal without scarring but may leave some skin pigmentation; growth and mental retardation may be present in the group with recessive inheritance; stenosis of the larynx due to scarring may occur.


Goltz Syndrome (Focal Dermal Hypoplasia Syndrome)

**General:** X-linked dominant inheritance; lethal in males; skin manifestations present at birth.

**Ocular:** Microphthalmia; strabismus; coloboma of iris and/or choroid; epiphora; blue sclera; nystagmus; anophthalmos; keratoconus.

**Clinical:** Skin atrophy and linear pigmentation; telangiectasias of trunk and extremities; superficial, localized fatty skin deposits; multiple papillomas of mucous membranes and periorificial skin (oral, genital, anal); anomalies of extremities with syndactyly, oligodactyly, adactyly; hypohidrosis; paper-thin nails may be present; spina bifida; hypoplasia of right clavicle; umbilical or inguinal hernia.


Gonorrhea

**General:** Caused by *Neisseria gonorrhoeae,* which is transmitted sexually.

**Ocular:** Conjunctivitis; eyelid edema; keratitis; uveitis.

**Clinical:** Pelvic inflammatory disease; arthritis; dermatitis; carditis; meningitis.


Goodpasture Syndrome

**General:** Chronic, relapsing pulmonary hemosiderosis, often in association with fatal glomerulonephritis; rare; occurs in young males.
Ocular: Episcleritis; juxtapapillary subretinal neovascular membranes; superficial retinal hemorrhages; bilateral peripheral retinoschisis; hemorrhagic and/or exudative retinopathy; nonrhegmatogenous retinal detachment (rare).

Clinical: Cough with recurrent hemoptysis; dyspnea; pulmonary infiltrates; hypochromic iron deficiency anemia; glomerulonephritis; progressive renal failure; diffuse hemorrhagic inflammation of lung.


Gopalan Syndrome (Burning Feet Syndrome; Nutritional Melalgia Syndrome)

General: Female preponderance; onset age 20 to 40 years; pantothenic acid deficiency; lack of nicotinic acid, and low-protein diet are contributory factors; found in malnourished populations, detainees in prison camps, and chronic alcoholics.

Ocular: Decreased vision; central or paracentral scotomata.

Clinical: Hyperalgesia; hyperesthesia; severe burning of palms and soles (more pronounced at night); excessive sweating; circulatory insufficiency; tachycardia; muscle atrophy.


Gorlin-Chaudhry-Moss Syndrome

General: Etiology unknown.

Ocular: Microphthalmia; hypertelorism; depressed supraorbital ridges; inability to open or close lids fully because of incomplete lid development; antimongoloid, oblique palpebral fissures; sparse eyelash development; lid defect (notching); horizontal nystagmus at extreme lateral gaze; limited upper gaze; astigmatism; marked hyperopia; corneal scars (possibly due to exposure keratitis); keratoconus.

Clinical: Craniofacial dysostosis; saddled appearance of upper face; high-arched, narrow palate; dental anomalies (size, number, position); hypertrichosis; hypoplasia of labia majora; patent ductus arteriosus; normal mental development; fatigue; frontal headache.


Gout (Hyperuricemia)

General: Genetic disease of purine metabolism and renal excretion of uric acid.

Ocular: Conjunctivitis; episcleritis; posterior scleritis; ocular motor disturbances; iritis; band keratopathy; interpalpebral paralimbal nodules.

Clinical: Acute inflammatory arthritis; accumulation of sodium urate deposits; uric acid nephrolithiasis; renal failure; tophi in any body tissue; marked swelling of feet and ankles.


Gradenigo Syndrome (Temporal Syndrome; Lannois-Gradenigo Syndrome)

General: Caused by extradural abscess of the petrous portion of the temporal bone; good prognosis.
Ocular: Ipsilateral paralysis (cranial nerve VI); transient involvement of cranial nerves III and IV occasionally present; severe pain in area of ophthalmic branch (cranial nerve V); photophobia; lacrimation; reduced corneal sensitivity; optic nerve involvement occasionally present.

Clinical: Inner ear infection with deafness; mastoiditis; facial paresis possible; temperature may be elevated; meningeal signs possible; can occur rarely as a complication of otitis media.


Graft Versus Host Disease

General: Major complication of bone marrow transplantation; donor T lymphocytes attack recipient's cells; targets are skin, liver, intestine, oral mucosa, conjunctiva, lacrimal gland, vaginal mucosa, and esophageal mucosa.

Ocular: Keratoconjunctivitis; photophobia; hemorrhagic conjunctivitis; proptosis; intraretinal hemorrhages; nerve palsy; herpes simplex/herpes zoster manifestations; uveitis; corneal epithelial denudement; conjunctival scarring; dry eye syndrome; corneal melt; dacrooadenitis; keratoconjunctivitis sicca; cataract; retinitis.

Clinical: Leukemia; aplastic anemia; exanthematous dermatitis; hepatitis; enteritis; scleroderma-like involvement of skin; chronic liver dysfunction; recurrent bacterial infections.


Granuloma Annulare

General: Benign; self-limited dermatosis; etiology unknown but reported to follow insect bites, sun exposure, trauma, viral infection, and psoralen ultraviolet radiation therapy; hereditary predisposition; seen in children and young adults.

Ocular: Granuloma of lid; predilection for lateral upper lid and outer canthus; lesions are clinically similar to the subcutaneous nodules of rheumatoid arthritis.

Clinical: Skin lesions localized, generalized, subcutaneous, perforating, or arcuate dermal erythema; papules often arranged in a complete or half circle.


Granuloma Faciale

General: Uncommon disease; etiology unknown; characterized by single or multiple cutaneous nodules usually occurring on the face; asymptomatic; most common in males; seen in whites, rarely in blacks and Japanese.

Ocular: Unusual eyelid nodules.

Clinical: Cutaneous nodules most often on face but may appear anywhere; lesions are soft, elevated, well-circumscribed nodules, from a few millimeters to several centimeters in size; extrafacial lesions are extremely rare but have been reported.


Granuloma Venereum

General: Donovania granulomatis; infective venereal disease; prevalent in black women; Chlamydia trachomatis is an intracellular bacterium lacking respiratory enzymes that has an affinity for mucosal epithelium; serotypes A through C have been epidemiologically associated with trachoma; serotypes E through K have been associated with genital infection and keratoconjunctivitis in sexually active adults and neonates; other serotypes have been associated with lymphogranuloma venereum and Reiter syndrome.

Ocular: Lid and orbit granulomas.
Clinical: Painless primary lesions; painful secondarily infected ulcers.

Gray Iris Syndrome

General: Excessive trauma of iris at time of lens implantation with loss of posterior iris pigment; originally had blue irides.
Ocular: Pigmentary glaucoma; gray iris; massive pigment deposits in chamber angle; nonfixated intraocular lens.
Clinical: None.

Grayson-Wilbrandt Syndrome (Reis-Buecklers Syndrome; Corneal Dystrophy of Reis-Buecklers)

General: Onset at end of first decade; infrequent episodes of eye redness and pain; autosomal dominant trait; electron microscopy reveals peculiar curly material in the subepithelial fibrous tissue that parallels the distribution of attachment proteins.
Ocular: Corneal changes variable from a mottled scarring to gray macular opacities of the anterior limiting membrane of the cornea; strabismus.
Clinical: None.

Greig Syndrome (Ocular Hypertelorism Syndrome; Hypertelorism; Primary Embryonic Hypertelorism; Hypertelorism Ocularis)

General: Condition is rare; sporadic or hereditary; autosomal dominant or sex linked; if not associated with mental deficiency, then adequate mental and physical development is found.
Ocular: Hypertelorism (wide spacing of orbits); enophthalmos; epicanthus; deformities of eyelids and brows; defects of the palpebral fissure; bilateral sixth nerve paralysis; esotropia; astigmatism; optic atrophy by tension on the optic nerve; strabismus.
Clinical: Skull may show mild malformations, including bitemporal eminences and decreased anteroposterior diameter; harelip; high-arched palate; cleft palate; broad and flat nasal root; mental impairment.

Grönblad-Strandberg Syndrome (Systemic Elastodystrophy; Pseudoxanthoma Elasticum; Elastorrhexis; Darier-Grönblad-Strandberg Syndrome)

General: Autosomal recessive; female-to-male ratio of 2:1; inheritance is usually autosomal recessive, but it also has reported as autosomal dominant.
Ocular: "Angioid streaks" of the retina; macular hemorrhages and transudates not infrequent; choroidal sclerosis; retinal detachment; keratoconus; cataract; paralysis of extraocular muscles (secondary to vascular lesions of central nervous system); subluxation of lens; exophthalmos; optic atrophy; vitreous hemorrhages; Salmon spot multiple atrophic peripheral RPE lesions; reticular pigment dystrophy of the macula; optic disk drusen; multiple small crystalline bodies associated with atrophic RPE changes.
Clinical: Pseudoxanthoma elasticum with thickening, softening, and relaxation of the skin; skin changes are symmetrical in skin folds near large joints (axilla, elbow, inguinal region, lower abdomen, neck); flattening of the pulse curve and peripheral vascular disturbances; gastrointestinal hemorrhages.


**Grouped Pigmentation of the Macula**

**General:** Autosomal recessive.

**Ocular:** Grouped pigmentation limited to foveal area; metamorphopsia; pigmented spots around a clear hole in the foveal area.

**Clinical:** None.


**Gruner-Bertolotti Syndrome**

**General:** Various causes, including pineal tumor, supranuclear lesions, thrombosis of anterior choroidal artery, aneurysm or tumor; combination of Parinaud and von Monakow syndromes (see Parinaud Syndrome; von Monakow Syndrome).

**Ocular:** Hemianopia; lid retraction; ptosis; extraocular muscle paralysis; papilledema.

**Clinical:** Vertigo; hemiplegia; sensory disturbances; brain tumors.


**Guillain-Barré Syndrome (Landry Paralysis; Acute Infectious Neuritis; Acute Polyradiculitis; Acute Febrile Polyneuritis; Acute Idiopathic Polyneuritis; Inflammatory Polyradiculoneuropathy; Landry-Guillain-Barré-Strohl Syndrome; Postinfectious Polyneuritis)**

**General:** Etiology unknown; occurs from age 16 to 50 years; Fisher syndrome is a variant (see Fisher Syndrome).

**Ocular:** Facial nerve paralysis with paralytic ectropion of the lower eyelid; mild-to-complete external ophthalmoplegia; optic neuritis; papilledema; ptosis; anisocoria; nystagmus; dyschromatopsia; scotoma; bilateral tonic pupils.

**Clinical:** Polyneuritis involving facial peripheral motor nerves and spinal cord; facial diplegia; bladder incontinence; variable degrees of paralysis, usually beginning in lower extremities; tendon reflexes absent; involvement of respiratory muscles possible; paresthesia (symmetrical).


**Gyrate Atrophy (Ornithine Ketoacid Aminotransferase Deficiency)**

**General:** Deficiency of the enzyme ornithine aminotransferase; autosomal recessive; chronic, progressive dystrophy; responsible human gene has been localized to chromosome 10.
Ocular: Chorioretinal atrophy; crystalline deposits associated with brown pigment in fundus; myopia; cataract; keratoconus; night blindness; constricted visual fields; axial hypermetropia; cobblestone-like peripheral lesions; blunting of ciliary processes; iris atrophy.

Clinical: Absence of enzyme ornithine ketoacid transaminase; elevated levels of amino acid ornithine in body fluids; seizures; abnormal electroencephalography; eosinophilic subsarcolemmal deposits are seen on muscle biopsy; massive cystinuria, and lysinuria; diabetes.


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Haemophilus Aegyptius (Koch-Weeks Bacillus) 545

**General:** Caused by gram-negative Koch-Weeks bacillus in warm-climate regions; characterized by a 24- to 48-hour incubation period; now classified as *Haemophilus influenzae* biotype III; *H influenzae* is divided into biotypes based on biochemical reactions (indole production, urease activity, ornithine decarboxylase activity) and into serotypes based on their capsular polysaccharides; common cause of purulent conjunctivitis and preseptal cellulitis in children.

**Ocular:** Conjunctivitis; corneal opacity; corneal ulcer; phlyctenular keratoconjunctivitis; keratitis; cellullitis of lid; pseudoptosis; uveitis; petechial subconjunctival hemorrhages.

**Clinical:** Coryza; systemic symptoms are rare.


Haemophilus Influenzae 546

**General:** Gram-negative rod.

**Ocular:** Conjunctivitis; cellulitis; tenonitis; uveitis; vitreous opacity; pannus; corneal opacity.

**Clinical:** Pharyngitis; epiglottitis; laryngotracheitis; pneumonia; bronchitis; otitis media; meningitis; cellulitis; septic arthritis; sinusitis.


Hajdu-Cheney Syndrome 547

**General:** Rare; autosomal dominant; disorder of bone metabolism

**Ocular:** Bilateral visual loss; choroidal folds; optic nerve head swelling; optic neuropathy; optic nerve meningocele

**Clinical:** Facial dysmorphism; progressive platybasia; syringomyelia; curvature of the spine; aplasia of facial sinuses

Hallermann-Streiff Syndrome (Dyscephalic-Mandibulo-Oculo-Facial Syndrome; Oculo-Mandibulo-Dyscephaly; Ullrich-Fremery-Dohna Syndrome; Francois Dyscephalic Syndrome; Mandibulo-Oculo-Facial Dyscephaly Syndrome; Francois-Hallermann-Streiff Syndrome; Hallermann-Streiff-Francois Syndrome; Audry I Syndrome; Dohna Syndrome; Francois Syndrome (1); Dyscephaly-Teeth Abnormality-Dwarfism; Dyscephalia Oculomandibularis-Hypotrichosis; Mandibulo-Ocular Dyscephalia Hypotrichosis; Fremery-Dohna Syndrome; Oculo-Mandibulo-Facial Dyscephaly)

**General:** Rare; familial occurrence and consanguinity; males and females equally affected.

**Ocular:** Microphthalmos (bilateral); proptosis; nystagmus; strabismus; cataracts; bilateral optic atrophy; coloboma of optic disk, choroid, and iris; keratoglobus; microcornea; antimongoloid slant; iris atrophy; uveitis; blue sclera; persistent pupillary membrane; secondary glaucoma.

**Clinical:** Malformations of skull (brachycephaly), facial skeleton, and jaws; erupted teeth at birth; diminished hair growth; hyperextensibility of joints; short stature; skin atrophy; mental deficiency; predisposition to upper airway compromise; obstructive sleep apnea.


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Hallervorden-Spatz Syndrome (pigmentary Degeneration of Globus Pallidus; Progressive Pallidal Degeneration Syndrome)

**General:** Etiology unknown; autosomal dominant; onset between age 7 and 9 years; globus pallidus and pars reticularis of the substantia nigra are involved with demyelinization and degenerative processes; perhaps a form of iron storage disease.

**Ocular:** Nystagmus; retinitis pigmentosa; optic nerve atrophy; degeneration of photoreceptors; retinal gliosis; narrowing and obliteration of blood vessels with perivascular cuffing; degeneration of retinal pigment epithelial cells.

**Clinical:** Slowly progressing spasticity and rigidity of the extremities; emotional disturbances (pseudobulbar type); dementia; clubfoot; dysphagia; ataxosis; dysphonia; choreoathetosis; rigidity; seizures; pyramidal signs; generalized dystonia.


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Hallgren Syndrome (Retinitis Pigmentosa-Deafness-Ataxia Syndrome; Usher Syndrome Type I)

**General:** Autosomal recessive inheritance.

**Ocular:** Horizontal nystagmus (10%); cataract; retinitis pigmentosa; retinal atrophy; narrow retinal vessels; optic atrophy; keratoconus.

**Clinical:** Congenital deafness (complete or at least severe auditory impairment); mental deficiency (25%); vestibulocerebellar ataxia (90%); schizophrenia-like symptoms (25%).

### Hamman-Rich Syndrome (Alveolar Capillary Block Syndrome; Diffuse Pulmonary Fibrosis Syndrome; Rheumatoid Lung Syndrome)

**General:** Etiology unknown; insidious onset with progressive exertional dyspnea; association with rheumatoid arthritis or scleroderma; autosomal recessive; occurs between age 40 and 50 years.

**Ocular:** Xerophthalmia; keratomalacia; retinal venous congestion and engorgement; ischemic retinopathy; cystic macular changes.

**Clinical:** Cyanosis; dyspnea; cough; weight loss; clubbing of fingers; high sodium and chloride concentrations in sweat; heart failure.


### Haney-Falls Syndrome (Congenital Keratoconus Posticus Circumscriptus Syndrome)

**General:** Etiology unknown; autosomal dominant or recessive.

**Ocular:** Hypertelorism (mild); lateral canthi are displaced upward; myopic astigmatism; sharply localized posterior curvature of the cornea; corneal nebulae.

**Clinical:** Mental retardation; retarded growth; broad nose; brachydactyly; pterygium colli; barrel chest.


### Hanhart Syndrome (Richner Syndrome; Recessive Keratosis Palmoplantaris; Pseudoherpetic Keratitis; Richner-Hanhart Syndrome; Tyrosinemia II; Tyrosinosis; Pseudodendritic Keratitis)

**General:** Autosomal recessive; consanguinity.

**Ocular:** Excess tearing; photophobia; dendritic lesions of the cornea with corneal sensitivity not affected; keratitis; papillary hypertrophy of conjunctiva; corneal haze; neovascularization of cornea; cataract; nystagmus.

**Clinical:** Dyskeratosis palmoplantaris; diffuse keratitis; dystrophy of nails; hypotrichosis; mental retardation (usually pronounced); sensorineural hearing loss.


### Hansen Disease (Leprosy)

**General:** Communicable disease caused by *Mycobacterium leprae*.
Ocular: Keratitis; leukoma; pannus; corneal ulcer; uveitis; iris atrophy; dacryocystitis; anisocoria; multiple pupils; decreased or absent pupillary reaction to light; paralysis of seventh nerve; episcleritis; blepharospasm; lagophthalmos; madarosis; secondary glaucoma; decreased intraocular pressure; subconjunctival fibrosis; punctate epithelial keratopathy; posterior subcapsular cataract; corneal hypesthesia; prominent corneal nerves; iridocyclitis; foveal avascular keratitis; scleritis; interstitial keratitis; iris pearls; dry eye.

Clinical: Disease affects primarily the skin, mucous membrane, and peripheral nerves.


Happy Puppet Syndrome (Puppet Children)

General: Etiology unknown; very rare form of infantile epilepsy.

Ocular: Optic atrophy; deficiency of choroidal pigment; lightly colored irides; Brushfield spots; retinal pigment epithelium abnormalities; heterotropia; blindness.

Clinical: Mental retardation; seizures; puppet-like ataxia; paroxysms of laughter; absent speech; microcephaly; horizontal occipital depression; brachycephaly; prognathism, abnormal electroencephalographic findings.


Harboyan Syndrome (Congenital Corneal Dystrophy and Sensorineural Hearing Loss; Maumenee Syndrome; Corneal Dystrophy, Congenital Hereditary Endothelial)

General: Autosomal recessive; both sexes affected; corneal edema present at birth; slow and progressive; both dominant and recessive forms of this disorder have been described.

Ocular: Bluish-white opacities of cornea with normal sensitivity and no vascularization; nystagmus; keratoconus.

Clinical: Sensorineural hearing loss with childhood onset.


Harlequin Syndrome (Bullous Ichthyosiform Erythroderma; Collodion Baby; Congenital Ichthyosis; Epidermolysis Hyperkeratosis; Ichthyosis; Ichthyosis Vulgaris; Lamellar Ichthyosis; Nonbullous Ichthyosiform Erythroderma; Xeroderma; X-Linked Ichthyosis)

General: Autosomal inherited disorder; affects both sexes; normal at birth; onset within first 7 days.

Ocular: Keratopathy; corneal scarring; keratitis; conjunctivitis; lagophthalmos; photophobia; ectropion; lid erythema; lacrimation.

Clinical: At birth, the skin surface is moist, red, and tender; within several days, thick verrucous scales form.


**Hartnup Syndrome (pellagra-Cerebellar Ataxia-Renal Aminoaciduria Syndrome; H Disease; Niacin Deficiency)**

**General:** Recessive; inborn error in amino acid metabolism with abnormal metabolism of tryptophan; both sexes affected; presents from infancy.

**Ocular:** Ectropion; symblepharon; nystagmus; scleral ulcers; corneal leukemia; photophobia; diplopia during attacks.

**Clinical:** Dermatitis (similar to pellagra) with skin eruptions; progressive mental retardation; cerebellar ataxia.


**Hays-Wells Syndrome (AEC Syndrome; Ankyloblepharon-Ectodermal Defects-Cleft Lip/Palate Syndrome)**

**General:** Autosomal dominant disease, as initially described, but may exist as an autosomal recessive disorder.

**Ocular:** Ankyloblepharon, filiforme adnatum (fused eyelids).

**Clinical:** Coarse, wiry, sparse hair; dystrophic nails; slight hypohidrosis; maxillary hypoplasia; cleft lip and palate.


**Headache Neurologic Defects and Cerebrospinal Fluid Lymphcytosis Syndrome**

**General:** Age range 7-52 years of age; no gender bias.

**Ocular:** Papilloma; homonymous hemianopia; photopsias; sixth nerve palsy.

**Clinical:** Headache; hemisensory defects; muscle weakness; aphasia; elevated intracranial pressure.


**Head-Riddoch Syndrome**

**General:** Occurs in quadriplegics; caused by distention of a viscus below the level of spinal cord lesion; seen most frequently in people with high cervical cord lesion; may follow catheter obstruction, fecal impaction, bladder calculi, urinary infection, or decubiti.

**Ocular:** Dilated pupils; blurred vision.

**Clinical:** Sweating; flushing; pilomotor activity; nasal stuffiness; headaches; generalized seizures; bradycardia; hypertension.


Head H, Riddoch G. The automatic bladder, excessive sweating and some other reflex conditions in gross injuries of the spinal cord. Brain 1917; 40:188-263.

**Heerfordt Syndrome (Uveoparotid Fever; Uveoparotitis; Uveoparotic Paralysis)**

**General:** Occurs in young adults, more frequently in females than in males; usual cause is sarcoidosis.

**Ocular:** Band keratopathy; keratoconjunctivitis sicca; uveitis; optic atrophy; papillodema; episcleritis; snowball opacity of vitreous; retinal vasculitis; proptosis; cataract; paralysis of seventh nerve; sarcoid nodules of eyelid, iris, ciliary body, choroid, and sclera; dacryoadenitis.

**Clinical:** Parotid gland swelling; facial paralysis; lymphadenopathy; splenomegaly; cutaneous nodules; facial nerve palsy.
Hemangioma

**General:** Can occur throughout the body, but particularly in the head; primary intrasosseous orbital hemangiomas is rare; capillary hemangioma of the orbit and eyelids generally is unilateral.

**Ocular:** Hemangiomas of lids or orbit; ptosis; strabismus; amblyopia; proptosis; optic atrophy; hypermetropia; cavernous hemangiomas are the most common benign orbital tumors of adults.

**Clinical:** Ipsilateral hemangiomas of the brain and meninges.


Hemeralopia

**General:** Autosomal dominant.

**Ocular:** Complete loss of the outer quadrant of visual field bilaterally; visual fields become progressively more constricted until blindness occurs; corneal ulcers; photoreceptor dysfunction.

**Clinical:** None.


Hemifacial Hyperplasia with Strabismus (Bencze Syndrome)

**General:** Autosomal dominant; abnormal growth of facial skeleton and soft tissue and viscera; left side prominent.

**Ocular:** Strabismus; amblyopia.

**Clinical:** Facial asymmetry; submucous cleft palate.


Hemifacial Microsomia Syndrome (Unilateral Facial Agenesis; Otomandibular Dysostosis; Francois-Hausrate Syndrome)

**General:** No inheritance pattern; left side of face seems to be more frequently involved; facial asymmetry usually most obvious finding; both sexes affected; alteration of intrauterine environment is possible cause.

**Ocular:** Microphthalmos; congenital cystic phthalmia; enophthalmos; strabismus; cataract; colobomata of iris, choroid, and retina.

**Clinical:** Microtia; macrostomia; failure of development of mandibular ramus and condyle; external auditory meatus may be absent; single or numerous ear tags; hypoplasia of facial muscles unilaterally; pulmonary agenesis (ipsilateral side); associated with Goldenhar syndrome.

Hemimacrosomia Syndrome (Steiner Syndrome; Hemigigantism; Hemifacial or Unilateral Hypertrophy)  

**General:** Occasionally hereditary, although true etiology is obscure; right side affected more frequently than left side; slight male preponderance.

**Ocular:** Dilated pupil on the affected side; eccentric pupillary location; hypochromic heterochromia.

**Clinical:** Unilateral facial enlargement may be associated with enlargement of half of entire body to varied extent and degree; thickened skin over involved area with increased activity of sebaceous and sweat glands; telangiectasias and multiple nevi; polydactyly; syndactyly; macrodactyly; scoliosis.


Hemochromatosis  

**General:** Iron metabolism disorder; genetically determined, but mode of inheritance unknown; male preponderance 10:1; inheritance is autosomal recessive.

**Ocular:** Eyelid hyperpigmentation; diabetic retinopathy.

**Clinical:** Hemosiderin pigment deposition in many tissues; diabetes mellitus; cutaneous hyperpigmentation; cirrhosis of the liver; hypermelanotic pigmentation of skin; heart failure.


Hemolytic Anemia of Newborns (Icterus Gravis Neonatorum; Erythroblastosis Fetalis)  

**General:** Rh-positive/negative infant carried by an Rh-positive/negative mother; isoimmunization of the mother by her fetus of different blood group.

**Ocular:** Retinal hemorrhages; ophthalmoplegia; optic atrophy; yellow conjunctiva and lids.

**Clinical:** Jaundice; edema; liver and spleen palpable; cutaneous purpura; bleeding from mucosa.


Hennebert Syndrome (Luetic-Otitic-Nystagmus Syndrome)  

**General:** Caused by congenital syphilis; manifestations in childhood; when a fistula in the labyrinth exists, compression of the external auditory meatus will produce nystagmus of a wide amplitude (diagnostic of fistula).

**Ocular:** Spontaneous nystagmus when the column of air in the auditory canal is compressed; interstitial keratitis; disseminated syphilitic chorioretinitis may be present.

**Clinical:** Vertigo; fistula in the labyrinth; deafness; other clinical manifestations of congenital syphilis may be present, such as “saddle” nose and Hutchinson teeth.


Henoch-Schönlein Purpura (Purpura; Anaphylactoid Purpura)  

**General:** Occurs chiefly in children, although it can affect persons of any age; frequently follows an upper respiratory tract infection within 3 weeks.

**Ocular:** Retinal hemorrhages; iritis; optic neuritis.

**Clinical:** Purpuric skin rash; concentrated on lower extremities; joint pain; abdominal pain; hematuria; central nervous system involvement.

Hepatic Failure 572

**General:** Liver failure from infections or from toxic or inflammatory causes.

**Ocular:** Visual field defects; scleral icterus; night blindness; abnormal color vision; eyelid retraction; lid lag; Kayser-Fleischer ring; yellow discoloration of the conjunctiva.

**Clinical:** Bilirubin accumulation; reduced vitamin A levels.


Hereditary Ectodermal Dysplasia Syndrome (Siemens Syndrome; Keratosis Follicularis Spinulosa Syndrome; Hypohidrotic Ectodermal Dysplasia; Christ-Siemens-Touraine Syndrome; Weech Syndrome; Anhidrotic Ectodermal Dysplasia; Ichthyosis Follicularis) 573

**General:** Autosomal recessive inheritance; strong male preponderance (about 95%); linked to X-chromosome.

**Ocular:** Complete loss of eyebrows (madarosis); follicular keratosis; blepharitis; entropion or ectropion; reduced tear formation or epiphora; myopia; keratoconjunctivitis; corneal erosions and ulcers (recurrent); corneal dystrophy; cataract; increased periocular pigmentation; mongoloid lid slant; photophobia; absence of iris; luxation of lens; papillary abnormalities; cataracts.

**Clinical:** Mental retardation; dry skin and anhidrosis (reduced number of sweat glands); hypotrichosis; follicular hyperkeratosis (neck, palms, soles); hypohidrosis.


**Hereditary Microcornea, Glaucoma, and Absent Frontal Sinuses** 574

**General:** Autosomal dominant.

**Ocular:** Microcornea; glaucoma; epicanthal folds; optic cupping.

**Clinical:** Thickened palmar skin; torus palatinus; frontal sinus hypoplasia.


**Hermansky-Pudlak Syndrome (Oculocutaneous Albinism and Hemorrhagic Diathesis)** 575

**General:** Autosomal recessive; mostly affects children or young adults; tyrosinase-positive oculocutaneous albinism; abnormal platelets; increased ceroid-like material in the reticuloendothelial system.

**Ocular:** Translucent irides; minimally pigmented ocular fundi; nystagmus; foveal hypoplasia; large refractive errors; strabismus; fundus hypopigmentation.
Clinical: Gingival bleeding; epistaxis; easy bruising; pale skin; interstitial lung disease; defect in platelet function; "imperfect" oculocutaneous albinism; restrictive lung disease; ulcerative colitis.


Hermit Syndrome

General: Seen in alcoholic men in their 60s and 70s living in relative social isolation; secondary to history of sun exposure or chronic irritation; high prevalence in the tropics or developing countries; masquerades as conjunctivitis or orbital cellulitis; develops from carcinoma in situ (see Bowen Disease).

Ocular: Squamous cell carcinoma of conjunctiva; proptosis; erythema of eyelids; edema of eyelids; conjunctival hyperemia; vitreous hemorrhage; ciliary body hemorrhage; choroidal hemorrhage; paresis of extraocular muscles; ocular pain; orbital cellulitis.

Clinical: Vitamin A deficiency; nutritional disorders; recurrent infections; actinic keratoses.


Herpes Simplex

General: Large, complex deoxyribonucleic acid (DNA) virus.

Ocular: Conjunctivitis; keratitis; iridocyclitis; corneal ulcer; uveitis; hypHEMA; hypopyon; iris atrophy; cataract; scleritis; dacryoadenitis; blepharitis; acute retinal necrosis.

Clinical: Recurrent skin vesicles on lids, perioral area, nose, and genitalia; meningitis, encephalitis.


Herpes Simplex Masquerade Syndrome

General: Acanthamoeba keratitis occurs in those who wear soft contact lenses daily; confused with herpes simplex; Acanthamoeba culbertsoni, Acanthamoeba castellanii, and Acanthamoeba polyphaga are causative agents; agents found in distilled water, hot tubs, and swimming pools (see Acanthamoeba).

Ocular: Keratitis; corneal ulcer; corneal cysts; stromal infiltrates and necrosis; scleritis; uveitis; epiphora; pseudodendrites.

Clinical: None.


Herpes Zoster

**General:** Caused by varicella zoster virus; about 75% of cases occur in persons over age 45 years; condition is more frequent with advancing age and in patients who are immunocompromised by drugs or disease; in particular, an increasing number of patients with herpes zoster ophthalmicus are immunosuppressed.

**Ocular:** Conjunctivitis; keratitis; recurrent corneal ulcer; neuralgia; zoster rash of eyelids; uveitis; iris atrophy; scleritis; cataract; optic neuritis; paralysis of third nerve; proptosis; paralysis of lids; orbital apex syndrome; retinitis; neurotrophic keratitis; acute retinal necrosis; progressive outer retinal necrosis; ocular motor nerve pareses; tonic pupil; encephalitis; vasculitis.

**Clinical:** Local lesions involving the posterior or root ganglia; nerve damage; tissue scarring.


Herrick Syndrome (Dresbach Syndrome; Sickle Cell Disease; Drepanocytic Anemia)

**General:** Usually occurs in members of the black race; poor prognosis.

**Ocular:** Secondary glaucoma; telangiectasis of conjunctival vessels; scleral icterus; vitreous hemorrhages; cataract; retinal hemorrhages, exudates, and neovascularization; retinitis proliferans; microaneurysms; thrombosis of retinal venules; retinal vascular sheathing; central vein occlusion; angiod streaks; retinopathy with "black sunburst sign" in patients with SS hemoglobin; "sea fan sign" in patients with SC hemoglobin; comma signs of conjunctiva; fan-shaped neovascularization of iris; sector ischemic atrophy of iris; optic atrophy; white cotton mass of vitreous; retinal holes; color vision defects; central retinal artery obstruction; branch retinal artery obstruction; white without pressure; venous tortuosity; sickling maculopathy.

**Clinical:** Severe anemia with hemolytic crises; bone and joint aches; hemarthrosis; jaundice; hepatosplenomegaly.


Heterochromia Iridis

**General:** Autosomal dominant; can be associated with Horner, Waardenburg, and Marfan syndromes; may occur as an isolated phenomenon.

**Ocular:** Different pigmentation in the two irides or in the sectors of one iris (heterochromia iridium).

**Clinical:** None.


HIE Syndrome (Hyperimmunoglobulinemia E Syndrome)

**General:** Mononuclear cells inhibit normal neutrophil and monocyte chemotaxis; caused by *Staphylococcus aureus* and *Candida trachomatis*; autosomal dominant phenotype; onset at age 1 to 8 weeks; marked elevation of immunoglobulin E.

**Ocular:** Atopic keratitis; keratoconjunctivitis; photophobia; corneal ulcer; chorioretinal scars; lid edema.

**Clinical:** Pruritic dermatitis; dry skin; skin abscesses; pneumonia; upper respiratory infections; reduced resistance; coarse facial features.

Hilding Syndrome (Destructive Iridocyclitis and Multiple Joint Dislocations)

**General:** Atrophy of body cartilages and joint dislocations without destruction of bone or joint surfaces.

**Ocular:** Severe ocular hypotony; severe plastic iridocyclitis; iris atrophy; corneal endothelial precipitates; mature cataracts; retrolental inflammatory membrane.

**Clinical:** Multiple joint dislocations; hyperlaxity of joint capsules; generalized cartilage destruction with nose and ear deformities.


Histidinemia (Hyperhistidinemia; Histidase Deficiency)

**General:** Autosomal recessive; abnormality of amino acid metabolism due to lack of enzyme histidine ammonia lyase; the histidine gene has been assigned to 12q22-q23 by situ hybridization techniques.

**Ocular:** Nystagmus; hypopigmentation of the macula.

**Clinical:** Speech defects; mental retardation; head nodding movements; dysarthria; defective hand grip.


Histiocytosis X (Hand-Schuller-Christian Syndrome; Lipoid Granuloma; Xanthomatous Granuloma Syndrome; Schuller-Christian-Hand Syndrome; Letterer-Siwe Syndrome; Acute Histiocytosis X; Eosinophilic Granuloma; Reticuloendotheliosis Syndrome)

**General:** The term histiocytosis X has been proposed to include Letterer-Siwe disease, Hand-Schuller-Christian disease, and eosinophilic granuloma of bone; there are sufficient grounds to treat Hand-Schuller-Christian and Letterer-Siwe together as different phases of the same disease process; eosinophilic granuloma most likely represents a reaction pattern, sharing some histologic features with the first two but nonetheless carrying a more benign prognosis; Letterer-Siwe disease is referred to as acute differentiated histiocytosis; Hand-Schuller-Christian disease is referred to as subacute differentiated or chronic differentiated histiocytosis; Letterer-Siwe etiology is unknown, onset is in infancy and early childhood, and prognosis is generally poor; Hand-Schuller-Christian etiology is unknown, onset is in childhood, male preponderance is 2:1, and prognosis is chronic with remissions; eosinophil may play a contributory pathophysiologic role.

**Ocular:** Exophthalmos; ocular pulsations; orbital roof defects; xanthelasmas; blepharitis; internal opthalmoplegia; nystagmus; retinal hemorrhages; papilledema; optic atrophy; uveitis; hypopyon; pannus; bullous keratopathy; corneal ulcer; hypochromic heterochromia; retinal detachment; cataract; scleritis.

**Clinical:** Hepatosplenomegaly; lymphadenopathy; skin lesions with papular eruptions; ecchymosis; purpura; bone lesions; anemia; fatigue; anorexia; fever; xanthoma of the skin; diabetes insipidus; skull defects; lung fibrosis; cardiac insufficiency.


Histoplasmosis (Histoplasmosis Choroiditis; Histoplasmosis Maculopathy; Presumed Ocular Histoplasmosis Syndrome)  586

General: Fungal infection caused by *Histoplasma capsulatum*.

Ocular: Circumpapillary atrophy; maculopathy; scattered yellow "hистo" spots; optic disk edema; disseminated chorioiditis (immunocompromised patients); vitreous hemorrhage; punched-out chorioretinal lesions; choroidal neovascular membrane; exogenous endophthalmitis (isolated report).

Clinical: Pulmonary infection; fever; malaise.


HLA-B27 Syndromes  587

General: HLA system is the major histocompatibility complex (MHC) found on chromosome 6; associated with inflammatory disease

Ocular: Uveitis; Reiters syndrome; keratitis; band keratopathy; iris bombe’; pigment dispersion; papillary miosis; iris nodules; CME; disk edema; pars plana exudates; choroiditis

Clinical: Ankylosing spondylitis; arthritis; inflammatory bowel disease; psoriatic arthritis


HMC Syndrome (Hypertelorism, Microtia, Facial Clefting Syndrome)  588

General: Developmental defect marked by an abnormally wide space between two organs or parts; autosomal recessive.

Ocular: Hypertelorism.

Clinical: Microtia; clefting of the lip, palate, and nose; psychomotor retardation; conductive hearing loss; mild micrognathia; microcephaly; thenar hypoplasia; ectopic kidneys; atretic auditory canals; congenital heart malformations.


Hodgkin Disease  589

General: Hodgkin disease begins in the lymph nodes and usually spreads in a predictable fashion along contiguous chains of nodes; etiology may be viral; prevalent in males.

Ocular: Keratitis; uveitis; cataract; retinal hemorrhages; vasculitis; Horner syndrome; cortical blindness; papilledema; paralysis of oculomotor nerve; episcleritis; visual field defects; infiltration of choroid, conjunctiva, lacrimal gland, and orbit; papillitis; retrobulbar neuritis; oposconulus-myoclonus; keratitis sicca; infiltrative optic neuropathy; association with Vogt-Koyanagi-Harada syndrome; bilateral serous detachments of the macula.

Clinical: Painless cervical, axillary, or inguinal lymph node swelling; fever; weight loss; anemia; generalized pruritus.


**Hollenhorst Syndrome (Chorioretinal Infarction Syndrome)**

**General**: Caused by inadvertent pressure on the eye by the headrest from faulty positioning of the patient's head during surgery; ocular findings were immediately discovered when patient regains consciousness after surgery.

**Ocular**: Slight proptosis; ecchymosis of lid; marked lid edema; dilated and fixed pupil; hazy cornea; retinal edema; serous retinal detachment; cherry-red spot of the macula; attenuations of retinal arteries; pigmentary retinopathy; optic nerve atrophy; ophthalmoplegia.

**Clinical**: None.


**Homocystinuria Syndrome**

**General**: Rare disorder of amino acid metabolism; autosomal recessive inheritance; approximately one third of patients with this disorder have normal intelligence.

**Ocular**: Dislocated or subluxated lenses; spherophakia; cataract; retinal detachment; optic atrophy; keratitis; ocular hypotony; iris atrophy; uveitis; situs inversus optic disk; central retinal artery occlusion; high myopia; strabismus; pupillary block glaucoma; lens zonular fibers have abnormal glycoprotein with elevated concentration of cystine; ectopia lentis (nearly constant feature).

**Clinical**: Mental retardation; sparseness of hair; thromboembolism; arachnodactyly; generalized osteoporosis; thrombotic lesions of arteries and veins; abnormal cystathionine synthetase.

Cogan D. Dislocated lenses and homocystinuria. *Arch Ophthalmol* 1965; 74:446.


**Hoof and Mouth Disease**

**General**: Viral etiology.

**Ocular**: Conjunctival painful red blisters.

**Clinical**: Mucous membranes develop painful red blisters; lymph glands swollen; preauricular lymphadenopathy.


**Horner Syndrome (Bernard-Horner Syndrome; Cervical Sympathetic Paralysis Syndrome; Claude-Bernard-Horner Syndrome; Horner Oculopupillary Syndrome)**

**General**: Paralysis of cervical sympathetic; hypothalamic lesion with first neuron involved or lesion in the pons or cervical portion of cord; syndrome present in Babinski-Nageotte, Cestan-Chenais, Dejerine-Klumpke, Pancoast, Raeder, and Wallenberg syndromes (see Babinski-Nageotte Syndrome; Cestan-Chenais Syndrome; Dejerine-Klumpke Syndrome; Pancoast Syndrome; Raeder Syndrome; Wallenberg Syndrome).
Ocular: Enophthalmos; ptosis or narrowing of palpebral fissure; ocular hypotony; miosis (degree of miosis depends on site of lesion; most pronounced when roots of cranial nerves VII and VIII and first thoracic nerve are involved); hypochromic heterochromia (children more than adults); pupil does not dilate with cocaine.

Clinical: Anhidrosis on ipsilateral side of face and neck; transitory rise in facial temperature; hemifacial atrophy; may result from a variety of conditions, including cluster headache, parasellar neoplasms or aneurysms, internal carotid dissection or occlusion, and Tolosa-Hunt syndrome.


Hunt Syndrome (Ramsay-Hunt Syndrome; Geniculate Neuralgia; Herpes Zoster Auricularis) 594

General: Herpes of the geniculate ganglion; course is prolonged; characterized by severe pain that frequently precedes skin and mucosal lesions and may persist for some time after lesions have disappeared; sulfuduronate enzyme deficiency.

Ocular: Diminished lacrimation; absence of motor corneal reflex on affected side, whereas consensual reflex of noninvolved eye remains normal.

Clinical: Herpes zoster lesions of external ear and oral mucosa; severe pain in area of external auditory meatus and pinna; diminished hearing; tinnitus; vertigo, facial palsy; diminution or total loss of superficial and deep facial reflexes; zoster lesions may involve the scalp, face, and neck; hoarseness; absence of auricular lesions has been reported; progressive dementia; extensive frontal white matter change; myoclonus; ataxia; facial paralysis; hearing loss; hyperacusis; vertigo; dysgeusia; seizures; cerebellar ataxia; schizophrenia-like symptoms.


Hunter Syndrome (MPS II Syndrome; Mucopolysaccharidosis II; Systemic Mucopolysaccharidosis Type II) 595

General: Sex-linked recessive inheritance; clinically less severe than Hurler syndrome (MPS I) with a longer life span (into adulthood); similar to MPS I (Hurler syndrome), with chondroitin sulfate B and heparitin sulfate excreted in excess in the urine (see Sanfilippo-Good Syndrome; Morquio-Brailsford Syndrome; Scheie Syndrome; Maroteaux-Lamy Syndrome); X-linked recessive inheritance; decreased iduronate sulfatase.

Ocular: Visual fields may be constricted; splitting or absence of Bowman membrane in the periphery; stromal haze may be present; pigmentary degeneration of the retina; night blindness; narrowed retinal vessels and central choroidal sclerosis; bushy eyebrows; coarse eyelashes; ptosis; optic atrophy; papilledema; proptosis; angle-closure glaucoma; corneal clouding; scleral thickening; uveal effusion.

Clinical: Dwarfism; stiff joints; hepatosplenomegaly; gargoyle-like facies.


### Hurler Syndrome (Pfaundler-Hurler Syndrome; Gargoylism; Dysostosis Multiplex; MPS IH Syndrome; Systemic Mucopolysaccharidosis Type IH; Mucopolysaccharidosis IH)

**General:** Autosomal recessive inheritance; in addition to corneal opacities and enlargement of the head at birth, other symptoms become apparent at the end of the first year; death occurs usually before age 20 years; gross excess of chondroitin sulfate Band heparitin sulfate in the urine (see Hunter Syndrome; Sanfilippo-Good Syndrome; Morquio-Brailsford Syndrome; Scheie Syndrome; Maroteaux-Lamy Syndrome). Jensen suggested that the pathogenesis of the various mucopolysaccharidoses is the same but that the variations in the defective enzymes cause the different types; most common mucopolysaccharidosis, decreased α-iduronidase.

**Ocular:** Proptosis; hypertelorism; thick, enlarged lids; esotropia; diffuse haziness of the cornea at birth progressive to milky opacity; retinal pigmentary changes may exist; macular edema and absence of foveal reflex; optic atrophy; megalocornea; bushy eyebrows; coarse eyelashes; mucopolysaccharide deposits of iris, lens, and sclera; enlarged optic foramen; retinal detachment; anisocoria; buphthalmos; nystagmus; secondary open-angle glaucoma; progressive retinopathy with vascular narrowing; hyperpigmentation of the fundus; bone spicule; papilledema.

**Clinical:** Dorsolumbar kyphosis; head deformities with depressed nose bridge; short cervical spine; short limbs; macroglossia; enlarged liver and spleen; short stature; facial dysmorphism; progressive psychomotor retardation.


### Hurler-Scheie Syndrome (MPS I H/S)

**General:** Clinical disorder with severity midway between the Hurler and Scheie syndromes; genetic compound of the two alleles Hand S; autosomal recessive.

**Ocular:** Corneal clouding; chronically elevated optic disk; diminished or extinguished electroretinogram; pigmented retinopathy; glaucoma; optic atrophy; retinal pigmentary degeneration.

**Clinical:** Shares some clinical features of both MPS IH (Hurler) and MPS IS (Scheie). Severe bone involvement; minor intellectual impairment (see Hurler Syndrome; Scheie Syndrome); cardiac failure; cardiomyopathy; conduction defects; valvular heart disease.


### Hutchinson-Gilford Syndrome (progeria)

**General:** Inheritance unknown; belongs to group of ectodermal dysplasias (see Werner Syndrome-Progeria of Adults); elevated hyaluronic acid of unknown etiology, likely sporadic dominant mutation.

**Ocular:** Microphthalmia; hypotrichosis; microcornea; cataract.

**Clinical:** Appearance of "old age" in children; short stature to dwarfism; dyscephaly; atrophy of skin and subcutaneous adipose tissue; aplasia of maxilla; oligodontia; arteriosclerosis (premature); progeria.


Hutchinson J. Congenital absence of hair and mammary glands with atrophic condition of the skin and its appendages in a boy whose mother had been almost wholly bald from alopecia areata from the age of six. *Trans Med Chir Soc Edinb* 1886; 69:473.

Hutchinson Syndrome (Adrenal Cortex Neuroblastoma with Orbital Metastasis; Pepper Syndrome) 599

**General:** Metastatic infraorbital neuroblastoma after hematogenous dissemination of primary tumor; occurs in infants and children up to age 6 years; poor prognosis; in children neuroblastoma commonly involves the orbit; 15% of patients with neuroblastoma had proptosis and ecchymosis.

**Ocular:** Exophthalmos; lid hematoma; extraocular muscle palsy; subconjunctival hemorrhages; choroidal metastatic tumor; papilledema; optic atrophy.

**Clinical:** Severe anemia; increased sedimentation rate; urinary excretion of 3-methoxy-4-hydroxy mandelic acid.

Cox RA. Proptosis due to neuroblastoma of the adrenal cortex (Hutchinson's syndrome). Arch Ophthalmol 1948; 39: 713.


Hydatid Cyst (Echinococcosis) 600

**General:** Caused by *Echinococcus granulosus* acquired by contact with a dog host.

**Ocular:** Conjunctivitis; keratitis; exophthalmos; ptosis bulbi; optic atrophy; optic neuritis; papilledema; abscesses of orbit and cornea; retinal detachment; retinal hemorrhages; cataract; hypopyon; secondary glaucoma; hydatid cysts of the conjunctiva, eyelid, orbit, and lacrimal system; acute visual loss; vitreous mass.

**Clinical:** Pruritus; urticaria; pulmonary cysts; brain cysts; anaphylactic shock; death.


Hydranencephaly 601

**General:** Rare; development disorder in which cerebral hemispheres are replaced by a cystic space filled with cerebrospinal fluid, covered by intact meninges; short life expectancy of weeks to months, but adulthood is possible; positive association with cocaine.

**Ocular:** Pupillary abnormalities; strabismus; nystagmus; ptosis; optic nerve hypoplasia; chorioretinitis; retinal vessel attenuation; incomplete anterior chamber cleavage; microphthalmia; blepharospasm.

**Clinical:** Irritability; convulsions; muscular rigidity; decerebrate posturing; increasing head circumference; quadriplegia; psychomotor retardation.


Hydroa Vacciniforme 602

**General:** Sensitivity to sunlight.

**Ocular:** Conjunctivitis; corneal vesiculae; keratitis; cicatricial ectropion.

**Clinical:** Vesicular skin eruptions in areas exposed to sunlight.


Hydrophobia (Lyssa; Rabies) 603

**General:** Acute viral zoonosis of the central nervous system.
**Ocular:** Lid retraction; widening of palpebral fissure; retinal hemorrhages; mydriasis; paralysis of third, fourth, fifth, or seventh nerve; bilateral optic neuritis; branch retinal artery occlusion; vaccine-induced autoimmune demyelinative optic neuritis.

**Clinical:** Fever; headache; nausea; numbness; tingling; acute sensitiveness to sound and light; laryngeal and pharyngeal spasms; increased muscle tonus; convulsions; delirium; coma; death.


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**Hyperammonemia I (Carbamyl Phosphate Synthetase Deficiency; Hyperammonemia II; Ornithine Transcarbamylase Deficiency; Hyperammonemia-Hyperornithinemia-Homocitrullinuria Syndrome)**

**General:** Hyperammonemias I and II are due to errors at or near the "start" of the urea cycle; in hyperammonemia I, a decrease in the activity of the enzyme carbamyl phosphate synthetase, responsible for the first step of the cycle, results in the accumulation of excess ammonia; in hyperammonemia II, the defect is in ornithine transcarbamylase; type II occurs only in infants.

**Ocular:** Ptosis and visual loss; retinal depigmentation and chorioretinal thinning.

**Clinical:** Vomiting; screaming; confusion; lethargy; ataxia; mental retardation; atrophy of cerebral cortex; decreased vibration sense; bucco-facial lingual dyspraxia; learning difficulties; widespread manifestations in the central and peripheral nervous systems.


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**Hyperkalemic Familial Periodic Paralysis (Adynamia Episodica Hereditaria)**

**General:** Recurrent paralysis of skeletal muscles; occurs by age 10 years; usually occurs during the day when patient is sitting in a chair without exercise; attacks last 30 minutes to 2 hours.

**Ocular:** Transient attacks of staring with lid elevation in younger children; sclera above cornea is visible in adults when they look down; lid lag present during attacks but repeated up-and-down movements of eyes help.

**Clinical:** Muscle weakness; difficulty swallowing and coughing; tremor; episodes of quadriplegia lasting 2 to 3 weeks; salt craving; intense thirst; stomach pain.


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**Hyperlipoproteinemia**

**General:** Metabolic disorder characterized by abnormally elevated concentrations of specific lipoprotein particles in the plasma.

**Ocular:** Arcus; lipid keratopathy; xanthelasma; lipemia retinalis; lipemia of limbal vessels; xanthomata of choroid, conjunctiva, eyelids, iris, and retina; central retinal vein occlusion; Schnyder crystalline corneal dystrophy (association).

**Clinical:** Deposition of lipids at various sites throughout the body, such as skin, tendons, and vascular system.


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**Hyperopia, High**

**General:** Defect in eyesight in which the focal point falls behind the retina, resulting in farsightedness; autosomal recessive; eye shorter than normal.

**Ocular:** Farsightedness.
Hyperparathyroidism

**General:** Increased secretion of parathyroid hormone. Glasslike crystals of conjunctiva; band keratopathy; optic atrophy; papilledema; vascular engorgement of retina; ptosis; scleral thinning; ectopic calcifications in the choroid and sclera; unilateral visual loss; ischemic optic neuropathy.

**Clinical:** Hypercalcemia; hypophosphatemia; brown tumor.


Hyperpigmentation of Eyelids

**General:** Autosomal dominant; causes may include drugs, sun, or too little adrenaline.

**Ocular:** Unusual darkening of eyelids.

**Clinical:** None.


Hypertension

**General:** Elevated blood pressure.

**Ocular:** Retinal arterial narrowing; arteriosclerosis; hemorrhages; retinal edema; cotton-wool spots; fatty exudates; optic disk edema; exudative retinal detachment; optic neuropathy; swollen optic nerve; central retinal vein occlusion; branch retinal vein occlusion; choroidal ischemia.

**Clinical:** Systemic hypertension; patchy loss of muscle tone in vessel walls; vascular decompensation.


Hypertrichosis (Hirsutism)

**General:** Excessive hair growth due to endocrinologic or physiologic states.

**Ocular:** Keratitis; excessive cilia; ectropic cilia; monilethrix; pili torti; polytrichosis; reduplication of ciliary follicles; trichomegaly; association between congenital hypertrichosis lanuginosa and congenital glaucoma.

**Clinical:** Excessive hair growth on any part of the body except palms and soles; eczema; psoriasis; lupus erythematosus; association with acquired immunodeficiency syndrome.


Hypertrichosis Cubiti (Hairy Elbow Syndrome) 612

**General:** Autosomal dominant; occurs in childhood and regresses spontaneously in puberty; long vellus hair on extensor surface of the distal third of the upper arms and proximal third of the forearms

**Ocular:** facial dysmorphias

**Clinical:** Mental retardation; short stature


Hypertrrophic Cardiomyopathy Syndrome 613

**General:** Autosomal recessive; four major findings are congenital cataracts, hypertrophic cardiomyopathy, mitochondrial myopathy of voluntary muscles, and exercise-related lactic acidosis; premature death.

**Ocular:** Congenital cataracts; nystagmus; myopia; strabismus; myopic fundus findings.

**Clinical:** Hypertrophic cardiomyopathy; mitochondrial myopathy of voluntary muscles; exercise-related lactic acidosis.


Hypertrophic Neuropathy 614

**General:** Autosomal recessive.

**Ocular:** Cataract.

**Clinical:** Elevated spinal fluid protein; severe distal sensory and motor loss.


Hypervitaminosis A 615

**General:** Excessive vitamin A ingestion.

**Ocular:** Papilledema; congenital cataract; congenital anophthalmos; night blindness; diplopia; exophthalmos; "hourglass" cornea and iris with reduplicated lens.

**Clinical:** Elevation of cerebrospinal fluid pressure; migratory polyarthritis; hepatosplenomegaly; skin changes.


Hypervitaminosis D 616

**General:** Excessive vitamin D ingestion.

**Ocular:** Band keratopathy; epicantal folds; osteosclerosis of orbital bones; nystagmus; papilledema;iritis; cataract; sluggish pupillary reaction.

**Clinical:** Hypercalcemia; calcium deposition in body tissues.


Hypocalcemia

**General:** Serum calcium level depressed; secondary hypocalcemia can result following foscarnet treatment for cytomegalovirus retinitis in patients with acquired immunodeficiency syndrome.

**Ocular:** Conjunctivitis; blepharitis; blepharospasm; madarosis; ptosis; cataract; papilledema; strabismus.

**Clinical:** Chronic renal failure; hypoparathyroidism; hypoproteinemia; hypomagnesemia; malabsorption; acute pancreatitis; osteoelastic metastases; rickets; osteomalacia; medullary carcinoma of the thyroid; neuromuscular abnormalities.


Hypogonadism-Cataract Syndrome

**General:** Autosomal recessive.

**Ocular:** Cataracts.

**Clinical:** Elevated follicle-stimulating hormone levels; myotonic dystrophy; infertility.


Hypolipidemia Syndrome (Hooft Syndrome)

**General:** Autosomal recessive inheritance; disorder of tryptophan metabolism; normal glycolysis disturbed, which in turn interferes with normal fat synthesis; low levels of serum phospholipids, present from birth.

**Ocular:** Tapetoretinal degeneration of posterior pole with irregular, small areas of grayish-yellow discoloration scattered over posterior aspect of the retina; increased shiny reflection from the macular region, with "wet" and shiny appearance.

**Clinical:** Mental retardation; disturbance in normal growth; anomalies of hair, nails, and teeth; erythematous squamous skin rash (mainly face, arms, and legs).


Hypomelanosis of Ito Syndrome (Incontinentia Pigmenti Achromians; Systematized Achromic Nevus)

**General:** Probable autosomal dominant transmission; cutaneous abnormality consisting of bizarre, patterned, macular hypopigmentation over variable portions of the body with multiple associated defects in other body systems; abnormal chromosome constitutions.

**Ocular:** Iridal heterochromia; myopia; esotropia; microphthalmia; hypertelorism; nystagmus; strabismus; corneal opacity; choroidal atrophy; exotropia; small optic nerve; hypopigmentation of the fundus; corneal asymmetry; pannus; atrophic irides with irregular pupillary margins; cataract; retinal detachment.

**Clinical:** Cutaneous manifestations consisting of macular hypopigmented whorls, streaks, and patches in a bilateral or unilateral distribution affecting almost any portion of the body surface; 50% have associated noncutaneous abnormalities, including central nervous system dysfunction (seizure, delayed development) and musculoskeletal anomalies.


**Hypoparathyroidism** 621

**General:** Deficient secretion of parathyroid hormone.

**Ocular:** Keratitis; blepharospasm; ptosis; cataract; madarosis; optic neuritis; papilledema; conjunctivitis; myopia; ocular colobomata.

**Clinical:** Decreased blood calcium; increased serum phosphate; tetany; muscle cramps; stridor; carpopedal spasms; convulsions; lethargy; personality changes; mental retardation; intracranial calcification; choreoathetosis; hemiballismus; renal agenesis.


**Hypophosphatasia (Phosphoethanolaminuria)** 622

**General:** Inborn error of metabolism that entails increased urinary excretion of phosphoethanolamine and associated low alkaline phosphatase and hypercalcemia; prevalent in females; may result from absence or abnormal circulating factor regulating expression of alkaline phosphatase.

**Ocular:** Papilledema; optic atrophy; exophthalmos; blue sclera; conjunctival calcification; lid retraction; cataract; corneal subepithelial calcifications.

**Clinical:** Defect in true bone formation associated with widespread skeletal abnormalities; low serum alkaline phosphatase activity; hypercalcemia; nausea; vomiting; bowing of legs; convulsions; premature loss of teeth.


**Hypoproteinemia Syndrome (plurideficiency Syndrome; Kwashiorkor Syndrome; Malnutrition Syndrome)** 623

**General:** Manifested in children aged 4 to 5 years; widespread in underdeveloped countries; lack of adequate intake of protein; good prognosis with adequate diet therapy, otherwise fatal outcome; pseudomotor cerebri.

**Ocular:** Dullness of cornea and conjunctiva; thick, sticky, and foamy conjunctival excretion; corneal infiltrations and cloudiness; corneal ulcers (minimally inflammatory); keratomalacia possible in prolonged cases without therapy, eventually leading to panophthalmitis.

**Clinical:** Muscle atrophy; generalized edema; anorexia; vomiting; diarrhea; hepatosplenomegaly; dermatitis with desquamation; pigmentation and dyspigmentation; irritability; apathy; changes in hair color; hair becomes straight; hepatomegaly; failure to grow; weak cry.


**Hypothalamique Carrefour Syndrome (Carrefour Hypothalamique Syndrome)** 624

**General:** Onset is sudden with hemiplegia; etiology unknown.

**Ocular:** Visual loss.

**Clinical:** Hypertension; hemianesthesia; apraxia; astereognosis; asynergias.
Hypothermal Injury (Cryoinjury; Frostbite)  625

General: Loss of body heat to the point of local cold injury or freezing of tissue.

Ocular: Localized cryoinjury that can cause choroidal atrophy, retinal hemorrhages, hyperpigmentation of retina, uveitis, corneal edema, neovascularization of cornea, ectropion, lid edema, madarosis, pseudoepitheliomatous hyperplasia, iris atrophy, and paresis of extraocular muscles.

Clinical: Vesicles and blebs of affected tissue, especially ears, fingers, toes, and nose; contractures; dry gangrene of affected tissues.


Hypothermia with juvenile macular dystrophy syndrome (HJMD)  626

General: Rare; autosomal recessive.

Ocular: Macular dystrophy; hypotrichosis; vitreous hemorrhage in newborns

Clinical: Early hair loss


Hypovitaminosis A (Xerophthalmia)  627

General: Deficient serum levels of vitamin A; principal cause of infantile blindness in the world; due to insufficient intake of vitamin A or interference with its absorption from the intestinal tract; transport or storage in the liver; obstruction of biliary tract or pancreatic ducts.

Ocular: Bitot spot; xerosis; keratomalacia; keratitis; corneal perforation and ulcer; corneal opacity; hyperkeratosis; retinal degeneration; scotoma.

Clinical: Inadequate dietary intake or interference with absorptive storage or transport capacities, as occurs in liver disease, sprue, regional enteritis, and chronic gastroenteritis; respiratory infection; diarrhea; reduced childhood mortality.


Hysteria (Malingering; Ophthalmic Flake Syndrome)  628

General: Willful or unwillful exaggeration or simulation of symptoms of an illness without physiologic cause; frequently secondary to a state of anxiety; may be seen more in children; physical or sexual abuse may be a predisposing factor.

Ocular: Anxiety-induced angiospastic or central serous retinopathy; self-induced conjunctivitis; traumatic epithelial erosions; herpetic keratitis; angioneurotic edema; contact dermatitis; ptosis; recurrent herpetic vesicles; anisocoria; peculiar pupillary reflexes; accommodative spasm; amaurosis fugax; anxiety-induced optic neuritis; disturbance of conjugate movement; dyschromatopsia; facial tic; hypersecretion glaucoma; increased or decreased tear secretion; night blindness; nyctagmus; photophobia; strabismus; visual loss; psychogenic amaurosis with headaches.

Clinical: Aphonya; deafness; paralysis of limb; hemiplegia; dissociative state; anxiety; insomnia; tachycardia; shortness of breath; fatigue; vertigo, chest pains.

Impetigo

**General:** Superficial primary pyoderma caused by streptococci and *Staphylococcus aureus*.

**Ocular:** Pustular, crusting lesions of lids and brows; conjunctivitis; corneal ulcer; cicatricial ankyloblepharon.

**Clinical:** Thin-roofed vesicles that develop a thin amber crust occur on face and exposed areas of the extremities; extremely common skin infections caused by *S. aureus* in patients infected with human immunodeficiency virus (HIV).


Incipient Prechiasmal Optic Nerve Compression Syndrome

**General:** Caused by an expansive prechiasmal tumor or other lesion slowly compressing the optic nerve; frequently unilateral.

**Ocular:** Gradually progressive dimming of vision with near-normal acuity; reduced color perception (dyschromatopsia); subtle monocular field defects (progressive); positive Marcus Gunn pupillary sign (afferent pupillary light defect); optic nerve atrophy, depending on duration of compression before removal of the lesion; central or hemicentral scotoma.


Infantile Neuroaxonal Dystrophy (Seitelberger Disease [2]; Spastic Amaurotic Axonal Idiocy)

**General:** Axonal disease; occurs chiefly in female infants, less frequently in older children; etiology possibly vitamin E deficiency; autosomal recessive; selective axonal degeneration in the retina.

**Ocular:** Nystagmus; blindness; degeneration of optic pathways and long tracts.

**Clinical:** Muscular hypotonia; decreased pain sense; arrest of development in late infancy; areflexia; atonic bladder; dementia; spasticity; ataxia; spread of axonal lesions in the posterior gray horns of spinal cord, restiform bodies, and tegmentum of the lower brainstem; atrophy of the cerebral cortex; degeneration of the caudate nucleus and putamen with accumulation of lipids in these nuclei and in the globus pallidus may be associated; accumulation of iron-containing pigment in the globus pallidus and putamen occurs.


Infantile Type of Neuronal Ceroid Lipofuscinosis (Ceroid Lipofuscinosis; Hagberg-Santavuori Syndrome; Haltia-Santavuori Syndrome; Santavuori-Haltia Syndrome)

**General:** Age of onset 8 to 18 months; autosomal recessive; widespread loss of photoreceptor function; cerebral deterioration; death occurs at age 5 to 9 years (see Ceroid Lipofuscinosis).
Ocular: Visual failure simultaneously with or before neurologic signs; blindness; brownish macula and other signs of macular degeneration with narrow retinal vessels; atrophic optic disk; hypopigmented dystrophic peripheral retina without pigments; nystagmus; exotropia; formed and unformed visual hallucinations.

Clinical: Psychomotor deterioration; generalized muscular hypotonia; ataxia; myoclonic jerks; "knitting" hyperkinesia; and microcephaly; convulsions rare.


Infectious Mononucleosis (Mononucleosis; Epstein-Barr Virus, Acute; Acute Epstein-Barr Virus)

General: Asymptomatic in childhood; manifested in late adolescence of early adulthood; associated with Burkitt lymphoma and nasopharyngeal carcinoma.

Ocular: Conjunctivitis; ptosis; hippus; dacryocystitis; episcleritis; hemianopsia; nystagmus; retinal and subconjunctival hemorrhages; optic neuritis; orbital edema; scotoma; paralysis of extraocular muscles; uveitis; peripheral choroiditis; keratitis; papilledema; scleritis; retrobulbar neuritis, Sjögren syndrome; retinitis, choroiditis.

Clinical: Fever; widespread lymphadenopathy; pharyngitis; hepatic involvement; presence of atypical lymphocytes and heterophile antibodies in the blood; fatigue.


Influenza

General: Acute respiratory infection of specific viral etiology.

Ocular: Conjunctivitis; subconjunctival hemorrhages; keratitis; tenonitis; ptosis; cellulitis of orbit and lid; dacryocystitis; retinal hemorrhage; cataract; episcleritis; hypopyon; optic neuritis; uveitis; panophthalmitis; vitreal hemorrhage; paralysis of third or fourth nerve; uveitis following vaccination for influenza.

Clinical: Headache; fever; malaise; muscular aching; substernal soreness; nasal stuffiness; nausea.


Inverted Y Syndrome

General: Etiology unknown.

Ocular: Partial A syndrome with additional deviation occurring only in downgaze; occurs primarily with exodeviation.

Clinical: None.


Iridal Adhesion Syndrome (Iris Adhesion Syndrome, Iridocorneal Endothelial Syndrome)

General: Surgically related phenomenon following intraocular surgery in which iris pigment epithelium proliferates and adheres to cut edge of anterior capsule, drawing iris posteriorly to posterior and anterior capsules.
**Ocular:** Posterior synechiae; irregular pupil.

**Clinical:** None.


**Iridogonodysgenesis (Iris Hypoplasia With Glaucoma)**

**General:** Autosomal dominant; similar to Rieger syndrome or hereditary juvenile glaucoma.

**Ocular:** Stroma of iris hypoplastic; light irides; congenital glaucoma; optic atrophy; microphthalmia; opacities in Descemet membrane.

**Clinical:** None.


**Iris Dysplasia Hypertelorism-Psychomotor Retardation Syndrome**

**General:** Autosomal dominant inheritance; some features in common with Rieger syndrome.

**Ocular:** Hypertelorism; telecanthus; hypoplasia of the iris stroma; abnormally prominent Schwalbe line; synechiae between iris and cornea; pear-shaped pupils.

**Clinical:** Bilateral or unilateral hip dislocation; facial anomalies; psychomotor retardation; hypotonia and hyperlaxity of joints.


**Iris Neovascularization with Pseudoexfoliation Syndrome**

**General:** Anoxia secondary to iris vessel obstruction; electron microscopic studies reveal endothelial thickening with decreased lumen size and fenestration of vessel walls.

**Ocular:** Material found on posterior and anterior iris surface, anterior lens surface, ciliary processes, zonules, and anterior hyaloid membranes; neovascularization of iris stroma; increased permeability of iris vessels.


**Iris Pigment Layer Cleavage**

**General:** Autosomal dominant; cleavage of pigment of iris and ciliary body.

**Ocular:** Cataracts; reduced sagittal and spherical lens diameters; glaucoma; retinal detachment; microphakia; spherophakia.

**Clinical:** None.

Iris Retraction Syndrome (Posterior Synechiae and Iris Retraction Syndrome) 641

**General:** Rhegmatogenous retinal detachment, hypotony, and retrodisplacement of the iris with seclusion of the pupil, often associated with ciliochoroidal detachment, inflammation, and posterior vitreous retraction; caused by lowering of pressure behind iris partially due to posterior removal of fluid from subretinal space.

**Ocular:** Retinal detachment; hypotony; iris retraction; angle closure glaucoma; iris bombs; cataract; vitreous retraction; seclusion of the pupil; following intraocular surgery.

**Clinical:** None.


Iris Nevus Syndrome (Cogan-Reese Syndrome; Chandler Syndrome; Iridocorneal Endothelial Syndrome; ICE Syndrome) 642

**General:** Usually unilateral but may be bilateral; usually in young adult women; nonfamilial; cause unknown; Chandler, Cogan-Reese, and iridocorneal endothelial syndromes have been considered three separate syndromes but are now recognized as a single spectrum of diseases.

**Ocular:** Unilateral glaucoma in eyes with peripheral anterior synechiae; multiple iris nodules; ectopic Descemet membrane; corneal edema; stromal iris atrophy; iris pigment epithelial atrophy; ectropion uveae; ectopic pupil; keratoconus; herpes simplex virus deoxyribonucleic acid (DNA) has been detected in patients with iridocorneal endothelial syndrome from corneal specimens.

**Clinical:** Glasslike membrane covering the anterior iris surface; corneal endothelial degeneration and accompanying ectopic endothelial membranes are responsible for occlusion of the filtration meshwork and subsequent pressure increase.


Iron Deficiency Anemia (Microcytic Hypochromic Anemia) 643

**General:** Conjunctival pallor; small retinal hemorrhages; diplopia; visual field defects; fixed and dilated pupil; swelling or pallor of optic disk; retrobulbar neuritis; optic atrophy; it represents a state of inadequate body stores of iron; diagnosis of iron deficiency anemia is the *sine qua non* of chronic iron loss; most usual source is the gastrointestinal tract.

**Clinical:** Lethargy; increased susceptibility to infections; lymphadenopathy; hepatomegaly; splenomegaly.


Irvine Syndrome (Irvine-Gass Syndrome; Hruby-Irvine-Gass Syndrome) 644

**General:** Failing vision after uneventful cataract extraction; caused by spontaneous rupture of vitreous face with vitreous adhesions to the wound followed by macular edema; no gender or race preference; more common in older adults; positive association with other systemic vascular disease; may follow Nd:YAG laser posterior capsulotomy.
Ocular: Failing improvement or decrease in visual acuity after cataract surgery, due to macular changes; permanent impairment if the condition is not restored before secondary degenerative changes occur; vitreous opacities; rupture of the hyaloid face (spontaneous); vitreous adhesions to the wound; cystoid macular edema; papilledema; optic atrophy.

Clinical: None.


### Ischemic Orbital Compartment Syndrome

<table>
<thead>
<tr>
<th>General</th>
<th>Associated with spine surgery in the prone position</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Proptosis; elevated intraocular pressure; decreased vision; periorbital pain; ischemic optic atrophy</td>
</tr>
<tr>
<td>Clinical</td>
<td>Facial swelling; spinal stenosis or other diagnosis that would require long surgical procedures on the prone position</td>
</tr>
</tbody>
</table>


### Isotretinoin Teratogen Syndrome

<table>
<thead>
<tr>
<th>General</th>
<th>Maternal ingestion of isotretinoin (Accutane) during early pregnancy.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Small palpebral fissures; deep orbits; systemic treatment with isotretinoin has been associated with blepharitis and conjunctivitis.</td>
</tr>
<tr>
<td>Clinical</td>
<td>Prominent forehead; low-set, small, undifferentiated ears; depressed nasal bridge; small chin.</td>
</tr>
</tbody>
</table>


### IVIC Syndrome (Radial Ray Defects, Hearing Impairment, Internal Ophthalmoplegia, Thrombocytopenia)

<table>
<thead>
<tr>
<th>General</th>
<th>Autosomal dominant; Institute Venezolano de Investigacionas Cientificas (IVIC); observed in 1800s from Canary Islands in Venezuela; has been observed in descendants of a family that migrated to Venezuela from the Canary Islands.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ocular</td>
<td>Strabismus; internal ophthalmoplegia.</td>
</tr>
<tr>
<td>Clinical</td>
<td>Malformed upper limb; short distal phalanx; hearing loss; thrombocytopenia; leukocytosis; imperforate anus; radial ray defect.</td>
</tr>
</tbody>
</table>


### Jabs Syndrome (Synovitis, Granulomatous Uveitis, and Cranial Neuropathies)

<table>
<thead>
<tr>
<th>General</th>
<th>Autosomal dominant.</th>
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<tbody>
<tr>
<td>Ocular</td>
<td>Granulomatous uveitis; iritis; sixth nerve palsy.</td>
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</tbody>
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**Clinical**: Granulomatous synovitis; corticosteroid responsive hearing loss; boggy polynovitis; boutonneuse deformities; granulomatous arthritis; skin involvement; fever; hypertension; large-vessel vasculitis.


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**Jadassohn-Lewandowsky Syndrome (Pachyonychia Congenita)**

**General**: Autosomal dominant inheritance; three variants: type I has symmetric keratoses of hands and feet and follicular keratosis of body; type II same as type I, plus leukokeratosis; type III same as type I with corneal changes; gene for this disorder has been found to be closely linked to the keratin gene cluster on 17q12-q21; disorder usually develops in early infancy.

**Ocular**: Dyskeratosis of the cornea; bilateral cataract.

**Clinical**: Keratosis and hyperhidrosis of palms and soles, whereas the remaining skin is usually rather dry; bullous lesions may occur with secondary infections, mainly during warm seasons; leukokeratosis of oral mucosa (mainly tongue); follicular keratosis; congenital pachyonychia (nails not only may be thickened but also may be frequently inflamed and lost with aggravation at sites of regrowth); hoarse voice; epidermoid cysts; oral leukokeratosis.


**Jansen Disease (Metaphyseal Dysostosis)** 653

**General:** Affects both sexes; etiology unknown; autosomal dominant.

**Ocular:** Exopthalmos.

**Clinical:** Mental retardation; muscular atrophy; flat nose; large mouth; dwarfism; deafness; metaphyses of all bones affected with marked widening.


**Japanese River Fever (Mite-Borne Typhus; Rural Typhus; Scrap Typhus; Tropical Typhus; Tsutsugamushi Disease; Typhus)** 654

**General:** Acute febrile illness by *Rickettsia tsutsugamushi* transmitted by the larval form of a mite.

**Ocular:** Keratitis; uveitis; paracentral scotoma; vitreous opacity; nystagmus; retinal hemorrhages; exudates; edema.

**Clinical:** Chills; fever; malaise; headache; lymphadenopathy; generalized aching.


**Jensen Disease (Juxtapapillary Retinopathy)** 655

**General:** Etiology unknown.

**Ocular:** Circumscribed inflammatory changes of the choroid; field defect.

**Clinical:** None.


**Jeune Disease (Asphyxiating Thoracic Dystrophy; Thoracic-Pelvic-Phalangeal Dystrophy)** 656

**General:** Autosomal recessive; similar to Ellis-van Creveld syndrome; positive associations of this disorder with cystinuria has been reported in two sisters.

**Ocular:** Retinal dysfunction; granular pigmentation of the choroid; nystagmus; small white patches in peripheral fundus; retinal degeneration; coloboma of iris; eyes symmetrically involved; retinal aplasia; photophobia; strabismus; pigmentary retinopathy.

**Clinical:** Long, narrow thorax; short anteriorly clubbed ribs forming a continuous tube with the abdominal cavity; dwarfing skeletal dysplasia; progressive renal failure; liver abnormalities; severe respiratory insufficiency; long, narrow trunk; dystrophic rib cage with respiratory distress; short limbs; polydactyly.


Johnson Syndrome (Adherence Syndrome; Adherent Lateral Rectus Syndrome)  657

**General:** Congenital delayed development; most frequent in children below age 3 years; spontaneous disappearance possible; two principal types of disturbances: (i) adhesions between sheaths of external rectus and inferior oblique with resulting limits in abduction; (ii) adhesions between sheaths of superior rectus and superior oblique with resulting limits in elevation.

**Ocular:** Forced muscle duction test may prove presence or absence of adherence versus paralysis.

**Clinical:** None.


Jones Syndrome (Cherubism; Mandibular Cystic Dysplasia)  658

**General:** Etiology unknown; both sexes affected; present at birth.

**Ocular:** White line beneath the iris on sclera; hypertelorism; optic atrophy.

**Clinical:** Rounded cheeks; jaw fullness; submandibular region swelling; narrow, V-shaped palate.


Joubert Syndrome (Familial Cerebellar Vermis Agenesis)  659

**General:** Autosomal recessive; both sexes affected; onset in early infancy.

**Ocular:** Choroidal coloboma; nystagmus; ocular fibrosis, telecanthus.

**Clinical:** Episodic hyperpnea; apnea; ataxia; psychomotor retardation; rhythmic protrusion of tongue; mental retardation; micrognathia; complex cardiac malformation; cutaneous dimples over wrists and elbows.


Jugular Foramen Syndrome (Vernet Syndrome)  660

**General:** Injuries, aneurysms, and tumors (more commonly due to metastatic lesion than primary neoplasms) affecting the foramen jugulare are the primary causes for the syndrome to develop; if sympathetic fibers surrounding the carotid artery are involved, this will produce Homer triad; note similarity of clinical findings of Villaret syndrome or "retroparotid space syndrome," which may include epiphora and lagophthalmos and in which cranial nerves IX to XII and the cervical sympathetics are involved.

**Ocular:** Enophthalmos; ptosis; miosis.

**Clinical:** Paralysis of the ninth, tenth, and eleventh cranial nerves with resulting impairment of related function, that is, dysphagia, loss of taste on the posterior third of the tongue, and nasal regurgitation; anhidrosis; paralysis of the sternocleidomastoid muscle and part of the trapezium (upper portion); hoarseness; tachycardia; dysarthria; weight loss.


Junius-Kuhnt Syndrome (Kuhnt-Junius Disease; Macular Senile Disciform Degeneration [I]; Macula Lutea Juvenile Degeneration [2])

**General:** Onset in advanced age or in juvenile period; etiology unknown; possible autosomal dominant or recessive inheritance.

**Ocular:** Impairment of central vision; central scotoma; atrophic macular degeneration surrounded by retinal hemorrhages, resulting in mountlike lesion; exudative and atrophic reaction with deposit in and about macula.

**Clinical:** None.


Juvenile Diabetes-Dwarfism-Obesity Syndrome (Mauriac Syndrome; Dwarfism-Hepatomegaly-Obesity-Juvenile Diabetes Syndrome)

**General:** Etiology is obscure, although nutritional deficiencies, metabolic disorders, and deficiency in insulin have been considered; develops slowly, with slow growth and difficulties in management of diabetic condition.

**Ocular:** Cataract; diabetic retinopathy with retinal hemorrhages, exudates, microaneurysms, neovascularization, vaso and glial proliferation (grades I to IV diabetic retinopathy; hypertensive retinopathy); occasional optic neuritis.

**Clinical:** Hepatomegaly; diminished growth; osteoporosis; hypertension; arteriosclerosis; obesity (with moon face); juvenile diabetes; abdominal colic.


Juvenile Rheumatoid Arthritis (JRA; Still Disease)

**General:** Onset before age 16 years; greater occurrence of systemic manifestations, monarticular and oligoarticular joint involvement, and iridocyclitis.

**Ocular:** Hypopyon; band keratopathy; uveitis; cataract; papillitis; glaucoma; macular edema; ocular pain; vitreous cells; synechiae; scleritis; presumed to have an autoimmune etiology; antiocular antibodies, including iris protein antibodies, have been found in the sera of patients.

**Clinical:** Salmon pink macular rash; arthritis; hepatosplenomegaly; leukocytosis; chronic pain; joint swelling; low-grade fever; anemia; rheumatoid nodules.


Juvenile Xanthogranuloma (JXG; Nevoxanthoendothelioma)

**General:** Childhood disease; unknown etiology.

**Ocular:** Uveal tract tumor presenting as spontaneous hyphema; secondary glaucoma; uveitis; corneal, lid, and epibulbar tumors; proptosis; retinal and choroidal lesions (rare).
**Clinical**: Multiple benign tumors, primarily of the skin; usually appear in the first 3 years of life; lesions appear as yellow-to-brown papules or nodules.


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**Kabuki Makeup Syndrome (Niikawa-Kuroki Syndrome)**

**General**: Etiology unknown; originally termed Kabuki makeup syndrome because dysmorphic facies resembled the stylized makeup worn by Kabuki actors; also seen in ethnic groups other than Japanese.

**Ocular**: Ectropion of lower eyelid; long palpebral fissures; sparse lateral half of eyebrows; highly arched eyebrows.

**Clinical**: Prominent ears; cleft or highly arched palates; brachydactyly; dermatoglyphics; padlike swelling of fingertips; short stature; mental retardation; susceptibility to infection; characteristic facies; developmental delay; mental and growth retardation with specific craniofacial malformations including a depressed nasal tip; musculoskeletal abnormalities, evolving phenotype over time suggesting an underlying defect of the connective tissue.


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**Kahler Disease (Myelomatosis; Multiple Myeloma)**

**General**: Disseminated malignancy of plasma cells located predominantly in the bone marrow.

**Ocular**: Tumor of orbit common, with proptosis or displacement of globe; conjunctival sludging and segmentation; crystalline deposits of cornea and conjunctiva; cotton-wool spots; retrobulbar neuritis; occlusion of central retinal artery and vein; palsy of sixth nerve; vitreous hemorrhage; dilated veins and hemorrhages; retinal microaneurysms; choroidal detachment; amaurosis fugax; myeloma infiltrates in orbit, iris, choroid, retina, sclera, and optic nerve; corneal opacities; ciliary body cysts; iritis; glaucoma; subluxation of lens; papilledema; corneal edema; cavernous sinus syndrome; bilateral superficial punctate keratitis; central retinal vein occlusion; crystalline keratopathy in a vortex distribution; spontaneous endocapsular hematoma.

**Clinical**: Bone pain; fractures; dehiscence; hypercalcemia; hyperuricemia; proteinuria; inclination to infection; hyperviscosity.


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**Kallmann Syndrome (Hypogonadotropic Hypogonadism-Anosmia Syndrome)**

**General**: Disorder of hypothalamic function involving the control of releasing factors, with hypogonadism and anosmia as the clinical signs; agenesis of olfactory bulbs; midline cranial anomalies (cleft lip, cleft palate, imperfect fusion); autosomal recessive phenotype.
**Ocular:** Color blindness (variable occurrence).

**Clinical:** Failure of sexual maturation; decrease in primary and secondary sex characteristics; loss of smell; hypertension; mental retardation; schizophrenia.


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**Kaposi Disease (Kaposi Sarcoma; Kaposi Hemorrhagic Sarcoma; Multiple Idiopathic Hemorrhagic Sarcoma; Kaposi Varicelliform Eruption)**

**General:** Vascular tumor of unknown cause; seen most often in males, Jews, and those from eastern Europe, the southern Mediterranean, and Africa; human immunodeficiency virus-related Kaposi syndrome is the most common type of cancer seen in acquired immunodeficiency syndrome patients.

**Ocular:** Ocular adnexa, varicelliform eruption, including lids, conjunctivae, lacrimal glands, and orbit, may be involved; hemorrhage; extensive injection and thickening of conjunctival tissues; conjunctival involvement more evident in bulbar conjunctiva.

**Clinical:** Vascular sarcomas usually occur on the legs, although widespread cutaneous and visceral tumors may develop; secondary malignancies are very common; lymphedema.


Volberding P. Therapy of Kaposi's sarcoma in AIDS. *Semin Oncol* 1984; 11:60.

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**Karsch-Neugebauer Syndrome (Nystagmus-Split Hand Syndrome)**

**General:** Autosomal dominant.

**Ocular:** Horizontal nystagmus; strabismus; cataract; fundus changes.

**Clinical:** Split hand and split foot deformities; monodactylous hands.


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**Kartagener Syndrome (Sinusitis-Bronchiectasis-Situs Inversus Syndrome; Bronchiectasis-Dextrocardia-Sinusitis; Kartagener Triad)**

**General:** Autosomal recessive; onset in early infancy; occasionally dominant; finding of various structural defects in patients with this condition suggests that there are several genetic determinants.

**Ocular:** Myopia; glaucoma; conjunctival melanosis; iris coloboma; tortuous and dilated retinal vessels; retinal pigmentary degeneration; pseudopapillitis.

**Clinical:** Immotile cilia; situs inversus; bronchiectasis; sinusitis; various cardiovascular and renal abnormalities; dyspnea; productive cough; recurrent respiratory infections; palpitation; otitis media; nasal speech; conductive hearing loss; nasal polyps; situs inversus viscercum with hepatic dullness on left side.


Kasabach-Merritt Syndrome (Capillary Angioma-Thrombocytopenia; Hemangioma-Thrombocytopenia; Thrombocytopenia Purpura-Hemangioma)  

**General:** Angioma causing sequestration of platelets and platelet deficiency.

**Ocular:** Capillary hemangiomas of the orbit; retinal detachments.

**Clinical:** Extraorbital hemangiomas found on trunk, extremities, and palate or in subglottic space; thrombocytopenia; found in infants; purpura and bleeding.


**Kaufman Oculocerebrofacial Syndrome**  

**General:** Autosomal recessive; significant positive and negative features.

**Ocular:** Hypertelorism; epicanthal folds; ptosis; mongoloid obliquity; microcornea; pale optic disk; laterally broad eyebrows sparse.

**Clinical:** Flat philtrum; congenital hypotonia; micrognathia; respiratory distress; high narrow palate; lordosis; constipation; flat feet.


**Kearns-Sayre Syndrome (Ophthalmoplegia Plus Syndrome; Kearns-Shy Syndrome; Kearns Disease)**  

**General:** Etiology unknown; sporadic (nonhereditary); onset before age 20 years; external ophthalmoplegia; complete heart block.

**Ocular:** Pigmentary degeneration of retina; progressive external ophthalmoplegia; corneal decompensation; optic neuritis.

**Clinical:** Abnormal mitochondria with paracrystalline inclusion in muscle cell; heart block; limb weakness; hyperglycemic acidotic coma; death; cerebellar dysfunction.


**Kenny Syndrome**  

**General:** Congenital syndrome.

**Ocular:** Nanophthalmos with hyperopia; papilledema; vascular tortuosity; macular crowding; bilateral optic atrophy.

**Clinical:** Dwarfism; thickened long bone cortex; transient hypocalcemia.


**Keratitis Fugax Hereditarla**

**General:** Autosomal dominant; onset from age 4 to 12 years; characterized by acute attacks of keratitis.

**Ocular:** Keratitis; corneal opacities.

**Clinical:** None.


**Keratoconus Posticus Circumscriptus (KPC; KPC with Associated Malformations)**

**General:** Autosomal recessive; rare; abnormality in corneal curvature centrally localized on its posterior surface in association with opacification of the overlying stroma; may be an anterior chamber cleavage defect with failure of normal separation of the lens and iris from the cornea.

**Ocular:** Corneal opacities; retinal coloboma; ptosis; hyperopia; iridocorneal adhesions; hypertelorism.

**Clinical:** Cleft lip; cleft palate; neck webbing; short stature; mental retardation; inguinal hernia; undescended testes; tight heel cords; vertebral anomalies; delayed bone age; double ureters; cone-shaped epiphyses; stubby limbs and digits; limitation of extension and supination of the elbows; brachydactyly; fifth finger clinodactyly; frequent urinary tract infections; prominent nose; mild maxillary hypoplasia; low posterior hairline; short, broad feet with bilateral pes cavus; bilateral ureteric reflux.


**Keratodermia Palmaris Et Plantaris (Palmoplantar Keratodermia; Keratosis Palmoplantaris)**

**General:** Autosomal recessive; hereditary disorder; diffuse or focal thickening of the palms and soles.

**Ocular:** Hyperkeratosis of lid and cornea; ectropion; leukemia; corneal ulceration; pronounced photophobia; hereditary optic atrophy; epiphora; conjunctivitis.

**Clinical:** Localized or disseminated hyperkeratotic changes of the palms and soles with a tendency toward fissure and secondary infection.

**Kiloh-Nevin Syndrome (Muscular Dystrophy of External Ocular Muscles; Ocular Myopathy)**

**General:** Etiology unknown; autosomal dominant.

**Ocular:** Ptosis; orbicularis muscle weakness; ocular myopathy; diplopia progressing to bilateral myopathic ophthalmoplegia; may be associated with pigmentary retinopathy and heart block (see Kearns-Sayre syndrome).

**Clinical:** Progressive muscular dystrophy in which facial muscles may be involved; occasionally, hereditary ataxia; pain; myokymia.


**Kimmelstiel-Wilson Syndrome (Diabetes Mellitus-Hypertension-Nephrosis Syndrome; Diabetes-Nephrosis Syndrome; Diabetic Glomerulosclerosis; Intercapillary Glomerulosclerosis; Renal Glomerulohyalinosis-Diabetic Syndrome)**

**General:** Occurs in patients with diabetes mellitus of several years' duration.

**Ocular:** Retinal lesions, including hemorrhages, exudates, and neovascularization.

**Clinical:** Hypertension; proteinuria; edema; glomerulonephrosis; arteriosclerosis; capillary or intercapillary glomerulosclerosis; eosinophilic nodules; hyaline degeneration of the renal arterioles.


**Kinsbourne Syndrome (Dancing Eyes Syndrome; Opsoclonus-Myoclonus Syndrome)**

**General:** Etiology unknown; occurs in infancy and early childhood; myoclonic encephalopathy; has been reported as the only manifestation of a postinfectious syndrome without evidence of encephalitis.

**Ocular:** Twitching of lids and eyebrows on occasion, more pronounced with activity than at rest; irregular vertical movements, jerky in appearance and sometimes with some lateral nystagmic components.

**Clinical:** Sporadic, jerky movements of head, trunk, and limbs, usually more pronounced when the child is active; lack of coordination; ataxia; irritability; mental retardation; chronic neurologic deficits.


**Kirk Syndrome**

**General:** Etiology unknown; raised hereditary masses over central cornea; rare, only one family known to be affected.

**Ocular:** Photophobia; excessive lacrimation; amyloid corneal deposits.

**Clinical:** None.


**Kjellin Syndrome**

**General:** Autosomal recessive disorder; degeneration progressive.

**Ocular:** Yellow retinal flecks that lie at the pigment epithelial level; poor visual perceptual skills; central retinal degeneration.

**Clinical:** Spastic paraparesis; dementia; progressive lower extremity weakness; dysarthric speech; muscle atrophy.


Kjellin K. Familial spastic paraplegia with myotrophy, oligophrenia, and central retinal degeneration. *Arch Neurol* 1959; 1: 133.


**Klein Syndrome**

**General:** Autosomal dominant; belongs to the group of iridodermatoauditive dysplasias.

**Ocular:** Hypertelorism; blepharoophimosis; hypertrichosis; blue irides; heterochromia.

**Clinical:** Bilateral labyrinthine deafness; mandibular retrognathism; skull deformities and arched palate; partial albinism of skin and hair; syndactylysm (cutaneous).

Klinefelter Syndrome (Gynecomastia-Aspermatogenesis Syndrome; XXY Syndrome; XXXY Syndrome; Reifenstein-Albright Syndrome) 684

**General**: Occurrence in 1% of retarded males; phenotypically males with positive female sex chromatin; karyotype shows 47 chromosomes, 44 autosomes, and 3 sex chromosomes with the complement XXY.

**Ocular**: Anophthalmos; coloboma; corneal opacities.

**Clinical**: Testicular hypoplasia; sterility; gynecomastia; eunuchoid physique; mental retardation; association with progressive systemic sclerosis and systemic lupus erythematosus.


Klippel-Feil Syndrome (Congenital Brevicollis; Synostosis of Cervical Vertebra) 685

**General**: Autosomal recessive inheritance; females more commonly affected; progressive paraplegia may develop late in life.

**Ocular**: Esotropia; hypertropia combined with torticollis; occasional horizontal nystagmus.

**Clinical**: Platybasia; congenital upward displacement of scapula (Sprengel deformity); brevicollis; immobility of neck (painless); low posterior hairline; peculiar facies; cleft palate; short stature; congenital brevicollis; vertebral abnormalities; autosomal dominant mode of inheritance has been reported.


Klippel- Trenaunay-Weber Syndrome (Parkes-Weber Syndrome; Angio-Osteo-Hypertrophy Syndrome) 686

**General**: Most frequently inherited as irregular dominant; however, reported to be recessive with parent consanguinity; association of Klippel-Trenaunay-Weber syndrome and Sturge-Weber syndrome has been reported.

**Ocular**: Enophthalmos; unilateral hydrophthalmos; conjunctival telangiectasia; atypical iris coloboma; cataract; irregular and dilated retinal vessels; choroidal angiomas; exudative outer retinal vascular masses.

**Clinical**: Vascular nevi; varicose vessels; capillary angiomas; lymphangioma; arteriovenous aneurysm; hypertrophy of soft tissues and bones (local); phlebitis; thrombosis; syndactyly; polydactyly; early eruption of teeth; hemifacial hypertrophy.


Kloeppfer Syndrome 687

**General**: Rare autosomal recessive disease; manifestations at age 2 months; death between ages 21 and 30 years.

**Ocular**: Progressive loss of vision to complete blindness associated with progressive dementia.

**Clinical**: Severe blistering in sunlight; no increase in weight and height after erythema subsides at age 5 to 6 years; mental age does not progress beyond the level of imbeciles; progressive degenerative dementia occurs during or immediately after adolescence.


Klüver-Bucy Syndrome (Temporal Lobectomy Behavior Syndrome)

General: Occurs after temporal lobectomy, carried out therapeutically for temporal lobe epilepsy.

Ocular: "Psychic blindness" or visual agnosia.

Clinical: Changes in emotional behavior (possible rage reactions); hypersexuality; bulimia (changes in dietary habits); loss of recognition of people; strong oral tendencies (i.e., licking, biting, chewing); deficiency of memory; psychic blindness; aberrant sexual behavior; hypermetamorphosis; aphasia; visual agnosia; memory deficit; speech disturbance; syndrome in adults is commonly associated with neurodegenerative conditions, following radiation therapy, or after temporal lobectomy; syndrome in children has been recognized almost exclusively in association with acute bitemporal injury or dysfunction.


Knies Dysplasia (Metatropic Dwarfism Type II)

General: Autosomal dominant disease; Swiss cheese pattern on cartilage biopsy specimen; due to either alteration metabolism of proteoglycans or abnormality in collagen synthesis; both sexes affected; collagen type II collagenopathy; produced by a single amino acid substitution in the type II collagen triple helix.

Ocular: Retinal detachment; severe myopia; cataracts; dislocated lenses; blepharoptosis; vitreoretinal degeneration; vitreous traction; congenital glaucoma; hypertelorism; mild synophrys; epicanthal folds; perivascular lattice degeneration, white without pressure.

Clinical: Severe short stature; typical facies with flat nasal bridge; cleft palate; hearing loss; joint contractures; lordosis; kyphosis.


Knobloch Syndrome

General: Autosomal recessive; retinal detachment with occipital encephalocele.

Ocular: High myopia; retinal detachment; vitreoretinal degeneration; persistent papillary membrane; posterior vitreous detachment; retinochoroidal staphylomas.

Clinical: Occipital encephalocele; normal intelligence; congenital midline scalp defect; unusual plantar creases.


Koby Syndrome (Floriform Cataract)

General: Autosomal dominant; both sexes affected.

Ocular: Multiple opacities of different shapes (annular, floriform, and polychromatic); found especially around embryonic nucleus.

Clinical: None.


Koerber-Salus-Elschnig Syndrome (Sylvian Aqueduct Syndrome; Nystagmus Retractorius Syndrome)  692

General: Caused by tumor or inflammation in region of aqueduct of Sylvius, third and fourth ventricle, or corpora quadrigemina.

Ocular: Lid retraction may be associated with midbrain lesions above the posterior commissure; paresis of vertical gaze; tonic spasm of convergence on attempted upward gaze; clonic convergence movements or convergence nystagmus; vertical nystagmus on gaze up or down; nystagmus retractorius with spasmodic retraction of the eyes when an attempt is made to move them in any direction; occasional extraocular muscle paresis.

Clinical: Headaches; dizziness; hypertension; possible hemiparesis; ataxia; hemitremor; Babinski’s sign.


Kohn-Romano Syndrome (BPES Syndrome)  693

General: Autosomal dominant transmittance; tetrad with telecanthus, ptosis, epicanthus inversus, and blepharophimosis; male preponderance; location of the abnormal gene responsible for this syndrome has been postulated to be a 3q2.

Ocular: Telecanthus; ptosis; epicanthus inversus; blepharophimoses; divergent strabismus; nystagmus; esotropia; anomalies of the lacrimal punctum; reduced corneal diameter.

Clinical: Highly arched palate; low-set ears with deformed pinnas.


Komoto Syndrome (Congenital Eyelid Tetrad; CET)  694

General: Autosomal dominant; all races affected; most patients are of normal intelligence.

Ocular: Ptosis; epicanthus inversus; blepharophimosis; telecanthus.

Krabbe (2) Syndrome (Sturge-Weber-Krabbe Syndrome; Galactocerebrosidase Deficiency)  695

General: Etiology unknown; some evidence of irregularly dominant transmission; variant of Sturge-Weber syndrome; both sexes affected; present from birth; appears that the GALC locus lies somewhere in the region 14q21-q31.

Ocular: Buphthalmos; conjunctival angiomas; choroidal angiomatosis; retinal aneurysm.

Clinical: Cerebral angiomas (possible calcium deposition); flat angioma of the skin in the distribution area of the trigeminal nerve (V) (nevus flammeus); mental deterioration due to progressive atrophy of the brain; contralateral hemiplegia; facial hemiatrophy.


### Krause Syndrome (Congenital Encephalo-Ophthalmic Dysplasia; Encephalo-Ophthalmic Syndrome) 696

**General:** No hereditary factors involved; no predilection for either sex; more frequent in premature infants; death frequently from intercurrent infections.

**Ocular:** Microphthalmos; enophthalmos; ptosis; strabismus; secondary glaucoma; iris atrophy; anterior and posterior synechiae; scleral atrophy; persistent remnants of hyaloids artery; intraocular hemorrhages and exudates; cyclitic membranes; cataracts; retinal hypoplasia and hyperplasia; choroidal and retinal malformation; retinal glial membranes; retinal detachment; choroidal atrophy; optic nerve malformation; optic atrophy.

**Clinical:** Congenital cerebral dysplasia; hydrocephalus or microcephaly; mental retardation; heterotopia.


### Kufs Disease (Adult Chronic GM2 Gangliosidosis; Gangliosidosis GM2 Adult Type; Hallervorden-Kufs Syndrome) 697

**General:** Autosomal recessive; hexosaminase A decrease; onset in the third and fourth decades of life; slow death within 19 or 20 years of onset; very rare type of adult neuronal ceroid lipofuscinosis; autosomal recessive and dominant inheritance have been reported.

**Ocular:** Retinal storage; ocular lesions usually are absent; rare macular discoloration; no pigmentary degeneration of the retina.

**Clinical:** Dementia and behavioral changes; progressive gait and postural deterioration; mild ataxia; dysarthria; ascending muscular atrophy; pes cavus; cerebral degeneration; apathy; progressive myoclonus epilepsy; aphasia; facial dyskinesias; lipid infiltration of the brain cells is the principal pathologic feature (see Ceroid Lipofuscinosis).


### Kugelberg-Welander Syndrome (Juvenile Muscular Atrophy) 698

**General:** Autosomal recessive; juvenile spinal muscular atrophy; affects both sexes; onset in late childhood or adolescence.

**Ocular:** Ptosis; ophthalmoplegia; exotropia; orbicularis oculi paresis.

**Clinical:** Slowly progressive proximal muscle atrophy; lower extremities usually are affected first, with the upper limbs being affected late; frequently, fasciculation; proximal muscle weakness, especially of the lower extremities; elevated serum creatine kinase levels.


Kuru Syndrome (Laughing Death)

**General:** Restricted to Fore tribe of eastern New Guinea; prevalent in children and adult women; etiology unknown, possibly related to the tribe's practice of cannibalism; uncertain whether significant genetic factors also are involved.

**Ocular:** Strabismus; nystagmus.


Kussmaul Disease (Kussmaul-Maier Disease; Necrotizing Angiitis; PAN; Periarteritis Nodosa; Polyarteritis Nodosa)

**General:** Progressive process of vascular inflammation and necrosis, manifested by numerous nodules along the course of small- and medium-sized arteries; lesions are segmental in distribution, have a predilection for bifurcation and involve all but the pulmonary arteries; arteries in gastrointestinal tract, kidneys, and muscles are particularly affected; affects primarily males between ages 20 and 50 years.

**Ocular:** Retinal detachment; cotton-wool patches; polyarteritis nodosa lesion of arteries; pseudoretinitis pigmentosa; conjunctivitis; corneal ulcer; tenonitis; ptosis; exophthalmos; uveitis; optic atrophy; cataract; scleritis; paralysis of extraocular muscles; neuroretinitis; anterior uveitis; macular star; peripheral ulcerative keratitis; retinal vasculitis; pseudotumor of the orbit; central retinal artery occlusion.

**Clinical:** Fever; myalgia; hypertension; gastrointestinal disorders; neuropathy; respiratory infection; weight loss; anginal pain; hemiplegia; convulsion; acute brain syndrome; skin lesions; diffuse erythema; purpura; urticaria; gangrene; tachycardia; pericarditis; aortitis; painful facial swelling; diplopia.


Kyrle Disease (Hyperkeratosis Follicularis and Parafollicularis in Cutem Penetrans; Hyperkeratosis Penetrans)

**General:** Etiology unknown; associated in siblings, suggesting heredofamilial condition; syndrome may belong in the diabetic syndromes; rare skin disorder; onset usually in the third to sixth decades of life.

**Ocular:** Subcapsular cataracts; corneal changes characterized as minute, yellow-brown subepithelial opacities, noted to be more dense and more deeply penetrating in the peripheral cornea than in the central cornea.

**Clinical:** Multiple flesh-colored, horny papules associated with hair follicles.

Lacrimal Duct Defect 702

General: Autosomal dominant.

Ocular: Imperforate nasolacrimal ducts with or without absence of puncta and canaliculi.

Clinical: None.


Lacrimal-Auriculo-Dento-Digital Syndrome (Ladd-Levy-Hollister Syndrome) 703

General: Autosomal dominant; all features have been reported as isolated traits.

Ocular: Aplasia or hypoplasia of puncta; obstruction of nasolacrimal ducts; bilateral lacrimal duct fistula; poor tear production; absent lacrimal puncta; chronic dacryocystitis; dry eyes; epiphora.

Clinical: Cup-shaped pinnas; mixed hearing deficit; small and peg-shaped lateral maxillary incisors; mild enamel dysplasia; fifth finger clinodactyly; duplication of the distal phalanx of the thumb; triphalangeal thumb; syndactyly; unilateral radial aplasia; poor saliva production; salivary gland hyposecretion; dental hypoplasia; dysplasia; cup-shaped ears with hearing loss; digital anomalies.


Langer-Giedion Syndrome (Trichorhinophalangeal Syndrome, Type II) 704

General: Rare congenital condition.

Ocular: Iris colobomata.

Clinical: Mental retardation, bulbous nose, sparse hair, cone-shaped epiphyses, microcephaly, multiple exostoses, redundant skin; less consistently, "floppy infants," hyperextensible joints, recurrent upper respiratory tract infections, delayed speech development, and characteristic facies.


Lanzieri Syndrome 705

General: Developmental anomaly that belongs to group of craniofacial malformations; present from birth.

Ocular: Microphthalmia; anophthalmos; iris coloboma; cataracts; retinal and choroidal coloboma; optic nerve coloboma.

Clinical: Dwarfism; dyscephalia; dental anomalies; hypertrichosis; skin atrophy; absence of fibula, some tarsal and metatarsal bones.


**Laron Syndrome**

**General:** Autosomal recessive; insulinlike growth factor I hormone resistance  
**Ocular:** Microphthalmia; reduced retinal vascularization; optic nerve hypoplasia; pseudopapilledema  
**Clinical:** Short stature; abnormally small extremities; subnormal head circumference, increased body fat and delayed sexual development  

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**Larsen Syndrome**

**General:** Etiology unknown; autosomal recessive; possibly dominant in some cases.  
**Ocular:** Hypertelorism; bilateral chronic keratitis; corneal neovascularization; lower lid entropion.  
**Clinical:** Frontal bossing; depressed nasal bridge; flat face; flat and broad thumbs; skeletal dysplasia with multiple joint dislocations; unusual faces; long, cylindrical fingers; spatulate thumbs; dental abnormalities; cardiac defects; hydrocephalus; laryngotraceomalacia; dislocation of the cervical spine; tracheomalacias; heart disease; severe respiratory infection; clubfeet; multiple joint deformities; hydrocephalus; tracheal stenosis.  

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**Lateral Sinus Thrombosis (Sigmoid Sinus Thrombosis)**

**General:** Predominant in children; acute onset secondary to chronic otitic infections; high mortality rate without treatment; any microorganism may be responsible for infection and secondary thrombosis; associated with oral contraceptive usage.  
**Ocular:** Pain behind the eye; muscle palsies; papilledema (50%).  
**Clinical:** Fever; headaches; nausea; vomiting; swelling over mastoid region; intracranial hypertension; seizures; hemiplegia.  

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**Lattice Degeneration and Retinal Detachment**

**General:** Autosomal dominant; progressive; lattice degeneration precedes retinal detachment by about 20 years; familial occurrence of lattice degeneration in nonmyopes has been reported.  
**Ocular:** Myopia; retinoschisis; peripheral retinal degeneration; lattice degeneration.  
**Clinical:** None.  

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**Laurence-Moon-Bardet-Biedl Syndrome (Bardet-Biedl Syndrome; Retinitis Pigmentosa-Polydactyly-Adiposogenital Syndrome)**

**General:** Recessive, dominant autosomal, and recessive sex-linked gene; male preponderance; onset in childhood; cases of Laurence-Moon belong to the group of heredoataxias.  
**Ocular:** Ptosis; epicanthus; nystagmus; strabismsus; night blindness; myopia; hypermetropia; iris coloboma; retinitis pigmentosa "bone corpuscles"; macular degeneration; attenuation of retinal vessels; choroidal atrophy; optic nerve atrophy; cataract; microphthalmia; keratoconus.
Clinical: Obesity (Fröhlich type); hypogenitalism; reduced intelligence and mental retardation; turriencephaly; shortness of stature; atresia ani; genu valgum; congenital heart disease; polydactyly; body hair scant or absent; pseudogynecomastia.


Laurence JZ, Moon RC. Four cases of “retinitis pigmentosa” in the same family and accompanied by general imperfections of development. *Ophthalmol Rev* 1866; 2:32.


Lead Poisoning

General: Now rare and mostly of industrial origin; cumulative poisoning; excreted slowly; absorption slow by any route; prolonged exposure required for development of symptoms; acute poisoning virtually nonexistent.

Ocular: Sclerosis and obliteration of choroidal vessels; retinal arterial spasms; retrobulbar neuritis; papilledema; optic atrophy; cortical blindness; divergence palsy; papillary paralysis; bilateral abducens paralysis; accommodative palsy; mechanism of ocular pathology with this condition is not well defined, although there is evidence pointing to the level of cyclic adenosine monophosphate.

Clinical: Loss of appetite; weight loss; colic; constipation; insomnia; headache; dizziness; irritability; moderate hypertension; albuminuria; anemia; blue line edge of gum; encephalopathy; peripheral neuropathy leading to paralysis; convulsions; mania; coma.


Leber Hereditary Optic Neuropathy (Optic Atrophy Amaurosis; Pituitary Syndrome; Leber Syndrome)

General: Male preponderance; in acute phase of neuropathy there are three characteristic fundus changes: circumpapillary micro angiopathy, pseudoeudaema around the disk, and absence of staining on fluorescein angiography; possibly a toxic metabolic disorder, an abnormality of cyanide metabolism, or an effect of smoking; maternally inherited disease affecting young males presenting with unilateral or bilateral visual loss; second eye becomes involved within weeks to months later; positive association with an inherited mutation in mitochondrial deoxyribonucleic acid (DNA).

Ocular: Sudden severe loss of vision, which usually reaches its maximum after 1 or 2 months; complete blindness rare; central vision remains seriously impaired; occasional considerable visual improvement; sheathing of retinal vessels; circumpapillary telangiectatic microangiopathy; initial low-grade optic neuritis, then bilateral optic atrophy (partial or complete); possible swelling of the disk with hemorrhages and exudates, but usually transitory; nystagmus; macular colobomas; optic disk edema; cataracts; keratoconus; hyperemia of the disk; swelling of peripapillary nerve fiber layer.

Clinical: Headaches and vertigo; Uhthoff sign.


Leber Tapetoretinal Dystrophy Syndrome  (Amaurosis Congenita; Retinal Aplasia; Retinal Abiotrophy; Pigmentary Retinitis with Congenital Amaurosis; Dysgenesis Neuroepithelialis Retinae; Alstrom-Olsen Syndrome)

**General:** Autosomal recessive inheritance; consanguinity; occurs from teens to 30 years of age.

**Ocular:** Nystagmus; keratoconus; narrow retinal arteries; yellowish-brown or gray macular lesions; grayish atrophic retinal lesions; salt-and-pepper-like retinal pigmentation or typical “bone corpuscle” pigmented changes; keratoglobus.

**Clinical:** Mental retardation; microcephaly; mongoloid appearance; oculodigital sign; association with Down syndrome has been reported; hypoplasia of the cerebellar vermis; mild-to-moderate ventriculomegaly.


Lecithin-Cholesterol Acytransferase (LCAT) Deficiency

**General:** LCAT enzyme involved in metabolism of cholesterol deficiency; autosomal recessive; rare.

**Ocular:** Cloudy cornea; diplopia; photophobia; corneal opacities.

**Clinical:** Autoimmune hyperlipoproteinemic anemia; renal failure; hypertension.


Legg-Perthes Disease (Legg-Calve-Perthes Disease; Legg-Calve-Waldenstrom Syndrome; Coxa Plana; Legg Disease; Legg-Callve Disease)

**General:** Etiology not established; occurs in children between ages 3 and 12 years; more common in boys; unilateral involvement more common than bilateral; retardation of bone age; occasionally familial; rare in blacks; possible role for protein C and S deficiency, thrombophilia, venous hypertension, and hypofibrinolysis in the pathophysiologic mechanism of this disease.

**Ocular:** Iris processes (pectinate ligaments in the anterior chamber angle).

**Clinical:** Osteochondrosis of the capital epiphysis of the femur; limpness associated with muscular spasm.


Leigh Syndrome (Subacute Necrotizing Encephalomyelopathy; Infantile Subacute Necrotizing Encephalomyelopathy; Hyperpyruvicemia with Hyper-Alpha-Alaninemia; Gangliosidosis GM2 Type 3)

**General:** Autosomal recessive; metabolic disease occurring in infancy and childhood with increased levels of serum lactate, serum pyruvates, blood α-ketoglutarate, and aminoaciduria; course is remittent with early neuro-ophthalmologic manifestations and psychomotor retardation; the later the onset of clinical manifestations, the longer the survival time; acute form in young infants, subacute form in older infants, and chronic course in juveniles; Mutation at nt8993 of mitochondrial DNA has been reported as a common cause of Leigh syndrome; biochemical analysis revealed cytochrome c oxidase deficiency with this condition.
Ocular: Nystagmoid movements or nystagmus; disconjugate ocular movements due to tegmental involvement of brainstem; degrees of visual impairment, depending on pathologic changes involving optic nerves and tracts; optic nerve atrophy; oculomotor palsy; supranuclear gaze palsy; blindness.

Clinical: Spasticity of extremities; ataxia; muscular weakness; hemiparesis; progressive mental deterioration; hearing defects; dysphagia; dyspnea; mild hypotonia; slow development; intermittent abnormal respiratory rhythm; cranial nerve palsies.


Leiomyoma

General: Rare, benign tumor that arises from smooth muscle; usually well encapsulated.

Ocular: Pigmented tumor of ciliary body; proptosis; distorted pupil; ectropion; iris tumor; glaucoma; cataract; preferential location: ciliary body, peripheral choroid, supraciliary or suprachoroidal space; has a predilection for younger patients and females.

Clinical: Metastases have not been described.


Leishmaniasis

General: Caused by protozoa Leishmania.

Ocular: Conjunctivitis; ulcerative keratitis; ulcerating granulomatous lid lesions; lid edema; interstitial keratitis; subacute focal retinitis; retinal hemorrhage; iridocyclitis; unilateral chronic granulomatous blepharitis.

Clinical: Lesions in spleen, liver, and large intestine; fever; leukopenia; cutaneous lesions on the face.


Lenoble-Aubineau Syndrome (Nystagmus-Myoclonia Syndrome)

General: Familial; pathogenesis not known; prevalent in males; manifest during first years of life; X-linked dominant inheritance has been reported in one family.

Ocular: Congenital nystagmus associated with fasciculations of muscles spontaneously elicited by mechanical stimulation or cold.

Clinical: Tremors of head and limbs; myoclonic movements of extremities and trunk; hypospadias; abnormalities of teeth; facial asymmetry; localized edema.

### Lens-Iris Diaphragm Retropulsion Syndrome

**General:** Associated with small incision phacoemulsification

**Ocular:** Infusion of fluid into the anterior chamber; posterior displacement of the lens-iris diaphragm; posterior iris bowing; pupil dilatation; ocular discomfort

**Clinical:** Deep anterior chamber with small incision phacoemulsification


### Lenz Microphthalmia Syndrome

**General:** X-linked recessive; female carriers.

**Ocular:** Microphthalmia; microcornea; ocular coloboma; colobomatous microphthalmia.

**Clinical:** Skeletal abnormalities of vertebral column, clavicles, and limbs; severe renal dysgenesis and hydroureter; dental anomalies; hypoplasias and bilateral cryptorchidism; severe speech impairment; shortness of stature; long, cylindrical, and thin thorax; flat feet.


### Leptomeningeal Adhesive Thickening (Chronic Adhesive Arachnoiditis)

**General:** Follows a chronic leptomeningeal infection, trauma, or subarachnoid hemorrhage; insidious onset.

**Ocular:** Diplopia.

**Clinical:** Headache; nausea; vomiting; vertigo; epileptic seizures.


### Leri Syndrome (pleonosteosis Syndrome; Carpal Tunnel Syndrome)

**General:** Autosomal dominant type of congenital osseous dystrophy; early epiphyseal bone formation of extremities; Morton metatarsalgia syndrome may result; onset in early infancy.

**Ocular:** Microphthalmia; anophthalmia; oculomotor paralysis; corneal clouding; cataract.

**Clinical:** Dwarfism (disproportionate); articular deformities; cutaneous deformities; carpal tunnel syndrome (median nerve compression); deformities of thumbs and great toes; laryngeal stenosis.


**Lemoyez Syndrome**

**General:** Form of Ménière disease; however, hearing acuity improves during the climax of the vestibular attacks; onset in third or fourth decade of life.

**Ocular:** Nystagmus (spontaneous) directed toward opposite side of involved vestibular system or to the side of the increased tonic state.

**Clinical:** Dizziness; vertigo; tinnitus; improvement of hearing during vestibular attacks; sweating; nausea; tremor; low tone hearing loss.


**Leroy Syndrome**

**General:** Possible mild increase in mucopolysaccharide excretion.

**Ocular:** Nasal epicanthic folds; corneal opacities.

**Clinical:** High, narrow forehead; narrow nasal bridge.


**Leukemia**

**General:** Acute or chronic blood disorder.

**Ocular:** Engorgement of conjunctival vessels; papillary hypertrophy; aggregations of tumor cells in conjunctiva, choroid, and orbit; secondary glaucoma; retinal venous engorgement and tortuosity with pronounced constrictions; retinal hemorrhages; retinal detachment; cotton-wool spots; macular edema; papilledema; optic atrophy; optic neuritis; paralysis of extraocular muscles; hypopyon; vitreous opacities; retinal sea fans; perilimbal subconjunctival infiltrates; corneal leukemic infiltration (rare); shallow serous retinal detachments; hyphema; iris neovascularization; central retinal vein occlusion; vitreous infiltrates.

**Clinical:** Frequent involvement of central nervous system; intracranial hemorrhage; thrombocytopenia; rising white cell count.


**Lewis Syndrome (Tuberoserpigineous Syphilid of Lewis)**

**General:** Lesions more common on nose and ears but may involve eyelids primarily; clinical manifestations are similar to lupus vulgaris.

**Ocular:** Lesions most frequently involve the lower eyelids; tear ducts may become involved, with lesions of mouth and nose as a direct extension or via the lymphatic route; granulomatous conjunctival lesions, usually an extension of involvement of buccal and nasal mucosa; iridocyclitis may occur; corneal ulcers as seen in tuberculous granulomatosis.

**Clinical:** Skin lesions similar to those of the face may be seen on other parts of extremities or trunk.

Lhermitte-Levy Syndrome (Hallucinosis-Red Nucleus; Lhermitte-Delthil-Gamier Syndrome)  

**General:** Lesion of unknown nature in the upper portion of peduncle and subthalamic region; occurs in the elderly.

**Ocular:** Visual hallucinations.

**Clinical:** Auditory hallucinations; paralysis; rhythmic trembling; stroke.


Lichen Planus  

**General:** Conjunctival disorder associated with dermatologic disorder; disappears spontaneously.

**Ocular:** Conjunctivitis; cicatrising conjunctivitis; keratin plaque on bulbar conjunctiva.

**Clinical:** Grayish-white papules; oral lesions may precede skin lesions.


Lignac-Fanconi Syndrome (Fanconi-Lignac Syndrome; Cystinosis Syndrome; Cystine Storage-Aminoaciduria-Dwarfism Syndrome; Renal Rickets; Nephropathic Cystinosis)  

**General:** Autosomal recessively inherited storage disorder in which nonprotein cystine accumulates within cellular lysosomes; occurs primarily in children; prognosis in children with renal tubular insufficiency and dwarfism poor, with survival past age 10 years rare without renal transplant.

**Ocular:** Cystine crystals located in conjunctiva, cornea, sclera, iris, ciliary body, lens, and perhaps choroid; general clouding of cornea caused by dense deposition of cystine crystals; pupillary block glaucoma; photophobia; band keratopathy; posterior synecchia with thickened stroma of iris; decreased visual function; patchy retinopathy; visual field constriction.

**Clinical:** Fanconi syndrome with rickets; dwarfism; glomerular dystrophy; renal failure; oral motor dysfunction.


Linear IgA Disease  

**General:** Bullous dermatosis with pruritic urticarial lesions with overlying vesicles or bullae; skin lesions heal without scarring; homogeneous deposition of immunoglobulin A (IgA) at the dermal-epidermal junction and, rarely, deposition of other immunoglobulin present; heterogeneous disease with regard to its clinical features, target antigens, and immunogenetics; association with HLA-B8, DR3, CW7, and the linked rare tumor necrosis factor-α allele; may be induced by amiodarone.

**Ocular:** Chronic conjunctivitis; subconjunctival fibrosis; symblepharon; chronic progressive conjunctival cicatrization.

**Clinical:** Recurrent blistering skin disorder consisting of urticarial macules and plaques with vesicular eruptions on trunk and extremities; subepidermal vesiculation.

Linear Nevus Sebaceous of Jadassohn (Nevus Sebaceous of Jadassohn; Jadassohn-Type Anetoderma; Organoid Nevus syndrome; Sebaceous nevus syndrome)

**General:** Skin nevus caused by failure of separation of skin appendages from adjacent epithelium during the third month of gestation.

**Ocular:** Proptosis; epibulbar lipodermoids; colobomata of eyelids, iris, and choroid; antimongoloid fissures; ocular motor palsies; nystagmus; teratomas of orbit and aberrant lacrimal glands; corneal vascularization; vision defects; conjunctival dermolipomas; choristomas of conjunctiva, sclera; corneal vascularization/opacification; colobomas of uvea, retina, optic disk, and lids; optic nerve hypoplasia; microphthalmia; anophthalmia; hemangioma of the sclera/conjunctiva.

**Clinical:** Circumscribed lesions of the face and scalp with excessively large sebaceous glands; papillomatous epidermal hyperplasia; seizures; skeletal abnormalities, particularly in skull; failure to thrive; convulsion; mental retardation.


Lipodystrophy (Kobberling-Dunnigan Syndrome)

**General:** Disturbance of the fat metabolism; autosomal dominant; affects females predominantly; occurs at puberty.

**Ocular:** Enophthalmos; lack of lid apposition; choroidal atrophy; optic disk pallor; corneal opacity.

**Clinical:** Progressive symmetrical loss of subcutaneous fat in upper part of body, including face and orbits; fat accumulation of neck, shoulders, buffalo hump, and genitalia; hyperthyroidism; lipoatrophic diabetes; hepatosplenomegaly; acanthosis nigricans; hyperlipemia; lean muscular limbs; phlebectasia; insulin resistance; hyperglycemia; type IV lipoproteinemia.


Liposarcoma

**General:** Aggressive malignant neoplasms of lipogenic cells; occurs at any age, but rarely before age 30 years and most commonly in the fifth decade; occurs almost exclusively in adults and is found most often in the thigh or retroperitoneum.

**Ocular:** Paresis of extraocular muscle; proptosis; orbital liposarcoma; eyelid edema.

**Clinical:** Neoplasms of deeper soft tissues; metastasis to lungs, liver, lymph nodes, and periosteum.


Lissencephaly Syndrome (Miller-Dieker Syndrome)

**General:** Autosomal recessive; consanguinity; association with deletion of the LIS1 gene located at chromosome 17p13.

**Ocular:** Hypertelorism.
Clinical: Microcephaly; small mandible; bizarre facies; failure to thrive; retarded motor development; mental retardation; dysphagia; decorticated and decerebrate postures; polydactyly; malformations of brain, heart, kidneys, and other organs; spastic paraplegia; agyri-apachygyria; inverted gray-to-white matter ratio; absence of white-gray interdigitations; hypoplastic brainstem; characteristic facial dysmorphism.


Listerellosis (Listeriosis)  736

General: Caused by Gram-positive bacillus Listeria monocytogenes. High mortality among pregnant women, their fetuses, and immunocompromised persons with symptoms of abortion, neonatal death, septicemia, meningitis, brain abscesses, endocarditis.

Ocular: Conjunctivitis; keratitis; corneal abscess and ulcer; blepharitis; uveitis; endophthalmitis; cataract; secondary glaucoma.

Clinical: Vomiting; cardiorespiratory distress; diarrhea; hepatosplenomegaly; maculopapular skin lesions.


Little Syndrome (Nail-Patella Syndrome; Hereditary Osteo-Onycho-Dysplasia; HOOD Syndrome)  737

General: Inherited as autosomal dominant; affects males and females equally.

Ocular: Hypertelorism; ptosis; epicanthus; microcornea; keratoconus; sclerocornea; cataract; microphakia; light pigmentation of iris root with dark pigmented "clover-leaf" spots, referred to as the Lester line, not seen in all cases.

Clinical: Absent or hypoplastic patella; hypoplastic or dislocated head of radius; exostosis of skull bones; bilateral horns of iliac crests; longitudinal ridging of fingernails; glomerulonephritis; renal involvement; bilateral antecubital pterygia; arthrogryposis; disorder has been mapped to the long arm of chromosome 9; sensorineural hearing loss.


Locked-In Syndrome  738

General: Usually caused by extensive pontine hemorrhage; awake but paralyzed patient; unable to communicate following basilar artery occlusion; trauma.

Ocular: Ocular bobbing; bilateral paresis of horizontal gaze; spared vertical eye movements, and hearing.

Clinical: Paralysis of all four extremities and the lower cranial nerves without interference with consciousness.


### Lockjaw (Tetanus)

**General:** Acute infectious disease affecting nervous system; causative agent is *Clostridium tetani*; bacteria enters body through a puncture wound, abrasion, cut, or burn.

**Ocular:** Chemosis; keratitis; nystagmus; uveitis; corneal ulcer; cellulitis of orbit; hypopyon; panophthalmitis; pupil paralysis; pseudoptosis; blepharospasm; paralysis of third or seventh nerve; may occur following perforating ocular injuries.

**Clinical:** Severe muscle spasms; dysphagia; trismus; facial palsy; muscle stiffness; irritability.


### Loffler Syndrome (Eosinophilic Pneumonitis)

**General:** Etiology unknown, but such considerations as drug hypersensitivity, parasites, mycoses, and periarteritis nodosa have been advanced; eosinophilia up to 80%; condition self-limited and benign; may occur after using crack cocaine, after administration of medications such as minocycline, or as an idiopathic disorder.

**Ocular:** Endophthalmitis; retinal infarction with hemorrhages and exudates.

**Clinical:** Dry cough; shortness of breath; increased body temperature; weight loss; malaise; anorexia; fever; dyspnea; pleural rales; pericardial effusion; prolonged expiration; wheezing.


### Longfellow-Graether Syndrome

**General:** Rare; etiology unknown.

**Ocular:** Grossly dilated retinal veins; intermittent attacks of uniocular blindness.

**Clinical:** None.


### Lost Lens Syndrome

**General:** Occurs when the intraocular lens is completely dislocated into the vitreous cavity, caused by luxation of the implant through a zonular disinsertion or an unrecognized opening in the posterior capsule or trauma.

**Ocular:** Decreased visual acuity; retinal detachment; cystoid macular edema.


### Louis-Bar Syndrome (Cephalo-Oculocutaneous Telangiectasis; Ataxia-Telangiectasia Syndrome)

**General:** Autosomal recessive; thymic abnormality leading to an immunologic deficiency has been suggested as the cause; chromosomal translocations are found in 5% to 10% of peripheral T cells from most patients.

**Ocular:** Rapid blinking in upward gaze; "pseudo-ophthalmoplegia"; fixational nystagmus (see Roth-Bielschowsky Syndrome) halting intermittently, mainly on lateral and upward gaze; on head turning, eyes are involuntarily directed to opposite side with slow return to the primary position; ocular motor apraxic movement; loss of optokinetic responses; poor convergence ability;
telangiectasias of anterior segment and sclera; fine, bright, symmetrical red streaks of the temporal and nasal conjunctiva (usually first seen at age 4 to 6 years); prominent veins in canthal regions of conjunctiva.

**Clinical:** Progressive cerebellar ataxia; slow and scanning speech; mental retardation; cutaneous telangiectasis and fine spots of pigmentation; recurrent sinopulmonary infections; hypotonia; diminished growth; cutaneous telangiectasis of ears, cheeks, and antecubital space; deficiency of IgA; lymphoreticular malignancy; high cancer risk in children with progressive cerebellar ataxia most commonly lymphoma (B-cell type) or leukemias.


### Louping III Syndrome

**General:** Tick-borne disease of sheep; occurs in Britain.

**Ocular:** Difficulty in blinking; retrobulbar neuritis; transient diplopia.

**Clinical:** Cerebellar ataxia; mild encephalitis; weak facial muscles.


### Lowe Syndrome (Oculo-Cerebro-Renal Syndrome)

**General:** Essential enzyme or protein abnormality is unknown; sex-linked recessive trait (male incidence only); onset in early infancy.

**Ocular:** Nystagmus; congenital glaucoma; miotic pupils; no pupillary reaction; ectropion uveae; malformation of the anterior chamber angle and of the iris; Schlemm canal may be absent with imperfect angle cleavage; blue sclera; cloudy cornea; cataracts; megalocornea; corneal dystrophy; buphthalmos; microphthalmos; microphakia; mydriasis; strabismus; lens punctate cortical opacities.

**Clinical:** Mental, psychomotor, and growth retardation; aminoaciduria; albuminuria; glycosuria; renal tubular acidosis; rickets; osteomalacia; muscular hypotony; hyporeflexia; hyperactivity with bizarre choreoathetoid movements and screaming.


### Lubarsch-Pick Syndrome (Primary Amyloidosis; Idiopathic Amyloidosis; Amyloidosis)

**General:** Rare condition of unknown etiology; inherited as a dominant trait, with male preponderance; characterized by amyloid accumulation in muscles and in gastrointestinal and genitourinary tracts.

**Ocular:** Internal and external ophthalmoplegia; diminished lacrimation; amyloid deposits in conjunctival, episcleral, and ciliary vessels; vitreous opacities; amyloid deposits in the corneal stroma; retinal hemorrhages and perivascular exudates; paralysis of extraocular muscles; pseudopodia lentis; strabismus fixus convergens; keratoconus.

**Clinical:** Peripheral neuropathy (extremities); heart failure; defective hepatic and renal functions with hepatosplenomegaly; waxy skin lesions; muscular weakness (progressive); multiple myeloma; hoarseness; chronic gastrointestinal symptoms.


### Lyme Disease

**General:** Caused by tick bite; symptoms resolve after treatment.

**Ocular:** Keratitis may occur up to 5 years after the first episode; diplopia; photophobia; ischemic optic neuropathy; iritis; panophthalmitis; conjunctivitis; exudative retinal detachment; choroiditis; vitreitis; multiple cranial nerve palsies; association with acute, posterior, multifocal, placoid, pigment epitheliopathy; branch retinal artery occlusion.

**Clinical:** Arthritis; increased intracranial pressure; effusion of knees; swelling of wrists.


### Lymphadenosis Benigna Orbitae

**General:** Onset from age 18 to 88 years; localized inflammatory process of undetermined origin; duration usually 2 to 12 months.

**Ocular:** Exophthalmos; well-circumscribed painless swelling around the eyes; glaucoma; restricted eye movements.

**Clinical:** Malignant tumor; insect bites; lymphadenosis benigna cutis.


### Lymphangioma

**General:** Poorly circumscribed infiltrating lesions consisting of lymphatics/dysplastic blood vessels; occurs predominantly in children and young adults.

**Ocular:** Conjunctival hemorrhages; cellulitis of lid; ptosis; exophthalmos; amblyopia; astigmatism; extraocular muscle imbalance; optic disk edema; retinal striae.

**Clinical:** Benign tumors of the lymph system.


### Lymphedema

**General:** Abnormal accumulation of lymph in the extremities; occurs from multiple causes.

**Ocular:** Conjunctival chemosis; ectropion; ptosis; strabismus; hyperpigmentation of eyelids; chorioretinal dysplasia; distichiasis; eyelid lymphedema.

**Clinical:** Abnormal lymphatic drainage; painless swelling; fibrosis of skin and subcutaneous tissues; skin becomes thickened, brown, multiple papillary projections (lymphostatic verruca); microcephaly; lymphedema.


Lymphocytic Choriomeningitis (Aseptic Meningitis)  751

**General:** Virus of which natural host is house mouse; more common in late fall and winter; usually benign.

**Ocular:** Palpebral edema; optic neuritis; ocular motor palsy; pupillary and accommodative pareses; strabismus; diplopia; conjunctival injection.

**Clinical:** Fever; meningeal irritation; headaches; confusion; coma; stiffness of neck; Babinski sign; may occur early in the course of human immunodeficiency virus infection; syndrome is considered to be usually viral in origin, with enteroviruses accounting for most cases, but rare bacterial organisms, such as *Mycobacterium tuberculosis*, *Leptospira* sp, *Brucella* sp, *Borrelia burgdorferi*, and others, may cause aseptic meningitis; drug-induced aseptic meningitis also should be considered.


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Lymphogranuloma Venereum (Nicolas-Favre Disease; Tropical Bubo LGV; Lymphogranuloma Inguinale)  752

**General:** Venereally transmitted infection caused by chlamydia.

**Ocular:** Conjunctivitis; chronic lid edema; keratitis; pannus; corneal ulcer; keratoconus; episcleritis; uveitis; tortuosity of retinal vessels; retinal hemorrhages.

**Clinical:** Enlargement of inguinal lymph nodes; lymphadenitis; lymphogranuloma.


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Lymphoid Hyperplasia (Reactive Lymphoid Hyperplasia; Lymphoid Tumors; Malignant Lymphoma; Pseudolymphoma; Pseudotumor; Burkitt Lymphoma; Neoplastic Angioendotheliomatosis)  753

**General:** Occurs in tropical Africa; young children; idiopathic orbital inflammation; systemic disease is rarely associated but occasionally occurs with either vasculitis or lymphomas; etiology of Burkitt lymphoma currently includes three factors: (i) Epstein-Barr virus, (ii) malaria, and (iii) chromosomal translocations activating the *c-myc* oncogene, which induces uncontrolled B-cell proliferation.

**Ocular:** Proptosis; extraocular motility disturbances; lesions of orbit, lacrimal gland, conjunctiva, and uvea; cortical blindness; retinal artery occlusion; retinal vascular and pigment epithelial alterations; vitreitis.

**Clinical:** Maxillary tumor; Epstein-Barr virus; cranial neuropathy.


Macular Halo Syndrome

**General:** Probably variant of Niemann-Pick disease; major differences are the ocular lesion described as macular halo with a granular appearance instead of the classic cherry-red spot and the lack of major visual symptoms.

**Ocular:** Crystalloidal opacities of foveolae; granular macula; macular halo.

**Clinical:** Hepatosplenomegaly; hyperlipidemia; histiocytes of bone marrow, spleen, and liver.


Macular Dystrophy

**General:** Sex-linked; macular dystrophy of fundus.

**Ocular:** Posterior pole changes; marked decrease in visual acuity; congenital stationary night blindness; color blindness; vitreotapetoretinal dystrophy; choroidal dystrophy; senile macular choroidal degeneration; retinoschisis; prognosis for retaining functional visual acuity is good.

**Clinical:** Lipidoses; neurologic disorders.


Macular Dystrophy, Concentric Annuli (Bull's-Eye Macular Dystrophy)

**General:** Autosomal dominant; no male-to-male transmission observed.

**Ocular:** Dyschromatopsia; foveal hyperpigmentation; perifoveal hyperpigmentation; perifoveal circular pigment epithelial atrophy.

**Clinical:** None.


Macular Dystrophy, ectodermal Dysplasia and Ectrodactyly (EEM syndrome)

**General:** Associated with abetaloproteinemia

**Ocular:** Macular dystrophy; hypotroichosis; vitreous hemorrhage; persistant hyloid artery


Macular Dystrophy, Fenestrated Sheen Type

**General:** Autosomal dominant; progressive; onset in the sixth decade.

**Ocular:** Yellowish retractible sheen in sensory retina at the macula; red fenestrations present within sheen; hypopigmentation of retinal pigment epithelium.

**Clinical:** None.


**Macular Edema, Cystoid**

**General:** Autosomal dominant; edema due to leaking perimacular capillaries.

**Ocular:** Retinal capillary leakage all over posterior pole of the eye; whitish punctate deposits in vitreous; hyperopia; "beaten bronze" atrophy to macula; strabismus.

**Clinical:** Patients show cystoid macular edema at a young age with gradual progressive decrease in visual acuity starting between the first and the fourth decades of life.


**Mad Hatter Syndrome**

**General:** Chronic mercury intoxication; symptoms seldom improve regardless of treatment.

**Ocular:** Constricted visual fields; lens opacities.

**Clinical:** Anorexia; peripheral neuritis; ataxia; hearing impairment; hyperreflexia; progressive mental depression; tremor; insomnia; fatigue; irritability; lethargy; hallucinations; shyness; withdrawal; gingivitis; teeth loosening.


**Maffucci Syndrome (Multiple Enchondromatosis; Progressive Dyschondroplasia and Multiple Hemangiomas; Kast Syndrome; Osteochondromatosis)**

**General:** Rare; etiology unknown; no hereditary factor; manifest in ages I to 5 years; characterized by numerous cartilage tumors involving mainly small bones of hands and feet; malignant transformation common; no simple mendelian inheritance.

**Ocular:** Hemangiomas of lid and retina.

**Clinical:** Multiple enchondromas with secondary bony deformities (dyschondroplasia of Ollier); chondrosarcomas; multiple hemangiomas and phlebolithiasis; orthostatic hypotension (depending on extent and size of hemangiomas); frequent fractures following minimal trauma; precocious pseudopuberty.


**Majewski Syndrome**

**General:** Autosomal recessive; normal chromosomes; perinatal mortality; has been suggested to be related to Mohr syndrome.

**Ocular:** Cataract; optic disk edema; optic atrophy; hypertelorism; absent lashes and brows; persistent pupillary membrane.

**Clinical:** Short rib polydactyly; cleft lip; cleft palate; narrow thorax; short tibia; hypoplastic epiglottis; lung and visceral abnormalities.


Malaria

**General:** Caused by *Plasmodium*, which is transmitted by mosquito bite, blood transfusion, or contaminated needles and syringes.

**Ocular:** Proliferative retinitis; vascular embolism; keratitis; ocular herpes simplex; blepharitis; optic atrophy; papilledema; papillitis; optic neuritis; anisocoria; Argyll Robertson pupil; vitreal hemorrhages and opacity; cataract; myopia; strabismus; uveitis; scleral icterus; scotoma; lagophthalmos; ptosis; subconjunctival hemorrhages; paralysis of third, fourth, or sixth nerve; epibulbar hemorrhage involving the conjunctiva, episclera, tendinous insertion of the medial rectus.

**Clinical:** Fever; anemia; splenomegaly; death.


Malignant Hyperpyrexia Syndrome (Postcataract Hyperpyrexia Syndrome; Postinduction Hyperpyrexia Syndrome)

**General:** Etiology uncertain, but believed to be a secondary response to suxamethonium and halothane used with general anesthesia; high mortality rate of about 70%.

**Ocular:** Malignant hyperpyrexia following congenital cataract or strabismus surgery under general anesthesia.

**Clinical:** Rapid elevation of body temperature and vastly enhanced metabolic activity; hyperapnea; tachycardia.


Malignant Hyperthermia Syndrome

**General:** Pharmacogenetic disease with uninhibited flow of calcium ion into muscle substance; leads to combined metabolic and respiratory acidosis and liberation of heat; cellular death may result; autosomal dominant; more common in children and young adults; unusual disorder of skeletal and cardiac muscle triggered by anesthetic agents.

**Ocular:** Blepharoptosis; squint; pupils fixed and dilated.

**Clinical:** Hernias; kyphosis; clubfoot; excessive muscular bulk; muscle cramps; unstable blood pressure; rapid and deep respiration; mottled cyanosis; arrhythmias; muscle rigidity; oliguria; anuria; deep tendon reflexes absent.


Mannosidosis

**General:** Rare; deficiency of alpha-mannosidase activity

**Ocular:** Lens opacities; corneal opacities; strabismus; late onset retinal dystrophy

**Clinical:** Coarse facial features; dysostosis multiplex; hearing defects; mental retardation; hepatosplenomegaly

### Marchesani Syndrome (Weill-Marchesani Syndrome; Inverted Marfan Syndrome; Brachymorphy with Spheroophakia; Dystrophia Mesodermalis Congenita Hyperplastica) 767

**General:** Pattern of inheritance uncertain; manifest at age 9 months to 13 years.

**Ocular:** Lenticular myopia; secondary glaucoma (rare), caused by luxation of the lens; iridodonesis; ectopia lentis; spheroophakia; optic atrophy; megalocornea; corneal opacity; acute pupillary block glaucoma.

**Clinical:** Brachydactyly; reduced growth; athletic build with abundant subcutaneous tissue; short neck and large thorax; short and clumsy hands and feet; decreased joint flexibility; hearing defects; inheritable connective tissue disorder, usually inherited as an autosomal recessive.


### Marcus Gunn Syndrome (Jaw-Winking Syndrome; Congenital Trigeminooculomotor Synkinesis) 768

**General:** Familial occurrence rare, although dominant inheritance has been reported; symptoms caused by abnormal connections between external pterygoid muscle and levator palpebrae, with supranuclear or supranuclear-nuclear involvement (see Marin Amat Syndrome).

**Ocular:** Unilateral congenital ptosis in more than 90% of cases; 10% have spontaneous onset, usually in older persons; lid elevates rapidly when mouth is opened or mandible is moved to one or the other side; left eye seems to be more frequently affected than right eye; high incidences of strabismus (36%); amblyopia (34%); bilateral jaw-winking; decreased abduction.

**Clinical:** Stimulation of ipsilateral pterygoid with chewing, opening mouth, sucking, or contralateral jaw thrusts.


### Marfan Syndrome (Dolichostenomelia; Arachnodactyly; Hyperchondroplasia; Dystrophia Mesodermalis Congenita) 769

**General:** Hypoplastic form of dystrophia mesodermalis congenita; autosomal dominant; affects both sexes; has been demonstrated that an abnormality of the gene coding for the connective tissue protein fibrillin is responsible for chronic Marfan syndrome.

**Ocular:** Exotropia; nystagmus; paralysis of accommodation; myopia (axial or lenticular); iridodonesis; miosis; persistent pupillary membrane; blue sclera; spheroophakia; lens dislocation; cataract; megalocornea; retinal detachment (less frequently); pigmentary retinopathy; colobomata of macula, iris, optic nerve, and uveal tract (less frequently); keratoconus; central retinal artery occlusion; rhegmatogenous retinal detachment; syringoma.

**Clinical:** Arachnodactyly; skeletal anomalies; asymmetric thorax; dolichocephaly and high-arched palate; dissecting aneurysm; mitral valve prolapse; prominent ears; kyphoscoliosis; pectus excavatum; flat feet; hammer toes; pulmonary and kidney defects.


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**Marin Amat Syndrome (Inverted Marcus Gunn Phenomenon)**

<table>
<thead>
<tr>
<th><strong>General</strong></th>
<th>Intrafacial connection between the orbicularis oculi and external pterygoid muscles; occurs primarily after peripheral facial palsy.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Ocular</strong></td>
<td>When mouth is opened and/or mandible is moved to side opposite ptosis, closure of the eye occurs; increased tearing during mastication.</td>
</tr>
<tr>
<td><strong>Clinical</strong></td>
<td>Signs of old facial palsy usually recognizable.</td>
</tr>
</tbody>
</table>


Marin Amat. Contribucion del Estudio de la Curabilidad de las Paralisis Oculares de Origen Traumatico. *Arch Ofalmol Hispanoamo* 1918; 71.

Wartenberg R. Inverted Marcus Gunn phenomenon (so-called Marin Amat syndrome). *Arch Neural Psychiatr* 1948; 60:584.

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**Marinesco-Sjögren Syndrome (Congenital Spinocerebellar Ataxia- Congenital Cataract-Oligophrenia Syndrome)**

<table>
<thead>
<tr>
<th><strong>General</strong></th>
<th>Autosomal recessive trait; onset when child learns to walk; mitochondrial disease.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Ocular</strong></td>
<td>Cataracts; aniridia; rotary and horizontal nystagmus; nystagmus; strabismus; optic atrophy.</td>
</tr>
<tr>
<td><strong>Clinical</strong></td>
<td>Cerebellar ataxia; oligophrenia; small stature; scoliosis; genu valgum; restricted extensibility of the knee; defects of fingers and toes; mental retardation; hair sparse; hypersalivation; sensorineural hearing loss.</td>
</tr>
</tbody>
</table>


Sjögren T. Hereditary congenital spinocerebellar ataxia combined with congenital cataracts and oligophrenia. *Confinia Neurol* 1950; 10[Suppl 46]:293.

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**Maroteaux-Lamy Syndrome (Systemic Mucopolysaccharidosis Type VI; MPS VI Syndrome; Mucopolysaccharidosis VI)**

<table>
<thead>
<tr>
<th><strong>General</strong></th>
<th>Onset in infancy; etiology unknown; autosomal recessive; excessive urinary excretion of chondroitin sulfate B; lysosomal storage disease; deficiency of the enzyme arylsulfatase B; multiple clinical phenotypes.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Ocular</strong></td>
<td>Corneal haziness and opacities; pupillary membrane remnants.</td>
</tr>
<tr>
<td><strong>Clinical</strong></td>
<td>Skeleton deformities; restriction of articular movements; dyspnea; heart murmur; hearing impairment.</td>
</tr>
</tbody>
</table>


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**Marquardt-Loriaux Syndrome (Wolfram Syndrome; Diabetes Insipidus-Diabetes Mellitus-Optic Atrophy-Deafness Syndrome; DIDMOAD Syndrome)**

<table>
<thead>
<tr>
<th><strong>General</strong></th>
<th>Autosomal recessive; present from childhood; age of onset varies.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Ocular</strong></td>
<td>Optic nerve atrophy; color blindness; visual field defects; anisocoria; diabetic retinopathy; nystagmus; cataract; pigmentation of retina.</td>
</tr>
</tbody>
</table>

198
Clinical: Juvenile diabetes mellitus; diabetes insipidus; neurosensory hearing loss; hypertension; cerebellar dysfunction; vertigo; atony of urinary tract; anosmia; peripheral neuropathy; mitochondrial abnormalities; moderate hearing loss.


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Marshall (D) Syndrome (Atypical Ectodermal Dysplasia)

General: Autosomal dominant; variant of ectodermal dysplasia; onset at birth.

Ocular: Myopia; congenital cataract (spontaneous absorption not uncommon); degenerative fluid vitreous; luxation of lens; cataract; shallow orbits.

Clinical: Facial malformation; saddle nose; hypohidrosis; partial deafness; flat or retracted midface.


---

Marshall (RE) Syndrome

General: Present from birth; etiology unknown; death usually from pneumonia before age 20 months.

Ocular: Exophthalmos; blue sclera; megalocornea; thick eyebrows.

Clinical: Underweight for length; long cranium; prominent forehead; hyperextension; small mandible; small upturned nose; broad middle and proximal phalanges; repeated respiratory infections; failure to thrive; mental retardation; accelerated skeletal growth.


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Marshall-Smith Syndrome

General: Rare congenital condition with advanced bone age, facial anomalies, and relative failure to thrive.

Ocular: Hypertelorism, protuberant eyes with shallow orbits.

Clinical: Feeding and respiratory difficulties; developmental delay; advanced bone age; characteristic facies.


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Martsolf Syndrome

General: Autosomal recessive; rare; cardiac abnormalities.

Ocular: Cataracts.

Clinical: Mental retardation; short stature; hypogonadism.


Masquerade Syndrome

**General:** Chronic blepharoconjunctivitis due to an underlying conjunctival carcinoma.

**Ocular:** Squamous cell and sebaceous carcinomas that mimic chalazion or other eyelid lesions; lacrimal secretory and excretory systems; lymph node involvement; hematogenous spread (rare).

**Clinical:** Orbital or regional involvement; intracranial extension and dural invasion.


Mastocytosis (Urticaria; Mast Cell Leukemia)

**General:** Increased mast cells found in tissues and organs; range from cutaneous to systemic condition.

**Ocular:** Conjunctival pigmentation; keratitis; pingueculae.

**Clinical:** Urticarial wheals; mast cells infiltrate into liver, spleen, gastrointestinal system, and bones.


Matsoukas Syndrome (Oculo-Cerebro-Articulo-Skeletal Syndrome)

**General:** Autosomal dominant; some of the features are found in Larsen syndrome, Schwartz syndrome, Hallermann-Streiff syndrome, Mietens syndrome, and Stickler syndrome; both sexes affected; onset at birth.

**Ocular:** Microphthalmia; myopia; increased pupillary distance; cataract; corneal sclerosis with vascular pericorneal net.

**Clinical:** Small stature; multiple joint dislocations; mental retardation; high palate; small mouth.


McFarland Syndrome

**General:** Autosomal recessive; duplication of chromosome 16q22 has been proposed; prominent amniotic fluid leakage.

**Ocular:** Hypertelorism; dysotopic canthi.

**Clinical:** Flat-appearing face; prominent forehead and frontal bossing; dislocation of joints (elbows, knees, hips most commonly); malformation of feet; short metacarpals; heart defects (usually ventricular septum); harelip; cleft palate; micrognathia.


McKusick-Weiblaecher Syndrome

**General:** Cataract; leg absence deformity; rare; two cases reported in Amish females whose parents share same ancestors; autosomal recessive.
Ocular: Congenital cataract; partial paralysis of oculomotor III nerve.

Clinical: Absence or deformity of leg; progressive scoliosis; partial duplication of foot; imperforate anus.


Measles (Morbilli; Rubèola) 783

General: Acute, extremely communicable disease that affects young school-aged children; caused by paramyxovirus; multiple sclerosis.

Ocular: Hypopyon; uveitis; conjunctivitis; Koplik (Hirschberg) spots of conjunctiva; keratitis; corneal ulcer; cellulitis of lid; dacryocystitis; congenital cataract; optic atrophy; optic neuritis; strabismus; pigmentary retinopathy; iris prolapse; hemianopsia; secondary glaucoma; central retinal artery occlusion; orbital cellulitis; accommodative spasm; paralysis of sixth nerve; keratoconus.

Clinical: Maculopapular rash; fever.


MEB Disease (Muscle-Eye-Brain Disease) 784

General: Autosomal recessive; possibly the same as Walker-Warburg syndrome. Ocular: Severe congenital myopia; congenital glaucoma; pallor of optic disk; retinal hypoplasia.

Clinical: Congenital muscular dystrophy; mental retardation; hydrocephalus; myoclonic jerks; high serum creatine phosphokinase.


Meckel Syndrome (Dysencephalia Splanchnocystic Syndrome; Gruber Syndrome) 785

General: Autosomal recessive; ocular manifestations are similar to those of trisomy 13-15 syndrome.

Ocular: Cryptophthalmos; clinical anophthalmos; microphthalmos; mongoloid slant of lid fissures; sclerocornea; microcornea; partial aniridia; cataract; retinal dysplasia; posterior staphyloma; optic nerve hypoplasia.

Clinical: Sloping forehead; posterior encephalocele; short neck; polydactyly and syndactyly (hands and feet); polycystic kidneys; cryptorchidism; cleft lip and palate; central nervous system abnormalities, including the Dandy-Walker malformation.


MELAS Syndrome 786

General: Changing of threonine at amino acid 109 to an alanine; A3245G mitochondrial DNA point mutation Cataracts; RPE abnormalities with age-related maculopathy
**Clinical:** Migraines; sensorineural hearing loss; grand mal seizures; stroke-like episodes; lactic acidosis; ragged-red muscle fibers


**Melkersson-Rosenthal Syndrome (Melkersson Idiopathic Fibroedema; Miescher Cheilitis Granulomatosis)**

**General:** Occurrence in childhood or youth; possible etiologies include viral infection, tuberculosis, sarcoidosis, and allergic reactions (all affecting parasympathetic cells in geniculate ganglia); facial palsy resembles Bell palsy; possible localization of this disorder to the gene at 9p11 has been reported.

**Ocular:** Lagophthalmos; lid edema; lacrimation secondary to the "crocodile tear" phenomenon from aberrant seventh nerve regeneration; exposure keratitis and corneal ulcers; corneal opacities.

**Clinical:** Chronic edema of face and lips; peripheral facial palsy (may be bilateral), which may precede edema by weeks to years; furrowed tongue; granulomatous cheilitis and glossitis; lingua plicata.


**Melnicke-Needles Syndrome (Osteodysplasty)**

**General:** Bone dysplasia; fewer than 30 cases reported; familial congenital autosomal trait; affect both sexes; onset at birth.

**Ocular:** Exophthalmos; hypertelorism; bilateral sclerocornea; mild cornea plana; strabismus.

**Clinical:** Misaligned teeth; micrognathia; multiple symmetric bone deformities; large ears; broad nose; frontal bossing; rosy cheeks; short stature; recurrent respiratory and ear infections; pneumosinus dilatans; abnormalities of the distal phalanges.


**Ménière Syndrome**

**General:** Etiology unknown; more common in males between ages 40 and 60 years.

**Ocular:** Nystagmus (rapid component toward the normal side), mainly during attacks; diplopia possible during and after attacks.

**Clinical:** Paroxysmal attacks of vertigo; tinnitus; gradually progressing deafness, although not prerequisite for diagnosis; during attacks, pallor, nausea, vomiting, and fainting; allergy; giant cell arteritis; facial paralysis.


**Meningioma**

**General:** Benign, slow-growing tumors that arise from the arachnoid matter, the middle layer of meninges that lies inside the dura mater, and outside the pia mater; more common in females; peak incidence in the seventh decade of life.
Ocular: Exposure keratopathy; paralysis of extraocular muscles; proptosis; optic nerve atrophy; papilledema; choroidal folds; hyperopia; visual field defect; afferent pupil defect; optociliary shunt veins.

Clinical: Headache; intracranial pressure; vomiting.


Meningococcemia (Neisseria Meningitides; Meningitis) 791

General: Systemic bacterial infection caused by Neisseria meningitides; can be present chronically in patients with immune deficiencies including deficient complement levels.

Ocular: Photophobia; conjunctivitis; chemosis; keratitis; uveitis; panophthalmitis; retinal endophlebitis; macular edema; papillitis; optic neuritis; paresis of sixth or seventh nerve; nystagmus; miosis; hippus; cortical blindness; papilledema (rare); conjunctival petechiae; strabismus.

Clinical: Meningitis; fever; malaise; joint pain; splenic enlargement.


Menkes (2) Syndrome (Kinky Hair Syndrome) 792

General: Etiology unknown; sex-linked recessive neurodegenerative disorder; focal cerebral and cerebellar degenerative changes involving the white and gray matter; affects only males; onset in early infancy.

Ocular: Decreasing visual function with progression of the disease.

Clinical: Spasticity; refractory motor seizures; retarded growth; dementia; abnormal pigmentation of hair with kinky, wiry texture; lack of facial expression; thick and dry skin; transient jaundice.


Mercury Poisoning (Minamata Syndrome) 793

General: Both sexes affected; onset several weeks or months after ingestion of fish from contaminated water or animals fed with contaminated grain; symptoms may be mild to severe.

Ocular: Constriction of visual fields; blindness.

Clinical: Paresthesia of mouth, tongue, and extremity; hearing loss; asthenia; fatigue; inability to concentrate; dysarthria; tremors; persistent vegetative state; peripheral neuropathy; cerebella ataxia; gait disturbance; sensory impairment; anosmia; loss of taste; bladder disturbance; mental deterioration.

Meretoja Syndrome (Finnish Type, FAP IV)  794

**General:** Lattice corneal dystrophy type II with familial amyloid polyneuropathy type IV; also called primary hereditary systemic amyloidosis.

**Ocular:** Lattice corneal dystrophy; cranial nerve palsies.

**Clinical:** Multiple neurologic symptoms such as severe itching, various nerve palsies, and diminished vibratory sensation; patients are said to develop a so-called bloodhound-like appearance due to skin and facial nerve degeneration.


MERRF Syndrome  795

**General:** Associated with mitochondrial tRNA [Leu(UUR)] A3243G mutation.

**Ocular:** Optic neuropathy; pigmentary retinopathy, ophthalmoparesis, and ptosis.

**Clinical:** Mitochondrial encephalomyopathy; lactic acidosis; stroke-like episodes.


Mesher's Macronecrotic Dystrophy of Retinal Pigment Epithelium (Butterfly-Shaped Dystrophy of Retinal Pigment Epithelium)  796

**General:** Autosomal recessive; autosomal dominant inheritance has been reported.

**Ocular:** Butterfly-shaped dystrophies of retinal pigment epithelium; macular degeneration associated with fundus flavimaculatus; drusen of Bruch membrane; choroidal folds; bull's-eye degeneration of macula; detachment of pigment epithelium.

**Clinical:** None.


Mesodermal Dysgenesis (Anterior Chamber Dysgenesis; Dysembryogenesis; Anterior Segment Ocular Dysgenesis Syndrome)  797

**General:** Mesodermal abnormalities, including oculocutaneous albinism; autosomal dominant.

**Ocular:** Capsular cataracts; external ophthalmoplegia; anterior chamber cleavage syndrome; atrophy of iris; ectropion; flat cornea; coloboma of iris and choroid; posterior embryotoxon; Axenfeld anomaly; Rieger anomaly; Peters anomaly; keratoconus; microphthalmos.

**Clinical:** None.


Metaphyseal Chondrodysplasia with Retinitis Pigmentosa  798

**General:** Autosomal recessive.

**Ocular:** Retinitis pigmentosa.
Clinical: Defective cartilage and growth of long bones, particularly the metacarpals and phalanges.


Metastatic Bacterial Endophthalmitis

General: Causative agent usually of low pathogenicity (e.g., Staphylococcus albus, Staphylococcus epidermidis); occasionally organisms of greater pathogenicity (e.g., Pseudomonas aeruginosa, Diplococcus pneumoniae); bilateral 45%; organisms originate in body or are introduced by drug addicts using nonsterile needles.

Ocular: Conjunctival hemorrhages; conjunctivitis; Roth spots; retinal arterial occlusion; uveitis; hypopyon; chorioretinitis; endophthalmitis; retinal hemorrhages.

Clinical: Manifestations are nonspecific.


Metastatic Fungal Endophthalmitis

General: Usually occurs in immunosuppressed or immunocompromised patients; usually asymmetrical; Candida albicans frequent etiologic agent.

Ocular: Anterior uveitis; vitreitis; focal retinitis; Roth spots; chorioretinitis; Fusarium solani also has been isolated from immunocompromised patients with endogenous endophthalmitis.

Clinical: May be evidence of other monocular foci of metastatic fungal disease.


Methemoglobinemia

General: Deficiency of enzyme; inherited or acquired, with acquired most common; caused by contact with drugs and chemicals; disorder disappears when offending chemical is eliminated.

Ocular: Pigmentation of conjunctiva and retina.

Clinical: Cyanosis; mental retardation; central nervous system involvement.


Meyer-Schwickerath-Weyers Syndrome (Microphthalmos Syndrome; Oculodentodigital Dysplasia)

General: Etiology unknown; two types recognized: (I) dysplasia oculodentodigitalis and (II) dyscraniopygophalangie; type I is characterized by microphthalmia with possible iris pathology and glaucoma, oligodontia and brown pigmentation of teeth, camptodactyly, and possible absence of middle phalanx of second to fifth toes; type II consists of severe microphthalmos to anophthalmos, polydactyly, and developmental anomalies of nose and oral cavity; both sexes affected; present from birth; abnormal cerebral white matter.

Ocular: Microphthalmos; hypotrichosis; glaucoma; iris anomalies (eccentric pupil; changes in normal iris texture; remnants of pupillary membrane along iris margins); microcornea; hypertelorism; myopia; hyperopia; keratoconus.
**Clinical:** Thin, small nose with anteverted nostrils and hypoplastic alae; syndactyly; camptodactyly (fourth and fifth fingers); anomalies of middle phalanx of fifth finger and toe; hypoplastic teeth; wide mandible; alveolar ridge; sparse hair growth; visceral malformations.


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<thead>
<tr>
<th>Micro Syndrome</th>
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<tr>
<td><strong>General:</strong> Autosomal recessive microcephaly and microcornea; Muslim Pakistani inheritance; present at birth; consanguinity; autosomal recessive.</td>
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<tr>
<td><strong>Ocular:</strong> Microcornea; congenital cataract; retinal dystrophy; optic nerve atrophy; ptosis; microphakia; microphthalmos; nuclear cataract; atonic pupils.</td>
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<tr>
<td><strong>Clinical:</strong> Severe mental retardation; hypothalamic hypogenitalism; hypoplasia of the corpus callosum; short stature; cortical visual impairment; microcephaly; developmental delay.</td>
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**Microcephaly with Chorioretinopathy**

| **General:** Autosomal dominant; congenital infection; exposure to irradiation, chemical agents, mother’s infection, or injury. |
| **Ocular:** Chorioretinopathy usually inactive. |
| **Clinical:** Microcephaly; slow growth of brain; mild mental retardation. |


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**Microcephaly, Microphthalmia, Cataracts, and Joint Contractures**

| **General:** Autosomal dominant; ocular features like Hagberg-Santavuori syndrome. |
| **Ocular:** Microphthalmia; cataracts; hypopigmented retinal degeneration. |
| **Clinical:** Microcephaly; shortening or wasting of muscle fibers, causing excess scar tissue over joints. |


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**Microphthalmia and Mental Deficiency**

| **General:** Autosomal recessive. |
| **Ocular:** Microphthalmia; corneal opacities. |
**Clinical:** Severe mental retardation; spastic cerebral palsy; glycinuria; abnormally small head.


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**Microphthalmos, Myopia, and Corectopia**

**General:** Autosomal dominant; characterized by microphthalmos, myopia, and corectopia.

**Ocular:** Microphthalmos; myopia; corectopia; glaucoma; cataract; hypoplastic macula; spherophakia; microphakia.

**Clinical:** None.


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**Microphthalmos, Pigmentary Retinopathy, Glaucoma**

**General:** Autosomal dominant; three disorders combined.

**Ocular:** Microphthalmos; pigmentary retinopathy; glaucoma.

**Clinical:** None.


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**Micropsia Syndrome (Lilliputian Syndrome)**

**General:** Psychosensory illusion produced by various mental derangements such as acute infections, alcoholism, toxic delirium, dementia, or trauma.

**Ocular:** Illusions, with misjudging of distance, position, and size of known objects (regarded as a psychovisual phenomenon).

**Clinical:** Fixed hallucinations or dreams are expressions of illusions and are misinterpreted by the patient.

Bender MB, Savitzky N. Micropsia and teleopsia limited to the temporal fields of vision. *Arch Ophthalmol* 1943; 29:904.


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**Microspherophakia with Hernia**

**General:** Autosomal dominant.

**Ocular:** Microspherophakia; glaucoma.

**Clinical:** Inguinal hernia.


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**MIDAS Syndrome (Microphthalmia, Dermal Aplasia and Sclerocornea)**

**General:** X-linked phenotype; male-lethal trait

**Ocular:** Bilateral microphthalmia; sclerocornea; blepharophimosis
Clinical: dermal aplasia; microcephaly; cardiomyopathy; ventricular fibrillation; congenital heart defect


Mietens Syndrome (Mietens-Weber Syndrome)

General: Etiology unknown; unclassifiable familial condition.

Ocular: Bilateral corneal opacities; horizontal and rotational nystagmus; strabismus; bushy eyebrows; ptosis.

Clinical: Growth failure; flexion contracture of the elbows; dislocation of the head of the radii; mental retardation; small pointed nose with a depressed root; low hairline; external ear defects; digital defects; hypertrichosis.


Migraine (Vascular Headache)

General: Recurrent attacks of pain in the head; usually unilateral; often familial.

Ocular: Abnormal visual sensations; scotoma generally restricted to one half of the visual field; complete blindness; unilateral transient visual loss; photopsia; branch retinal artery occlusions; anisocoria.

Clinical: Nausea; vomiting; anorexia; sensory, motor, and mood disturbances; fluid imbalance; headache.


Mikulicz-Radecki Syndrome (Mikulicz Syndrome; Dacryosialoadenopathy; Mikulicz-Sjögren Syndrome)

General: Not an individual disease but a manifestation of tuberculosis, syphilis, leukemia, lymphosarcoma, sarcoidosis, Hodgkin disease, mumps, Waldenström macroglobulinemia, or lymphoma; exhibits a chronic course with frequent recurrences; milder form of Sjögren syndrome (see Schaumann Syndrome).

Ocular: Bilateral painless enlargement of lacrimal glands with bulging of upper lid; decreased or absent lacrimation; conjunctivitis, uveitis; optic atrophy; optic neuritis; phlyctenules; keratoconjunctivitis; dacryoadenitis; retinal candlewax spots; periphlebitis.

Clinical: Symmetrical, perhaps marked, enlargement of salivary glands; dryness of mouth and pharynx; hoarseness; neurologic complications.


Millard-Guber Syndrome (Abducens-Facial Hemiplegia Alternans)

General: Vascular, infectious, or tumorous lesion at the base of the pons affecting the nuclei of the sixth and seventh nerves and fibers of the pyramidal tract; demyelinating disease.

Ocular: Diplopia; esotropia; paralysis external rectus muscle (often bilateral); in unilateral cases, there is deviation of eyes to side opposite lesion and inability to move them toward side of lesion; abduction of eye prevented by destruction of sixth nerve nucleus; opposite eye cannot be voluntarily adducted but can converge and move in this position by rotatory and caloric stimulation.
Clinical: Ipsilateral facial paralysis; contralateral hemiplegia of arm and leg from involvement of pyramidal tract.


Miller Syndrome (Postaxial Acrofacial Dysostosis; Genee-Wiedemann Syndrome) 816

**General:** Cause unknown; sporadic and familial cases known as Genee-Wiedemann Syndrome.

**Ocular:** Ectropion.

**Clinical:** Malar hypoplasia; cleft palate and lip; postaxial limb deficiency; cup-shaped ears.


Miller Syndrome (Wilms Aniridia Syndrome; WAGR Syndrome; Wilms Tumor-Aniridia-Genitourinary Abnormalities-Mental Retardation Syndrome) 817

**General:** Etiology unknown; manifests an association of aniridia, which is inherited as a dominant autosomal trait, and Wilms tumor; this is one of the best studied continuous gene syndromes as defined by Schmickel.

**Ocular:** Glaucoma; bilateral aniridia (aniridia often not complete, with remnants of iris root present as rudimentary forms); cataract.

**Clinical:** Wilms tumor; mental retardation with microcephaly; genital malformations with cryptorchidism and hypospadias; hemihypertrophy; kidney anomalies (horseshoe kidney).


Mirror Image Syndrome (Autoscopic Syndrome; Lukianowicz Phenomenon) 818

**General:** Patient's delusion that he or she is seeing a double of himself or herself; seen in patients with schizophrenia, epilepsy, migraine, and even depression; the "double" usually appears suddenly and is of white or gray hue (see Capgras Syndrome, of which this syndrome is a variant).

**Ocular:** Hallucination in the form of seeing a double of self.

**Clinical:** Migraine; schizophrenia; epilepsy; depression.


Misdirected Third Nerve Syndrome

**General:** May occur with a variety of inflammatory infections and parainfections, vascular lesions, tumors, and degenerative and demyelinating diseases that may involve the nerve anywhere; may occur as primary aberrant regeneration without prior history of acute oculomotor nerve palsy.

**Ocular:** Bizarre eyelid movements that may accompany various eye movements; lid may rise as the medial rectus, the inferior rectus, or the superior rectus muscle contracts; iridoplegia; ptosis.

**Clinical:** None.


ML I (Mucolipidosis I; Lipomucopolysaccharidosis; Dysmorphic Sialidosis; Spranger Syndrome)

**General:** Rare storage disease; autosomal recessive; increased sialic acid and deficiency of the enzyme alpha-N-acetyllneuraminidase in cultured mucolipidosis I fibroblasts.

**Ocular:** Variable corneal clouding; macular cherry-red spot; optic atrophy; lens opacity; pupillary reflexes anomaly; grayish area around cherry-red spot.

**Clinical:** Moderate progressive mental retardation; skeletal changes of dysostosis multiplex; peripheral neuropathy; myoclonic jerks; tremor; cerebellar signs; gait abnormalities.


ML II (I-Cell Disease; Mucolipidosis II)

**General:** Autosomal recessive mucolipidosis is a Hurler-like disorder with some radiologic features, striking fibroblast inclusions, and no excess mucopolysacchariduria; abnormal N-acetylglucosamine phosphotransferase.

**Ocular:** Minimal corneal clouding; glaucoma; megalocornea.

**Clinical:** Congenital dislocation of the hips; thoracic deformities; hernia; hyperplastic gums; retarded psychomotor development and restricted joint mobility; dysmorphic facies; skeletal deformities; organomegaly; short stature; mental retardation.


ML III (Pseudo-Hurler Polydystrophy; Mucolipidosis III)

**General:** Autosomal recessive disorder, almost indistinguishable biochemically from mucolipidosis II; decreased levels of N-acetylgalactosamine phosphotransferase.

**Ocular:** Increased corneal thickness; wrinkled maculopathy; granular pigmented changes of fundus; papilledema; hyperopic astigmatism; corneal opacities; retinal vascular tortuosity; visual field defects.

**Clinical:** Joint stiffness; coarse facial feature; short stature; aortic valve disease; arm and hand deformities; self-mutilation of the distal phalanges; carpal tunnel syndrome.


ML IV (Mucolipidosis IV; Berman Syndrome)

**General:** Storage disease in which corneal clouding is an early sign with no evidence of systemic involvement until age 1 year; autosomal recessive; cases seen in Ashkenazi Jews; abnormal neuraminidase.

**Ocular:** Corneal clouding; corneal opacities; epithelial edema; retinal atrophy; pale optic nerve; diffuse corneal clouding present at birth or in early infancy.

**Clinical:** Progressive psychomotor retardation; skeletal dysplasia; facial anomalies.


MMMM Syndrome (Neuhauser Syndrome; Megalocornea, Macrocephaly, Mental and Motor Retardation)

**General:** Rare

**Ocular:** Megalocornea

**Clinical:** Mental retardation; hearing loss; sensorineural complications; hypoplasia; corpus callosum; macrocephaly


Möbius I Syndrome (Hemicrania, Hemiplegic; Hemiplegic-Opthalmoplegic Migraine; Hemiplegic Familial Migraine)

**General:** Etiology unknown; indirect indications of unilateral cerebral edema due to vasomotor phenomena; occurs in young adults; recovery usually follows after a few days; no clear etiology has been determined, including a vascular theory of embryopathogene-sis, a chromosome translocation, and exposure to teratogens.

**Ocular:** Extraocular palsy; permanent damage of oculomotor nerve III.

**Clinical:** Hemicrania; hemiparesis; aneurism of internal carotid; neoplasia; headache.


Möbius II Syndrome (Congenital Facial Diplegia; Congenital Paralysis of the Sixth and Seventh Nerves; Congenital Oculofacial Paralysis; Von Graefes Syndrome)

**General:** Congenital; possibly failure of development of facial nerve cells or primary defect of muscles deriving from first two brachial arches or both; recovery in a few weeks or nonprogressive permanent paralysis of face; asymmetrical; if incomplete, usually spares lower face and platysma.

**Ocular:** Ptosis; weakness of abductor muscles; normal convergence; limitation to internal rotation in lateral movements; esotropia.

**Clinical:** Facial diplegia; deafness; loss of vestibular responses; webbed fingers or toes; clubfoot.


Möhr-Claussen Syndrome (Oral-Facial-Digital Syndrome Type II; OFD Syndrome; Orofaciadicatal Syndrome II) 827

**General:** Rare; autosomal recessive; certain features similar to Papillon-Leage-Psaume, Carpenter, Laurence-Moon-Bardet-Biedl, and Ellis-Van Creveld syndromes (see Papillon-Leage-Psaume Syndrome; Laurence-Moon-Bardet-Biedl Syndrome; Ellis-Van Creveld Syndrome).

**Ocular:** Epicanthus; bridged chorioretinal colobomata.

**Clinical:** Clefts and fibroma of tongue; polydactyly; broad nasal bridge; narrow-arched palate; short humerus, femur, and tibia; irregular teeth; hypotonia; mental retardation; deafness; thin and fair hair; cerebellar atrophy.


Möller-Barlow Disease 828

**General:** Vitamin C deficiency in children.

**Ocular:** Hemorrhages around and, in rare cases, in eyes; yellow-brown discoloration from hemosiderin may remain for some time after resolution of a subconjunctival hemorrhage; hemorrhage in eyelids, conjunctiva, anterior chamber, and retina; proptosis in infantile scurvy.

**Clinical:** Tenderness of the lower extremities; ecchymoses.


Molluscum Contagiosum 829

**General:** Etiologic agent of this disease is a poxvirus that can cause proliferative skin lesions anywhere on the body; commonly found in patients who are immunosuppressed.

**Ocular:** Lesions of lid, lid margin, conjunctiva, and cornea; conjunctivitis; keratitis; corneal ulcer.

**Clinical:** Well-defined, pearly appearing papules with umbilicated centers of varying size (3 to 10 mm); eczematization of the surrounding skin.


Monbrun-Benisty Syndrome (Ocular Stump Causalgia) 830

**General:** Sympathetic irritation of resected sympathetic fiber to the eye; occurs after trauma of eye.

**Ocular:** Severe refractory pain of orbital cavity.

**Clinical:** Pain of face and the corresponding hemicranium; congestion and hyperhidrosis of region involved.

Monofixation Syndrome (Blind Spot Syndrome; Primary Monofixation)

General: No hereditary factor; uncommon.

Ocular: Deviation of eight prism diopeters or less by simultaneous prism and cover test; central scotoma; stereopsis; good fusional vergences found in patients with congenital esotropia; unilateral syphilitic optic perineuritis (rare); congenital esotropia (inherited in a multifactorial fashion).

Clinical: Syphilis (rare).


Moraxella Lacunata

General: Gram-negative rod; causes chronic angular blepharoconjunctivitis; without treatment, may persist for months or years; normally found in flora of respiratory tract; seen more frequently in alcoholics and those with poor sanitary habits; *Moraxella* organisms produce proteases, although those are not related directly to their pathogenetic mechanism.

Ocular: Catarrhal angular conjunctivitis; corneal ulcer; hypopyon, chronic blepharitis; eczema; lateral canthal skin erythema; iridocyclitis.

Clinical: Alcoholism; impaired nutrition; dermatitis.


Morgagni Syndrome (Hyperostosis Frontalis Interna Syndrome; Intracranial Exostosis; Metabolic Cranioathy)

General: Dominant inheritance; onset around age 45 years; occurs almost exclusively in females. Cataract; optic nerve injury within the optic canal by bony protrusions, with resulting blindness.

Clinical: Hyperostosis frontalis interna; obesity (mainly trunk and proximal portions of limbs); hirsutism; menstrual disorders; hypertension; arteriosclerosis; headache; hypertrichosis; no case of male-to-male transmission is known; hyperprolactinemia.


Morning Glory Syndrome (Hereditary Central Glial Anomaly of the Optic Disk)

General: No hereditary factor; rare; anomaly of optic disk with deep excavation resembling the flower for which syndrome is named.

Ocular: Strabismus; abnormality of embryologic development of anterior chamber (anterior chamber cleavage syndrome); remnants of hyaloid system; chorioretinal pigment surrounding optic disk; narrow branches of retinal arteries at edge of optic
disk; retinal exudates and detachment; subretinal hemorrhages and retinal neovascularization; enlarged pink optic disk, funnel shaped with a central white fluffy dot; nerve head surrounded by elevated annulus of chorioretinal pigment, unilateral.

**Clinical:** Midline cranial facial defects such as hypertelorism, cleft lip/palate, basal encephalocele, agenesis of corpus callosum, sphenoid encephalocele defects in the floor of the sella turcica; cranial, facial, and neurologic associations; pituitary dwarfism; association with the CHARGE syndrome.


**Morphea (Localized Scleroderma; Circumscribed Scleroderma)**

**General:** Localized chronic connective tissue disease of unknown etiology; etiology remains unknown, although there is a possible association with *Borrelia burgdorferi* infection.

**Ocular:** Circumscribed plaque like lesions of the eyelid; prevalent in females; onset usually in second to fourth decades of life; onset occasionally associated with trauma, pregnancy, or menopause.

**Clinical:** Firm skin plaques over entire body, but most frequently on trunk, lower extremities, face, and genitalia; abdominal pain; migraine; generalized joint pain; renal crisis; Raynaud phenomenon; systemic sclerosis; eosinophilia; positive antinuclear factor; increased immunoglobulin (immunoglobulin G [IgG]); seizures; skin sclerosis; alterations in tryptophan metabolism.


**Morquio Syndrome (Morquio-Brailsford Syndrome; Brailsford-Morquio Dystrophy; Familial Osseous Dystrophy; Keratosulfaturia; MPS IV; Mucopolysaccharidosis IV; Spondyloepiphyseal Dysplasia; Osteochondrodystrophy Deformans; Infantile Hereditary Chondrodysplasia; Hereditary Poly topic Enchondral Dysostosis; Hereditary Osteochondrodystrophy; Eccentro-Osteochondrodysplasia; Dysostosis Enchondralis Meta- Epiphysaria; Morquio-Ullrich Syndrome; Atypical Chondrodystrophy; Chondrodystrophy Tarda; Chondro-Osteodystrophy)**

**General:** Autosomal recessive dystrophy of cartilage and bone; slight predilection for males; apparent between ages 4 and 10 years; excess production of keratosulfate (see Hurler Syndrome; Hunter Syndrome; Sanfilippo-Good Syndrome; Scheie Syndrome; Maroteaux-Lamy Syndrome); autosomal recessive; abnormal N-acetylgalactosamine-G-sulfate sulfatase.

**Ocular:** Enophthalmos; ptosis; excessive tear secretion; ocular hypotony; miosis; occasionally hazy cornea; bushy eyebrows; optic nerve atrophy; moderate-to-late corneal clouding.

**Clinical:** Dwarfism; skeletal deformities (progressive); delayed ossification of epiphyses; decreased muscle tone; deafness; weak extremities; waddling gait; coarse broad mouth; spaced teeth; aortic regurgitation; normal intelligence.


Mort d'Amour Syndrome (Death of Love Syndrome)  837

**General:** Sudden death during sexual intercourse.

**Ocular:** Pupillary dilation.

**Clinical:** Hypertension; arrhythmia; heart ischemia; rupture of cerebral aneurysm.


Mosse Syndrome (Polycythemia-Hepatic Cirrhosis Syndrome)  838

**General:** Unknown etiology.

**Ocular:** Scleral icterus; marked retinal venous tortuosity and dilation; retinal artery occlusion (occasionally); papilledema.

**Clinical:** Thrombosis portal vein secondary to polycythemia; hepatosplenomegaly; ascites; clinical features of liver cirrhosis.


Moyamoya Disease (Multiple Progressive Intracranial Arterial Occlusion)  839

**General:** Almost exclusively seen in Japanese infants and children; cerebrovascular disorder that results in occlusion of the large vessels at the base of the brain; slight female prevalence; collateral vascular networks (secondary to bilateral carotid occlusive disease) resembling puffs of smoke (Moyamoya in Japanese).

**Ocular:** Hemianopsia; nystagmus; papilledema; central retinal vein occlusion; visual field defects; amaurosis fugax; diplopia; optic pallor; ischemic chiasmal syndrome; bilateral renal artery stenosis.

**Clinical:** Loss of consciousness; seizures; hemiplegia; hemiparesis; intracranial hemorrhage; mental retardation; speech disturbances; unsteady gait; headache; psychiatric manifestations; focal epileptic attacks; chronic cerebrovascular disorder; intracerebral hemorrhage; pituitary adenoma (association).


Moynahan Syndrome (XTE; Xeroderma, Talipes, and Enamel Defect)  840

**General:** Autosomal dominant inheritance; xeroderma, talipes, and enamel defect.

**Ocular:** Absence of eyelashes of lower lid.

**Clinical:** Cleft palate; hypohidrosis; defective enamel; nail anomalies; coarse and dry hair; short-lasting skin bullae.


Mucocele (Pyocele)  841

**General:** Accumulation and retention of mucoid material within the sinus as a result of continuous or periodic obstruction of the sinus ostium.

**Ocular:** Paralysis of extraocular muscles; exophthalmos; lacrimation; erosion of bony walls of orbit; decreased visual acuity; diplopia; elevation of lower lid; ptosis; compression optic neuropathy; globe distortion; enophthalmos; epiphora; scleral indentation; choroidal folds; discharging lesion of the upper lid; pseudotelecanthus; spontaneous nontraumatic enophthalmos; local anestheisa.
Clinical: Headaches; epidural abscess; subdural empyema; meningitis; brain abscess; occlusion of nasal passage; loosening of teeth.


**Mucocutaneous Lymph Node Syndrome (MLN Syndrome; Kawasaki Disease)**

**General:** Multisystem syndrome with worldwide distribution; occurs from age 2 months to 9 years; increased incidence in summer; etiology unknown, but allergic reactions to chemicals or abnormal reactions to numerous infections have been suggested.

**Ocular:** Severe conjunctival congestion and hyperemia (88%); uveitis; dacryocystitis; anterior uveitis (commonly bilateral); punctate keratitis.

**Clinical:** Fever (1 to 2 weeks; does not respond to antibiotics) "strawberry tongue"; red palms and soles; indurative edema; membranous desquamation from fingertips; polymorphous exanthema; arthritis; myocarditis; enlarged cervical lymph nodes.


**Mucormycosis (Phycomycosis)**

**General:** Acute, often fatal infection caused by saprophytic fungi; associated with diabetes mellitus and ketoacidosis.

**Ocular:** Corneal ulcer; striate keratopathy; ptosis; panophthalmitis; proptosis; cellulitis of orbit; immobile pupil; retinitis; optic neuritis; paralysis of extraocular muscles; central retinal artery thrombosis.

**Clinical:** Epistaxis; nasal discharge; facial pain; facial palsies; anhidrosis; cranial nerve or peripheral motor and sensory nerve deficits may occur.


**Mucous Membrane Pemphigoid**

**General:** Immune mediated disease characterized by autoantibodies to the basement membrane zone at the sub-epithelial junction of mucous membranes

**Ocular:** Progressive cicatrizing conjunctivitis; symblepharon; corneal clouding

**Clinical:** Nasal and oral mucosa cicatrization; trachea and esophagus cictrization


**Mucus Fishing Syndrome**

**General:** Excessive mucus production created by mechanical damage of conjunctival epithelium; epithelial injury created by mechanical removal of excess mucus, which leads to further increased mucus production, causing a continuous cycle; initial mucus pro-duction may be associated with an underlying ocular disease, such as pterygium, keratoconus, blepharitis, or keratoconjunctivitis.
Ocular: Excessive mucus production; blepharitis; pterygium; keratoconjunctivitis; keratoconus; corneal foreign body; floppy lid syndrome; map-dot fingerprint dystrophy; squamous cell cancer of conjunctiva.

Clinical: None.

### Muir-Torre Syndrome

**General:** Rare; autosomal dominant; characterized by sebaceous tumors and internal malignancies

**Ocular:** Sebaceous tumor of the eyelids

**Clinical:** Gastrointestinal, breast and genitourinary malignancies; sebaceous neoplasms


### Mulibrey Nanism Syndrome (Perheentupa Syndrome)

**General:** Autosomal recessive inheritance; progressive growth failure; myocardial fibrosis.

**Ocular:** Alternating esotropia and exotropia; yellowish retinal dots and scattered pigment dispersion in clusters (especially in the midperiphery); drusen of Bruch membrane; hypoplasia of choriocapillaries (diagnostic sign).

**Clinical:** Triangular face with bulging forehead and low, broad nasal bridge; hypotonia; hepatomegaly; ascites; pulmonary congestion; cardiac enlargement with possible heart failure; pericardial constriction; dwarfism; immunoglobulin deficiency; isolated growth hormone deficiency.

### Multiple Endocrine Neoplasia 2B or 3 (MEN 2B or 3)

**General:** Autosomal dominant inheritance; multiple endocrine neoplasia type 3 (MEN3) has been separated from MEN2 because of low incidence of associated parathyroid disease in these cases; poor prognosis; several different point mutations in the RET protooncogene on chromosome 10 have been associated with the multiple endocrine neoplasia type 2 syndromes.

**Ocular:** Prominent corneal nerves in clear stroma; diffuse, nodular thickening of eyelids; nasal displacement of lacrimal puncta; rostral displacement of eyelashes; eversion of eyelids; subconjunctival neuromas; thickened conjunctival nerves; decreased tear formation; prominent eyebrows; impaired pupillary dilation; thickened iris nerves; increased intraocular pressure (rare); localized orbital neurofibromas (orbit, conjunctiva); lesions of the tongue resembling neuromas.

**Clinical:** May be present at birth or develop later; 50% show complete syndrome of multiple neuromas (lips, tongue, eyelids), bumpy lips, pheochromocytoma, and medullary carcinoma; others exhibit variable combinations of the preceding, without the pheochromocytoma; diarrhea; marfanoid habitus.


**Multiple Evanescent White-Dot Syndrome (MEWDS)**  
**General:** Unilateral disease including multiple white dots at the level of the pigment epithelium; etiology unknown; recurrent; no systemic involvement.

**Ocular:** Retinal pigment granularity in the macula; reduced visual acuity; vitreitis; loss of retinal sensitivity; white dots of pigment epithelium; uveitis; blind spot enlargement; choroidal neovascularization.

**Clinical:** None.


**Multiple Lentigines Syndrome (LEOPARD Syndrome)**  
**General:** Familial occurrence; classic features include lentigines (small focal hyperpigmentations of skin), electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonary stenosis, abnormal genitalia, retardation of growth, and deafness (LEOPARD).

**Ocular:** Hypertelorism; exophthalmos; epicanthal folds; strabismus; nystagmus; keratoconus.

**Clinical:** Lowset ears; receding chin; deafness; lentigines; pulmonary stenosis; genital abnormalities; growth retardation; skeletal malformations (bony fusion involving cervical vertebrae, ossicles, carpal and tarsal bones, scoliosis); hyposmia; heart murmur; mental retardation; hypospadias; congenital heart defect; thoracic deformities; respiratory insufficiency.


**Multiple Mucosal Neuromata with Endocrine Tumors Syndrome (Multiple Mucosal Neuromas, Pheochromocytoma, and Medullary Thyroid Carcinoma Syndrome)**  
**General:** Pheochromocytoma can be inherited and found in combination with neurofibromatosis and other brain tumors (meningioma, spongioblastoma, ependymoma, astrocytoma, cerebellar hemangioblastoma), multiple mucosal neuromas, and medullary carcinoma of the thyroid.

**Ocular:** Conjunctival neuromas; multiple, white, myelinated nerve fibers in corneal stroma arising at limbus with anastomoses in center of cornea.

**Clinical:** Neuromas of lips or anterior portion of tongue or, more rarely, of buccal, gingival, or laryngeal mucosa; hypertension; flushing; weakness; sweating; palpitations; headaches.


**Multiple Pterygium Syndrome, Lethal Type (Pterygium, Multiple, Lethal Type)**  
**General:** Autosomal recessive; nonconsanguineous parents; lethal.

**Ocular:** Hypertelorism; epicanthal folds.
Clinical: Multiple pterygia involving chin to sternum, cervical, axillary, antecubital, and popliteal areas; flexion contracture of multiple joints; small chest; hydrops; markedly flattened nasal bridge with hypoplastic nasal alae; cleft palate; micrognathia; lowset, malformed ears; short neck with a cystic hygroma on back of neck, head; pulmonary and cardiac hypoplasia; hypoplastic teeth; elongated clavicles; cryptorchidism; foot deformities; arthrogryposis; hypoplasia of the left arm, leg, pelvis, and kidney.


Multiple Sulfatase Deficiency 853

General: Autosomal recessive

Ocular: Congenital peripheral cataract; retinal degeneration; cherry red-spot of macula

Clinical: Progressive neurologic disease with death in the teenage years; ichthysis


Mulvihill-Smith Syndrome 854

General: Progeroid disorder.

Ocular: Keratoconus; conjunctivitis.

Clinical: Patients have short stature, microcephaly, unusual facies, numerous pigmented nevi, hypodontia, sensorineural hearing loss, immunodeficiency (low IgG), and a high-pitched voice.


Mumps 855

General: Viral infection.

Ocular: Conjunctivitis; keratitis; corneal ulcer; tenonitis; exophthalmos; microphthalmos; optic atrophy; optic neuritis; papillitis; scleritis; uveitis; cortical blindness; congenital punctal occlusion; paralysis of extraocular muscles; dacryoadenitis; iritis; paralysis of accommodation; internal and external ophthalmoplegias.

Clinical: Affects the parotid glands, but infection of other glandular tissue occurs, including the lacrimal gland and testicles; encephalitis; meningitis.


Muscular Dystrophy, Congenital, with Infantile Cataract and Hypogonadism 856

General: Autosomal recessive; rare; reported in isolated Norwegian village.

Ocular: Infantile cataract.

Clinical: Ovarian agenesis in females; Klinefelter syndrome in males; congenital muscular dystrophy.


Myasthenia Gravis, Neonatal or Infantile (Infantile Myasthenia Gravis; Neonatal Myasthenia Gravis) 857

**General:** Occurs in newborn of myasthenic mother; caused by compound in circulation received through placenta (see Erb-Goldflam Syndrome).

**Ocular:** Transient diplopia; ptosis of upper eyelids; internuclear ophthalmoplegia.

**Clinical:** Excessive fatigue musculature; symptoms appear and increase as day progresses; expressionless face; sagging jaw; sucking, swallowing, and respiratory difficulties.


Mycosis Fungoides Syndrome (Sézary Syndrome; Malignant Cutaneous Reticulosis Syndrome) 858

**General:** Lymphoma characterized by abnormal lymphocytes having hyperchromatic, hyperconvoluted nuclei; malignant, cutaneous T-cell lymphoma, which initially presents as a nonspecific erythematous cutaneous eruption that progresses to form plaques and tumors.

**Ocular:** Thick and swollen eyelids; ectropion; blepharitis; loss of eyelashes; keratoconjunctivitis; uveitis; retinal edema and exudates; papilledema; retinal hemorrhage; endophthalmitis; pupillary dilatation; scleritis; corneal opacity; optic disk swelling.

**Clinical:** Pruritus followed by thickening and edema of the skin; dermatitis exfoliativa; eczema; pyoderma; pigmentary changes of body and extremities (mottled appearance); hyperhidrosis.


Myelinated Optic Nerve Fibers 859

**General:** Autosomal dominant; transmission has been reported as autosomal recessive.

**Ocular:** White area adjacent to the disk caused by myelin sheath; pseudopapilledema.

**Clinical:** None.


Myopathy, Mitochondrial, with Cataract 860

**General:** Autosomal dominant.

**Ocular:** Early-onset bilateral cataracts; severe ophthalmoplegia; mitochondrial abnormalities in the inferior oblique muscle.

**Clinical:** Facial weakness; myocardial and skeletal myopathy of the mitochondrial type.


**Myopia, Infantile Severe** 861

**General:** Autosomal recessive; consanguineous parents.

**Ocular:** High myopia; optic disk cupping.

**Clinical:** None.


**Myopia, Sex-Linked** 862

**General:** Sex-linked; linkage to factor VIII gene; probable location Xq28.

**Ocular:** Myopia; hemeralopia; deuteranopia in males; hypoplasia of the optic nerve head.

**Clinical:** Short stature.


**Myopia-Ophthalmoplegia Syndrome** 863

**General:** Sex-linked; characteristics seen in males; carried by females.

**Ocular:** Ptosis; myopia; complete or partial ophthalmoplegia; abnormal pupil; progressive degeneration of retina and choroid.

**Clinical:** Patellar reflex absent; Achilles reflex absent; spina bifida; cardiac defects; absent deep tendon reflex in carriers only.


**Myotonic Dystrophy Syndrome (Myotonia Atrophica Syndrome; Dystrophia Myotonica; Curschmann-Steinert Syndrome)** 864

**General:** Rare autosomal dominant disease; onset about age 20 years; condition is worsened by administration of neostigmine (Prostigmin); associated with an unstable deoxyribonucleic acid (DNA) sequence composed of varying numbers of CTG triplet repeats (which allows a specific molecular test for this disorder).

**Ocular:** Mild ptosis (occasionally); myotonic cataract with small, dotlike subcapsular cortical opacities during early stage, with polychromatic properties on biomicroscopic examination; corneal epithelial dystrophy; loss of corneal sensitivity; tapetoretinal degeneration; macular red spot; macular degeneration; chorioretinitis; pilomatrixomas; ocular hypotony; pattern pigmentary changes; abnormal saccades.

**Clinical:** Progressive muscular atrophy with selection of certain muscles (mainly stern-oclidoemastoid, temporalis, dorsiflexor muscles of the ankle, anterior oblique); myotonia; bland facial expression; speech disturbance due to involvement of vocal cords and palatal muscles; dysphagia; endocrine disturbances.


**Myxomas, Spotty Pigmentation, and Endocrine Overactivity Syndrome (NAME Syndrome)**

**General:** Autosomal dominant.

**Ocular:** Eyelid myxomas; pigmented lesions of the caruncle or conjunctival semilunar fold.

**Clinical:** Cardiac, cutaneous, or mammary myxomas; acromegaly; adrenal, pituitary, and testicular neoplasms; Cushing syndrome; sexual precocity; spotty skin pigmentation.


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**Naegeli Syndrome (Melanophoric Nevus Syndrome; Franceschetti-Jadassohn Syndrome; Naegeli Incontinentia Pigmenti; Reticular Pigmented Dermatosis)**

**General:** Autosomal dominant; separate entity from incontinentia pigmenti (Bloch-Sulzberger syndrome) based on mode of inheritance; blisters and inflammatory lesions of skin as seen in incontinentia pigmenti not present in Naegeli syndrome; skin pigmentation appears at age 2 years.

**Ocular:** Nystagmus; strabismus; pseudoglioma; papillitis; optic atrophy.

**Clinical:** Reticular pigmentary skin changes; hypohidrosis; defective teeth with yellow spotting; keratoderma of palms and soles; disturbed regulation of temperature by reduction of number of sweat glands.


**Naffziger Syndrome (Scalenus Anticus Syndrome)**

**General:** Compression of brachial plexus and subclavian artery by scalenus anticus muscle; symptoms vary from mild, with remissions and exacerbations, to severe.

**Ocular:** Enophthalmos; ptosis (unilateral); small pupil.

**Clinical:** Weakness of ipsilateral hand grip; reduced ipsilateral biceps reflex; diminution of pulse volume on affected side; numbness and coldness in hand and fingers.


**Nager Syndrome (Nager Acrofacial Dysostosis) 868**

**General:** Rare congenital syndrome characterized by mandibulofacial dysostosis with associated radial defects.

**Ocular:** Downward slanting palpebral fissures; absent eyelashes in the medial third of the lower lids.

**Clinical:** Mandibular and malar hypoplasia; dysplastic ears with conductive deafness; variable degrees of palatal clefting; upper limb malformation (often bilateral hand deformities).


**Nance-Horan Syndrome 869**

**General:** Rare; X-linked oculodental trait; occurs in both sexes; present from birth.

**Ocular:** Posterior sutural cataracts in females; zonular cataracts in males; decreased visual acuity; nystagmus; microcornea; punctate opacities of cornea; affected males have dense nuclear cataracts and frequently microcorneas; carrier females may show posterior Y sutural cataracts with small corneas and only slightly reduced vision.

**Clinical:** Dental anomalies, including supernumerary central incisors; short fourth metacarpals; diastemata in females.


**NARP Syndrome 870**

**General:** Neurogenic weakness (N), ataxia (A), and retinitis pigmentosa (RP) syndrome.

**Ocular:** Retinitis pigmentosa; bull's-eye maculopathy; salt-and-pepper retinopathy.

**Clinical:** NARP syndrome patients develop ataxia, weakness, and have retinitis pigmentosa, causing gradual visual field constriction.


**Negative Acceleration Syndrome (Hydrostatic Pressure Syndrome; Supersonic Bailout Syndrome) 871**

**General:** At rapid deceleration, blood volume is shifted to the brain and face of the individual due to rapid elevation of intravascular blood pressure; the abrupt rise in hydrostatic pressure simultaneous with the decelerative force results in the signs of cerebral concussion, confusion, retrograde amnesia, and circulatory shock.

**Ocular:** Periorbital edema; ecchymosis of lids; transient visual loss; subconjunctival hemorrhages; retinal hemorrhages and exudates; retinal arteriolar spasm.

**Clinical:** Mental confusion; shock; possible perforation of internal organs.


Nelson Syndrome

**General:** Caused by elevated, incompletely suppressible, or nonsuppressible levels of circulating adrenocorticotropic hormone (ACTH) and possibly melanocyte-stimulating hormone; originally reported as hyperpigmentation with associated pituitary inactivity following bilateral adrenalectomy for Cushing syndrome; appears that age at the time of adrenalectomy is an important predictive factor for development of this disorder.

**Ocular:** Progressive visual deterioration; recurrent visual field defects with bilateral hemianopsia caused by a pituitary mass diagnosed as chromophobe adenoma.

**Clinical:** Hyperpigmentation; intermittent headaches.


Nematode Ophthalmia Syndrome (Visceral Larva Migrans Syndrome; Toxocariasis)

**General:** Usually found in children; invasion by larvae of *Toxocara canis* and *Toxocara cati* of viscera and eyes; pronounced eosinophilia; as many as 30% of asymptomatic children demonstrate serologic evidence of prior *Toxocara* infestations.

**Ocular:** Leukocoria; uveitis; cataract; marked vitreous reaction with large floaters; choroiditis; large, cystlike white masses extending into vitreous; optic neuritis; papillitis; strabismus; hemorrhagic, exudative, or granulomatous retinitis; retinal detachment; endophthalmitis; larvae present in the cornea.

**Clinical:** Hepatosplenomegaly; pulmonary infiltration; fever; cough; lack of appetite.


Neonatal Hemolytic Disease of Hyperbilirubinemia (Kernicterus; Bilirubin Encephalopathy)

**General:** Condition with severe neural symptoms associated with high levels of bilirubin in the blood.

**Ocular:** Disturbance of supranuclear vertical gaze; mild ocular motility defects; involuntary levator palpebrae inhibition.

**Clinical:** Athetosis; hearing loss; possible central nervous system (CNS) damage, characterized by deep yellow staining of the basal nuclei, globus pallidus, putamen and caudate nucleus, cerebellar and bulbar nuclei, and gray substance of the cerebrum.


Nephrotic Syndrome (Lipoid Nephrosis; Idiopathic Nephrotic Syndrome; Epstein Syndrome)

**General:** Unknown etiology, although some level of connection seems to exist between use of various drugs (penicillamine, heavy metals, trimethadione) and certain disease entities, such as intercapillary glomerulosclerosis, renal vein thrombosis, renal amyloidosis, and congestive heart failure; age at onset usually 1 to 6 years.

**Ocular:** Diffuse periorbital swelling; lid edema (mainly upper lids) more pronounced in the morning; uveitis; retinal edema around optic disk.

**Clinical:** Impaired renal function (depending on severity of renal involvement); anasarca (generalized and changing with time).


Neurilemoma (Schwannoma; Neurinoma)  876

General: Slow-growing encapsulated neoplasm from the Schwann cells of nerves; seen most frequently with patients with von Recklinghausen disease.

Ocular: Ptosis; exophthalmos; visual loss; pupillary dilation; lacrimal sac mass.

Clinical: Facial numbness; retroorbital headaches; intermittent pain radiating from the distribution of the appropriate sensory nerve branch.


Neuroblastoma  877

General: Highly malignant solid tumor arising from undifferentiated sympathetic neuroblasts of the adrenal medulla, sympathetic ganglia, ectopic adrenal, and theoretically the ciliary ganglion; autosomal dominant.

Ocular: Ptosis; exophthalmos; propotis; optic atrophy; optic neuritis; papilledema; metastatic tumor of orbit; retinal hemorrhage; convergent strabismus; paralysis of sixth or seventh nerve; nonreactive pupil; primary differentiated neuroblastoama of the orbit also has been reported; tonic pupils; microphthalmia; choroidal metastases (rare); iris metastases (rare).

Clinical: Skeletal metastasis to the cranium.


Neurocutaneous Syndrome  878

General: Triad of linear nevus sebaceous; seizures; mental retardation.

Ocular: Colobomas of irides and choroid; nystagmus; keratoconus; corneal vascularization; optic glioma; epibulbar choristomas; connective tissue nevi of the eyelids.

Clinical: Multiple nevi; seizures; mental retardation; failure to thrive; hydrocephalus; deformities of skull; lipoma of the cranium; alopecia of the scalp.


Neurodermatitis (Lichen Simplex Chronicus)  879

General: Skin altered due to chronic rubbing or scratching.

Ocular: Keratoconjunctivitis; lid edema; lid pigmentation; lid lichenification; atopic cataracts; keratoconus.

Clinical: Pruritus; dermatitis; seborrheic dermatitis; contact dermatitis; lichenification of skin; skin hyperpigmentation.

Neuronal Ceroid Lipofuscinosis (Neuronal Intranuclear Inclusion Disease; NIID)  880

General: Probably autosomal recessive.

Ocular: Nystagmus; sluggish pupil reaction; restricted ocular movements; optic disk pallor; loss of nerve fiber around macula; loss of pigment; widespread loss of photoreceptor function with abnormal electroretinogram.

Clinical: Slurred speech; extrapyramidal and lower motor neuron abnormalities; atrophy of skeletal muscles; bronchopneumonia; cerebral deterioration.


Neu Syndrome  881

General: Microcephaly-growth retardation-flexion deformities; both sexes affected; autosomal recessive; early death.

Ocular: Hypertelorism; absent eyelids.

Clinical: Flexion deformities; overlapping fingers; rocker-bottom feet; protruding heels; toe syndactyly; microcephaly; short neck; tiny nose; brain atrophy.


Newcastle Disease (Fowlpox)  882

General: Acquired directly by people handling chickens (see Parinaud Oculoglandular Syndrome); self-limiting conjunctivitis caused by a paramyxovirus.

Ocular: Acute follicular conjunctivitis, unilateral; keratitic precipitates; lid edema; decreased accommodation and visual acuity.

Clinical: Fatigue; fever; headache; pulmonary complications; preauricular lymphadenopathy.


Niacin Overdose (Nicotinic Acid Overdose)  883

General: B vitamin used in large doses to lower serum cholesterol and triglyceride levels or as a vasodilator.

Ocular: Cystoid maculopathy without leakage on fluorescein angiography, usually resolves when patient stops taking niacin.

Clinical: Facial flushing; gastric irritation; liver function impairment.


Nicolau Syndrome (Nicolau-Hoigne Syndrome)  884

General: First described as a nonallergic reaction following injection of bismuth; assumed to be caused by emboli of medication inadvertently introduced into an artery.

Ocular: Visual loss, depending on degree of involvement of retinal arteries.

Clinical: Tachycardia; acoustic sensations; somnolence; motoric irritation; sudden pain in extremities and abdomen; pallor, cyanosis, and edema of extremities; shock; paresis-paralysis; arterial hypotension.


Nieden Syndrome (Telangiectasia-Cataract Syndrome)

**General:** Etiology unknown; familial occurrence; onset from birth.

**Ocular:** Sparse eyebrows; glaucoma; dyscoria; defects of iris mesenchyme; bilateral cataract (cortical or mature).

**Clinical:** Telangiectasia of face and upper extremities; pigmented changes of the neck; thick, atrophic skin; heart enlargement; congenital valvular defect.


Nielsen Syndrome (Exhaustive Psychosis Syndrome; Neuromuscular Exhaustion Syndrome)

**General:** Chronic infections; postoperative phases frequently are associated with extreme stress and fatigue; similar manifestations often have been reported during and after prolonged combat; develops subacutely after severe overexertion during a period of euphoria.

**Ocular:** Paralysis of extraocular muscles; diplopia.

**Clinical:** Physical exhaustion; extreme weakness; delirium; fascicular twitching; poor attention; restlessness; lack of concentration; irritability; depression; anxiety; pain in muscles; weight loss; muscle flaccidity; absence of deep reflexes.


Niemann-Pick Syndrome (Essential Lipoid Histiocytosis; Sea-Blue Histiocytosis; Sphingomyelinase Deficiency)

**General:** Phospholipidosis with degeneration of ganglion cells of CNS and lipid storage involving entire reticuloendothelial system and parenchymatous tissue; autosomal recessive; occurs during first few months of life; similar to Tay-Sachs syndrome; predisposition for Jews (see Tay-Sachs Syndrome); lysosomal storage disease resulting from diminished activity or deficiency of sphingomyelinase.

**Ocular:** Vision may be reduced but usually not complete blindness; cherry-red spot of macula, similar to that of Tay-Sachs syndrome; progressive optic atrophy; supranuclear ophthalmoplegia; periorbital fullness; macular halo syndrome; abnormal visual evoked potentials.

**Clinical:** Mental retardation; extensive hepatosplenomegaly; epileptic seizures; progressive physical deterioration; deafness; skin pigmentation; abdominal enlargement.


9p- Syndrome

**General:** Congenital mental retardation syndrome due to a 9p deletion.

**Ocular:** Mongoloid (down-slanting) eyes; usually glaucoma.

**Clinical:** Mental retardation; sociable personality; trigonocephaly; wide, flat nasal bridge; anteverted nostrils; long upper lip; short neck; long digits; predominance of finger whorls; choanal atresia; usually seizures.


Nocardiosis

**General:** Aerobic Actinomycetaceae that may cause a chronic suppurative process; aerobic Gram-positive filamentous bacteria with branching pattern which resemble fungi.

**Ocular:** Conjunctivitis; keratitis; corneal ulcer; uveitis; lid involvement; orbital cellulitis; endophthalmitis; glaucoma; external ophthalmoplegia; scleritis; canaliculitis; preseptal cellulitis.

**Clinical:** Granuloma; draining sinuses; brain abscess; meningitis.


Nonne-Milroy-Meige Disease (Chronic Hereditary Lymphedema; Milroy Disease; Meige Disease; Meige-Milroy Syndrome; Nonne-Milroy Syndrome; Chronic Hereditary Edema; Chronic Hereditary Trophedema; Chronic Trophedema; Elephantiasis Congenita Herediteria; Familial Hereditary Edema; Hereditary Edema; Idiopathic Hereditary Lymphedema; Pseudoedematous Hypodermal Hypertrophy; Pseudoelephantiasis Neuroarthritica; Oromandibular Dystonia; Blepharospasm-Oromandibular Dystonia; Congenital Trophedema; Tropholymphedema; Trophoneurosis; Elephantiasis Arabum Congenita)

**General:** Autosomal dominant; prevalent in females; two types: praecox at birth to 35 years and tarda after age 35 years.

**Ocular:** Lid and conjunctival edema; blepharoptosis; distichiasis; strabismus; buphthalmos; ectropion.

**Clinical:** Lymphedema; mandibulofacial dysostosis; unilateral or bilateral edema of ankle ascending to the knee and eventually above; rough, pigmented skin over swollen parts.


Noonan Syndrome (Male Turner Syndrome)

**General:** Similar to Turner syndrome, but with normal chromosomlal analysis; X-linked dominant inheritance; X-linked dominant phenotype.

**Ocular:** Hypertelorism; exophthalmos; ptosis (unilateral or bilateral); antimongoloid-slanting palpebral fissure; myopia; keratoconus; optic disk coloboma.

**Clinical:** Valvular pulmonary stenosis; short stature; webbed neck; low hairline in the back; cubitus valgus; deformed chest wall; micrognathia; low-set ears; mild mental retardation.


Nothnagel Syndrome (Ophthalmoplegia-Cerebellar Ataxia Syndrome)

**General:** Lesion of superior cerebellar peduncle, red nucleus, and emerging oculomotor fibers, such as pineal tumor, or tumor or vascular disturbance in corpora quadrigemina or vermis cerebelli (see Bruns Syndrome).

**Ocular:** Oculomotor paresis; gaze paralysis most frequently upward, combined with some degree of internal or external ophthalmoplegia.

**Clinical:** Cerebellar ataxia; poor upper extremity movements; neoplasia; infarction; midbrain lesion.


Nystagmus Blockage Syndrome (NBS)

**General:** Congenital (see Ethan Syndrome, Primary; Ethan Syndrome Secondary; Nystagmus Compensation Syndrome); convergence is used to diminish nystagmus.

**Ocular:** Esotropia; nystagmus; amblyopia; most patients with this syndrome prefer to fixate with one eye, but others show alternating fixation.

**Clinical:** Abnormal head position.


Nystagmus Compensation Syndrome (NCS)

**General:** Congenital (see Ethan Syndrome, Primary; Ethan Syndrome Secondary; Nystagmus Blockage Syndrome).

**Ocular:** Esotropia; amblyopia; onset may be preceded by manifest nystagmus; abnormal head posture toward the adducted fixing eye; nystagmus reduced or absent, with the fixing eye adducted.

**Clinical:** Abnormal head position.


Nystagmus, Congenital (Congenital Idiopathic Nystagmus)

**General:** Autosomal dominant; pattern of inheritance in congenital nystagmus, whether of the "motor" or "sensory" type, may be autosomal dominant, recessive, or sex-linked.

**Ocular:** Vertical and horizontal nystagmus; this nystagmus occasionally is vertical or torsional; in addition, periodic alternating, downbeat, and see-saw nystagmus may be present at birth; normal electroretinogram.

**Clinical:** None.


Nystagmus, Hereditary Vertical

**General:** Autosomal dominant.

**Ocular:** Motor-type vertical and horizontal nystagmus; hyperactive vestibuloocular response; strabismus.

**Clinical:** Mild ataxia; poor line walking.


Nystagmus, Primary Hereditary (Congenital Nystagmus)

**General:** Autosomal recessive, sex-linked or irregular dominant; may be associated with albinism.

**Ocular:** Horizontal nystagmus; myopia.

**Clinical:** Head spasms; carpopedal spasms (Trousseau sign); Chvostek sign.


Nystagmus, Voluntary

**General:** Autosomal dominant; usually is purely horizontal, but it may be vertical or-torsional.

**Ocular:** Voluntary rapid to-and-fro synchronous movements of eyes.

**Clinical:** Simultaneous head tremor has been associated with this condition.


Obesity-Cerebral-Ocular-Skeletal Anomalies Syndrome

**General:** Rare, autosomal recessive disease; similar to Prader-Willi and Laurence-Moon-Bardet-Biedl syndromes (see Prader-Willi Syndrome; Laurence-Moon-Bardet-Biedl Syndrome).

**Ocular:** Microphthalmia; antimongoloid slant of lid fissure; asymmetrical size of fissure; strabismus; myopia; iris and chorioretinal colobomata; mottled retina; prominent choroidal vessels.
**Clinical:** Obesity (mid-childhood onset); hypotonia; mental retardation; craniofacial anomalies with microcephaly; tapering extremities; hyperextensibility at elbows and proximal interphalangeal joints; cubitus valgus; genu valgum; simian creases; syndactyly.


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**Ocular Dominance**

**General:** Autosomal dominant; 97% of people with normal vision have a sighting-dominant eye; experimental data suggest a relationship between eye dominance and head tilt.

**Ocular:** Sighting-dominant eye.

**Clinical:** None.


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**Ocular Myopathy with Curare Sensitivity**

**General:** Autosomal recessive.

**Ocular:** Static ophthalmoparesis.

**Clinical:** Limb weakness; sensitivity to tubocurarine.


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**Ocular Toxoplasmosis (Toxoplasmic Retinochoroiditis; Toxoplasmosis)**

**General:** Parasite infestation caused by *Toxoplasma gondii*; cell-mediated immunity is believed to be the major defense mechanism against *Toxoplasma* infection; ocular toxoplasmosis occurs in approximately 1% of patients with acquired immunodeficiency syndrome (AIDS); AIDS-related toxoplasma retinochoroiditis may have several atypical clinical manifestations.

**Ocular:** Keratitis; uveitis; optic atrophy; papillitis; anisocoria; persistent pupillary membrane; focal retinochoroiditis; scleritis; cataract; microphthalmos; myopia; nystagmus; esotropia.

**Clinical:** Cysts are seen in many organs, including brain and muscle; hydrocephalus; intracerebral calcification; various CNS complaints.


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**Ocular Vaccinia**

**General:** Occurs from virus used for smallpox vaccination; transferred from vaccination site to eye.

**Ocular:** Chemosis; nonfollicular, catarrhal, or purulent conjunctivitis; punctate epithelial keratitis; corneal vascularization; lid edema; vesicles of lid; uveitis; choroiditis; extraocular muscle palsies; optic neuritis; orbital cellulitis; ocular vaccinia may mimic signs of herpes simplex virus, varicella-zoster virus, and acanthamoeba keratitis.

**Clinical:** Dermatitis; encephalitis.


**Oculocerebellar Tegmental Syndrome**

**General:** Vascular lesion of mesencephalon with softening in peduncular area.

**Ocular:** Paralysis of associated ocular movements (internuclear anterior ophthalmoplegia).

**Clinical:** Sudden onset of hemiplegia with rapid recovery; bilateral cerebellar syndrome.


**Oculocerebral Syndrome with Hypopigmentation (Amish Oculocerebral Syndrome; Cross Syndrome)**

**General:** Autosomal recessive.

**Ocular:** Spastic ectropion; microphthalmos; enophthalmos; microcornea; corneal opacification; corneal vascularization; palpebral conjunctival injection; narrow lid fissures; aniridia; nystagmus; bilateral optic atrophy.

**Clinical:** Spastic diplegia; cutaneous hypopigmentation; mental retardation; hypogonadism; growth retardation; developmental defects of the CNS, such as cystic malformation of the posterior fossa of the Dandy-Walker type.


**Oculocerebrocutaneous Syndrome (Delleman Syndrome)**

**General:** Congenital; possibly autosomal recessive or a result of an environmental problem; Dutch descent appears to be a predisposing factor; in most cases, involvement is unilateral, with left to right 2: 1.

**Ocular:** Orbital cyst; absence of orbital structures; microphthalmos; anophthalmia.

**Clinical:** Cerebral malformations; focal dermal hypoplasia; aplasia; epilepsy; developmental delay; cranial skin appendages; cutaneous punched-out lesions.


**Oculodental Syndrome (Peters Syndrome; Rutherford Syndrome)**

**General:** Similar to Rieger syndrome and Meyer-Schwickerath-Weyer syndrome; Peters syndrome inherited as autosomal recessive with defect of corneogenetic mesoderm characterized by incomplete separation of lens vesicle, causing central opacities of cornea, shallow anterior chamber, synechiae, and remnants of pupillary membrane; anterior pole cataract; Rutherford syndrome inherited as autosomal dominant; exhibits iris and dental anomalies and mental retardation.

**Ocular:** High myopia; corneoscleral staphyloma; aniridia; macrocornea; opacities of the corneal margin; ectopia lentis with deposits of pigment; macular pigmentation; large excavation of optic nerve with atrophy.

**Clinical:** Oligodontia; microdontia; hypoplasia of enamel; abnormal tooth positions; hypertrophy of gums; failure of tooth eruption.

Oculogastrointestinal Muscular Dystrophy 908

**General:** Autosomal recessive; visceral myopathy with external ophthalmoplegia; intestinal pseudo-obstruction with external ophthalmoplegia.

**Ocular:** Ptosis; ophthalmoplegia.

**Clinical:** Chronic diarrhea; abdominal distention; diffuse abdominal pain; impaired motility of the lower esophagus; demyelinating and axonal neuropathy with focal spongiform degeneration of the posterior columns; proximal muscle weakness and atrophy; primary myopathy of the smooth muscles of the stomach and intestine; dilated duodenum and jejunum; recurrent intestinal obstruction in childhood or adolescence.


**Oculo-Oro-Genital Syndrome (Riboflavin Deficiency Syndrome; Gopalan II Syndrome; Jolliffe Syndrome)** 909

**General:** Vitamin B deficiency and possible vitamin A deficiency; prognosis good with diet therapy.

**Ocular:** Conjunctivitis, varying from mild to severe; keratitis; optic atrophy; corneal vascularization.

**Clinical:** Stomatitis; glossitis; scrotal dermatitis with pruritus, erythema, erythema of pharynx and soft palate; small sensitive ulcers of buccal membranes; diarrhea; fatigue; muscular weakness; painful feet with erythema, exfoliation, and ulceration; burning; itching; mental depression; dizziness; oral mucosa becomes pale and macerated with fissuring of skin.


**Oculo-Osteocutaneous Syndrome** 910

**General:** Autosomal recessive.

**Ocular:** Strabismus; myopia; distichiasis; nystagmus; lenticular opacities.

**Clinical:** Short stature; brachydactyly; hypoplastic maxilla; scanty hair; hypopigmentation; mental retardation.


**Oculo-Oto-Oro-Reeno-Erythropoietic Disease** 911

**General:** Etiology unknown; slowly progressive course; ocular lesions similar to those of tuberous sclerosis, but no other signs of that disorder.
Ocular: Exotropia; progressive visual loss to complete blindness; secondary glaucoma with iris bombe; iridocyclitis; chronic uveitis; vitreous opacities; cataract; retinal hemorrhages and exudates.

Clinical: Anemia; atypical development of periodontia and teeth with caries; high-arched palate; abdominal colic; genu valgum and pectus excavatum; dizziness.


Oculopalatocerebral Dwarfism (OPC Dwarfism)

General: Autosomal recessive; persistent hyperplastic primary vitreous.

Ocular: Persistent hypertrophic primary vitreous; microphthalmos; leukocoria; retrorenal fibrovascular membrane.

Clinical: Microcephaly; mental retardation; spasticity; cleft palate; short stature.


Oculopalatoskeletal Syndrome

General: Autosomal recessive; developmental defect of eye and occipital bone; skeletal abnormalities.

Ocular: Triad of blepharophimosis, blepharoptosis, and epicanthus inversus; corneal stromal opacities; limitation of upward gaze.

Clinical: Cleft lip/palate; spina bifida occulta; cranial asymmetry; radioulnar synostosis; short fifth finger; abnormality of occipital bone.


Oculopharyngeal Syndrome (progressive Muscular Dystrophy with Ptosis and Dysphagia; Oculopharyngeal Muscular Dystrophy)

General: Etiology unknown; autosomal dominant inheritance; no CNS pathology; muscles of pharynx, hypopharynx, and proximal third of esophagus involved with myopathy; onset late in life; progressive hereditary myopathy in which the levator palpebrae and pharyngeal muscles are selectively involved; progressive usually symmetrical blepharoptosis with or without dysphagia appears in the fifth decade.

Ocular: Ptosis.

Clinical: Dysphagia; occasionally, weakness of facial muscles.


Murphy SF, Drachman DB. The oculopharyngeal syndrome. JAMA 1968; 203:1003.


Oculorenocerebellar Syndrome (ORC Syndrome)

General: Autosomal recessive.

Ocular: Progressive tapetoretinal degeneration with loss of retinal vessels.
Clinical: Mental retardation; continuous jerky movements; spastic diplegia; glomerulopathy with most renal glomeruli completely sclerosed.


Oguchi Disease

General: Autosomal recessive; usually Japanese; form of congenital hemeralopia; one form of essential, congenital night blindness.

Ocular: Fundus has white-gray coloration, especially around the optic disk and in macular region after light exposure (30 minutes to 1 hour); color sometimes is more brown; after 2 to 3 hours in the dark (Mizuo-Nakamura phenomenon), the posterior pole appears normal; the pathogenesis of Mizuo phenomenon has been postulated to be secondary to an excess of extracellular potassium in the retina as a result of a decreased potassium scavenging capacity of retinal Muller cells; central visual acuity and visual fields normal; night blindness always present.


OHABA Syndrome (Ophthalmoplegia, Hypotonia, Ataxia, Hypoacusis, Athetosis)

General: Ophthalmoplegia, hypotonia, ataxia, hypoacusis, athetosis (OHARA) are distinguishing symptoms; sudden onset of deafness at an age after patient has learned to speak.

Ocular: Kernicterus; strabismus; nystagmus; ocular migraine; ophthalmoplegia; vascular spasm in branches of the ophthalmic artery; intact convergence.

Clinical: Hemiplegia; athetosis; choreoathetosis; tremor; hypoxia; corticospinal tract disease; diabetes mellitus; ataxia; medulloblastoma; asynergia; dysdiadochokineses; Holmes rebound phenomenon; acute cerebellar lesion; dysmetria; dysarthria; Fox syndrome; vascular occlusion; congenital athetosis.


Okihiro Syndrome

General: Autosomal recessive syndrome of Duane syndrome (retraction syndrome).

Ocular: Narrowing of palpebral fissures on adduction, widening on abduction; primary global retraction.

Clinical: May be associated with craniofacial abnormalities and various associated syndromes, such as Duane syndrome, cervico-oculo-auditory syndrome, acrorenalocular syndrome, cat's-eye syndrome; association with cardiac defects, urinary tract anomalies, Duane anomaly associated with mental retardation, thenar hypoplasia, and radial ray abnormalities.


Oliver-McFarlane Syndrome (Trichomegaly Syndrome)

General: Rare syndrome.

Ocular: Trichomegaly; pigmentary retinal degeneration.
Clinical: Prenatal onset growth failure; anterior pituitary deficiencies; peripheral neuropathy; mental retardation, sparse scalp hair, endocrinologic deficiencies, and koilonychias may be found.


Olivopontocerebellar Atrophy III (OPCA III; OPCA with Retinal Degeneration) 920

General: Autosomal dominant; neurologic lesion; dominant with variable penetration.

Ocular: Retinopathy variable: peripheral, macular, and circumpapillary; retinal degeneration; blindness; external ophthalmoplegia; variable electroretinogram function.

Clinical: Ataxia.


Onchocerciasis Syndrome (River Blindness; Onchocerca Volvulus Infestation) 921

General: Nematode infestation; positive diagnosis made with microfilariae from a skin biopsy and the presence of microfilariae in anterior chamber; infection results from the bite of the blackfly genus Simulium; recent findings suggest an autoimmune etiology for the occurrence of chorioretinopathy.

Ocular: Punctate keratitis (fluffy opacities); sclerosing keratitis; chronic iritis; pear-shaped pupil deformation; chorioretinitis; optic atrophy; uveitis; iris atrophy; papillitis; glaucoma; retinal degeneration; perivascular sheathing; microfilariae present in the anterior chamber, cornea, and vitreous; pannus; synechiae; cataracts; secondary glaucoma.

Clinical: Subcutaneous nodules; pruritus; atrophic skin changes; pretibial pigmentation; dermatitis; lymphadenitis.


One-and-a-Half Syndrome 922

General: Lesions in medial longitudinal fasciculus and paramedian pontine reticular formation; lateral gaze palsy on the side of the lesion and contralateral internuclear ophthalmoplegia; most common etiologies are ischemic infarction, demyelinating lesions, compressive lesion, and infections.

Ocular: Paralysis of abduction; lateral gaze palsy; nystagmus; ocular bobbing; exotropia; esotropia.

Clinical: Dysarthria; dysphagia; hemiparesis; multiple sclerosis frequently associated in young patients with this condition.


Ophthalmodynia Hypertonica Copulationis Syndrome  
**General:** Provocation of a diagnosed and otherwise controlled angle-closure glaucoma by prolonged sexual intercourse in the prone position.

**Ocular:** Acute attacks of angle-closure glaucoma during copulation in prone position.

**Clinical:** Nausea; vomiting.


Ophthalmopathic Syndrome (Thyrohypophysial Syndrome)  
**General:** Exophthalmos, usually bilateral; pituitary hypersecretion or enhanced tissue sensitivity to exophthalmos-producing factor that has been separated from thyroid-stimulating hormone.

**Ocular:** Exophthalmos; paralysis of ocular muscles.

**Clinical:** Thyroid disorders.


Ophthalmoplegia, Familial Static  
**General:** Autosomal dominant; forms include internal, external, and total ophthalmoplegia.

**Ocular:** Ptosis; almost completely fixed eyes; nystagmoid movements; unequal pupils; pupil paralysis.

**Clinical:** None.


Ophthalmoplegia, Progressive External  
**General:** Autosomal recessive; progressive limitation of ocular motility with clinical sparing of pupillary function; underlying pathogenesis is secondary to a mitochondrial cytopathy; appearance of ragged red fibers in the abnormal muscles is primarily caused by mitochondrial accumulations beneath the plasma membrane and between the myofibrils.

**Ocular:** Oculopharyngeal muscular dystrophy; retinitis pigmentosa; progressive external ophthalmoplegia; some patients show only ptosis and ophthalmoplegia; most patients have multisystem involvement.

**Clinical:** Heart block; ataxia.


Ophthalmoplegia, Progressive External, with Ragged Red Fibers  
**General:** Autosomal dominant.

**Ocular:** Progressive ophthalmoplegia.
Clinical: Ragged red fibers in skeletal muscle from the extremities; subsarcolemmal clusters of mitochondria containing paracrystalline inclusions.


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**Ophthalmoplegia, Progressive External, with Scrotal Tongue and Mental Deficiency**

General: Autosomal dominant.

Ocular: Progressive external ophthalmoplegia; progressive chorioretinal sclerosis; bilateral ptosis; convergence paresis; myopia; optic atrophy; retinitis pigmentosa.

Clinical: Bilateral facial weakness; linguæ scrotalis; mental retardation; cerebellar ataxia; weakness and spasticity of the limbs.


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**Ophthalmoplegic Migraine Syndrome**

General: Symptoms produced by ipsilateral herniation of hippocampal gyrus of temporal lobe through incisura tentorii; dependent upon unilateral cerebral edema due to vascular or vasomotor phenomena, intracranial aneurysm, or tumor; incidence may be greater in women with the initial attack in the first decade of life; pathogenesis is unclear, but it is likely secondary to ischemia of the oculomotor nerve.

Ocular: Severe unilateral supraorbital pain; ptosis; transitory partial or complete homolateral oculomotor paralysis; fourth or sixth nerve occasionally involved; retinal hemorrhages; papilledema (may be bilateral); moderate to severe headache with partial to complete cranial nerve III paresis including the pupil; more than one oculomotor nerve may be affected.

Clinical: Migraine headache, not present in all instances; dizziness; diminution in sense of smell; hypalgesia contralateral side of face; nausea/vomiting may be present; recurrent sinus arrest.


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**Ophthalmoplegic Neuromuscular Disorder with Abnormal Mitochondria**

General: Autosomal recessive.

Ocular: Ptosis; external ophthalmoplegia.

Clinical: Involvement of cranial nerves and skeletal muscles; morphologic alterations of mitochondria; diffuse low-density deep cerebral white matter; weak limbs.


Ophthalmoplegic Retinal Degeneration Syndrome (Barnard-Scholz Syndrome)

**General:** Onset at all ages (see Kearns-Sayre Syndrome).

**Ocular:** Unilateral or bilateral progressive weakness of muscles of eyelids, up to severe ptosis; progressive ocular myopathy up to complete ophthalmoplegia; retinitis pigmentosa.

**Clinical:** Facial, neck, and shoulder muscle weakness; hearing defects; heart block.


Kiloh LG, Nevin S. Progressive dystrophy of the external ocular muscles (ocular myopathy). Brain 1951; 774: 115-143.

Optic Atrophy, Cataract, and Neurologic Disorder

**General:** Autosomal dominant; similar to syndromes of Behr, Marinesco, Sjögren, and Friedreich, which are autosomal recessive.

**Ocular:** Cataract; optic atrophy.

**Clinical:** Neurologic disorder.


Optic Atrophy with Demyelinating Disease of CNS

**General:** Autosomal dominant; demyelinated optic nerves appear smaller than normal and are pale white or gray in color; blood vessels may seem to be less prominent than normal.

**Ocular:** Optic neuritis; Leber optic atrophy.

**Clinical:** Ataxia; leg weakness; dysarthria; hemiparesis.


Optic Atrophy, Juvenile (Optic Atrophy, Congenital; Kjer-Type Optic Atrophy; Optic Atrophy, Kjer-Type; OAK Syndrome [Optic Atrophy, Kjer Type])

**General:** Autosomal dominant; dominant pattern distinguishes it from Leber optic atrophy; insidious onset; onset in childhood; pathogenetic mechanism may be primary degeneration of retinal ganglion cells.

**Ocular:** Central scotoma; color defects; choroidal sclerosis; optic neuritis; temporal optic atrophy; aggregation of retinal pigment epithelium; tortuosity of retinal arteries and veins; reduced central vision; retinal lesions; may present with mild-to-moderate reduction of visual acuity with 50% of patients having vision between 20/60 and 20/200; visual field defect associated may be a central, paracentral, or cecocentral scotoma.

**Clinical:** Keratosis pilaris on the extremities; approximately 10% of patients present with mental abnormalities and approximately 80% of patients with neural hearing loss.


Optic Atrophy, Nerve Deafness

**General**: Autosomal recessive.

**Ocular**: Degeneration of optic nerves.

**Clinical**: Degeneration of the acoustic nerves; progressive polyneuropathy, distally.


Optic Atrophy, Non-Leber-Type, with Early Onset

**General**: Sex-linked; onset early in life.

**Ocular**: Optic atrophy.

**Clinical**: Mental retardation; hyperactive knee jerks; absent ankle jerks, extensor plantar reflexes; dysarthria; tremor; dysdiadochokinesia; difficulty with tandem gait.


Optic Atrophy-Spastic Paraplegia Syndrome

**General**: Degenerative disorder of CNS associated with optic atrophy; sex-linked.

**Ocular**: Optic atrophy.

**Clinical**: Neurologic spastic paraplegia,


Optic Canal Syndrome

**General**: Caused by severe blow to the head, usually the face or occiput.

**Ocular**: Unilateral blindness (rarely bilateral); amaurotic pupillary paralysis; pathogenesis is a shearing or tearing injury to the nerve at the transition point from where it is fixed in the optic canal and the free intracranial portion.


Optic Disk Traction Syndrome

**General**: Optic disc traction with elevation associated with posterior vitreous detachment or vitreopapillary fibrous membrane may be associated with retinal surgery

**Ocular**: Central retinal vein occlusion; optic disk traction (vitreopapillary); localized retinal detachment

Optic Nerve Hypoplasia, Familial (Bilateral, Unilateral)  

**General:** Autosomal dominant; congenital defect of optic nerve and retina that occurs in both unilateral and bilateral forms; onset at birth; majority of cases are sporadic, although there are reports of familial cases; association of this condition with maternal ingestion of various substances, including quinine, lysergic acid diethylamide (LSD), and anticonvulsants.

**Ocular:** Optic nerve hypoplasia; diameter of optic disk is one third normal size; nystagmus; peripapillary halos; situs inversus of disk; strabismus; choroidal atrophy; coloboma of choroid and/or optic disk; blepharophimosis; ptosis; aniridia; ocular motor nerve palsy.

**Clinical:** CNS defects; chromosomal abnormalities; cerebral malformations; vascular hypertension.


Optic Pit Syndrome  

**General:** Congenital.

**Ocular:** Serous detachment of macula; situs inversus; peripapillary chorioretinal changes; cilioretinal vessels; large optic disk; tortuous retinal vessels; retinoschisis.

**Clinical:** None.


Oral-Facial-Digital Syndrome (OFDS)  

**General:** Group of syndromes characterized by congenital anomalies of the oral cavity, face, and digits; see-saw walking; hypertelorism strabismus.

**Ocular:** Chorioretinal coloboma, optic nerve coloboma, retinal hamartoma.

**Clinical:** Patients with OFDS have been known to have various congenital anomalies, including cleft palate, tongue hamartomas, bifid tongue, multiple hyperplastic frenula, hypoplastic nasal cartilage, syndactyly, polydactyly, brachydactyly, clinodactyly, and the ocular ophthalmologic findings; nine distinct types of OFDS have been described.


ORF Syndrome (Ecthyma Infectiosum)  

**General:** Infectious disease; transmitted between animals; worldwide distribution; human infection from sheep.

**Ocular:** Pigmentation of lids.

**Clinical:** Single or multiple lesions of hands and other parts of body; itching; fever; concurrent aseptic meningitis caused by enterovirus.

Ormond Syndrome (Retroperitoneal Fibrosis)  944

General: Periureteral fibrosis that constricts ureter; most frequent in males; average age at onset is 46 years in males and 32 years in females; etiology possibly is a fasciculitis of collagen disease; clinical variant of multiple fibrosclerosis.

Ocular: Exophthalmos; orbital pseudotumor; nongranulomatous panuveitis.

Clinical: Pain in back progressing to ureteral colic or abdominal pain without specific localization; vomiting; nausea; anorexia; malaise; weight loss; constipation; diarrhea; pyelonephritis.


Orzechowski Syndrome (Truncal Ataxia-Opsoclonia; Encephalitis-Opsoclonia- Tremulousness)  945

General: Etiology unknown; follows a benign upper respiratory infection; spontaneous resolution; opsoclonus has been described in patients with encephalitis with Epstein-Barr virus, mumps, and coxsackievirus infections.

Ocular: Opsoclonia; excessive blinking.

Clinical: Incapacitating postural tremulousness of the body; fever; headache.


Osteogenesis Imperfecta Congenita, Microcephaly, and Cataracts  946

General: Autosomal recessive.

Ocular: Cataracts; blue sclera; keratoconus.

Clinical: Brain abnormally small; multiple prenatal bone fractures; calvaria soft; shortening and bowing of lower limbs.


Ota Syndrome (Nevus of Ota; Oculodermal Melanocytosis; Nevus Fuscoceruleus-Ophthalmomaxillaris Syndrome)  947

General: Affects mainly black and Japanese populations; female preponderance (4:1); mode of transmission unknown; most frequently unilateral; pigmentary changes frequently spread during puberty, but no malignant transformation occurs; malignant transformation to melanoma in the uvea and orbit has been reported.

Ocular: Congenital benign periorbital pigmentation of brown, slate to bluish-black coloration, involving area of first and second (rarely third) division of trigeminal nerve; unilateral hyperchromic heterochromia iridis; possible scleral and conjunctival pigmentation; trabeculae heavily pigmented; slate-gray hyperpigmentation of fundus; optic disk pigmentation (occasionally).

Clinical: Pigmentation of temples, nose, forehead, and malar region; "Mongolian spot" in sacral area (present at birth but usually disappears after puberty).

Otocephaly

**General:** Birth defect; extreme malformation of first brachial arch characterized by almost complete aplasia of its parts.

**Ocular:** Bilateral anophthalmos.

**Clinical:** Mouth deformities; absence of lower jaw; joining of ears on the neck.


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**Outer Retinal Ischemic Infarction Syndrome**

**General:** Complication that may occur during the course of cataract extraction or closed vitrectomy due to obstruction of choroidal circulation.

**Ocular:** Acute loss of central and paracentral vision; whitening of the outer retinal layers in posterior fundus; mottled changes in the pigment epithelium.

**Clinical:** None.


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**Oxalosis (Primary Hyperoxaluria; Lepoutre Syndrome)**

**General:** Autosomal recessive and acquired forms; metabolic disorders with accumulation of oxalic acid in tissues; type I: glycolic aciduria, defect of 2-oxoglutarate/ glyoxylate carboligase; type II: glyceric aciduria, defect of D-glyceric dehydrogenase.

**Ocular:** Calcium oxalate deposits may be found in the retina; retinitis punctate albescens; macular degeneration of Stargardt; pigmentary retinopathy; black, geographic central macular subretinal patches; oxalate may deposit in the retinal pigment epithelium, outer plexiform layer, and nuclear layers of the retina.

**Clinical:** Recurrent calcium oxalate nephrolithiasis and nephrocalcinosis; progressive renal insufficiency; continuous excessive synthesis and excretion of oxalic acid; nausea; vomiting; abdominal pain; renal colic; calculi in urine; tetany.


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**Page Syndrome (Hypertensive Diencephalic Syndrome)**

**General:** Irritation of parasympathetic and sympathetic centers in diencephalon; intradermal histamine 0.25 mg reproduces syndrome; prognosis unpredictable; more frequent in women; onset age 18 to 30 years; spontaneous attacks lasting several minutes.

**Ocular:** Excessive lacrimation; arteriosclerotic and hypertensive fundus changes.
Clinical: Vasomotor blush over face, neck, and trunk, followed by perspiration; tachycardia; palpitations; elevated baseline blood pressure with additional 20 to 30 mm Hg rise during attacks; reduced respiration, perhaps labored; increased bowel sounds; salivation; sexual frigidity; tremor of hands and generalized tremulousness.


Paget Disease (Osteitis Deformans; Congenital Hyperphosphatemia; Hyperostosis Corticalis Deformans; Pozzi Syndrome; Chronic Congenital Idiopathic Hyperphosphatemia; Osteochalasis Desmalis Familiaris; Familial Osteoectasia)

General: Autosomal dominant; more frequent in men, but more severe in women; onset after age 40 years; characterized by diffuse cortical thickening of involved bones with osteoporosis, bowing deformities, and shortening of stature; osteogenic sarcoma not infrequent.

Ocular: Shallow orbits with progressive unilateral or bilateral proptosis palsy of extraocular muscles; corneal ring opacities; cataract; retinal hemorrhages; pigmentary retinopathy; macular changes resembling Kuhnt-Junius degeneration; angioid streaks; papilledema; optic nerve atrophy; blue sclera; exophthalmos.

Clinical: Skull deformities; kyphoscoliosis; hypertension and arteriosclerosis; muscle weakness; waddling gait; hearing impairment; osteoarthrosis.


Paget J. On a form of chronic inflammation of bones (osteitis deformans). Med Chir Trans (Lond) 1877; 60:37.


Palatal Myoclonus Syndrome

General: Vascular disorders of brainstem that involve inferior olive and olivodentate connection; continuous rhythmic contractions of the palate that occur 100 to 180 times per minute.

Ocular: Opsoclonia; ocular bobbing; nystagmus; torsional, pendular nystagmus; gaze-evoked nystagmus.

Clinical: Contraction of pharynx, larynx, tongue, floor of the mouth, neck, and diaphragm; nodding of head; tremor of hand; sleep apnea; multiple sclerosis.


Pallidal Degeneration, Progressive, with Retinitis Pigmentosa (Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, and Pallidal Degeneration; HARP Syndrome)  

**General:** Autosomal recessive; destruction of global pallida and reticular portions of substantia nigra; also may be associated with hypoprebetalipoproteinemia and acanthocytosis; various combinations of components of HARP syndrome may be caused by several distinct genetic diseases or may represent variable manifestations of a contiguous gene defect.

**Ocular:** Retinitis pigmentosa.

**Clinical:** Progressive extrapyramidal rigidity; dysarthria.


Pallister-Killian Syndrome (PKS; Mosaic Tetrosomy)  

**General:** Variable condition caused by a mosaic tetrasomy of chromosome 12p.

**Ocular:** Reported manifestations include hypertelorism, narrow palpebral fissures, prominent epicanthal folds, lower eyelid entropion, ptosis, sparse eyebrows, sparse eyelashes, hypoplastic supraorbital ridge, miotic pupils, sluggish pupils, iris atrophy/hypoplasia, iris heterochromia, cataracts, nystagmus, esotropia, exotropia, optic nerve atrophy, optic disks, fundus hypopigmentation, ambylophia, and retinal pigmentary mosaicism.

**Clinical:** Patients have "coarse" facial features, midface malformations, psychomotor delay, hypotonia, scalp hair sparsity, and variegated lightly and darkly pigmented skin.


Palpebral Coloboma-Lipoma Syndrome (Nasopalpebral Lipoma-Coloboma)  

**General:** Autosomal dominant; described in a Venezuelan family.

**Ocular:** Coloboma of upper and lower lids at junction between their middle and inner thirds; fat deposits of both upper lids; malposition of lacrimal puncta; hypertelorism; telecanthus.

**Clinical:** Broad nasal bridge; fatty accumulations on nasal bridge and nasolabial area; maxillary hypoplasia.


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Pancoast Syndrome (Hare Syndrome; Superior Pulmonary Sulcus Syndrome)

**General:** Mass occupying lesion in pulmonary apex; erosion of first three ribs frequent; primary bronchogenic carcinoma most frequent cause; symptomatology similar to lower radicular (Dejerine-Klumpke) syndrome and scalenus anticus (Naffziger) syndrome; Horner syndrome caused by involvement of sympathetic chain (see Dejerine-Klumpke Syndrome; Horner Syndrome; Naffziger Syndrome); also can be caused by locally invasive fungus such as *Cryptococcus neoformans* or lymphomatoid granulomatosis.

**Ocular:** Mild enophthalmos; ptosis; narrowing of the palpebral fissure; miosis.

**Clinical:** Pulmonary apical tumor; severe shoulder pain; paresthesias, pain, and paresis of the homolateral arm with atrophy of arm and hand muscles.


Pancreatitis

**General:** Inflammation of pancreas.

**Ocular:** Xerosis; night blindness; multiple branch retinal artery occlusions; cotton-wool patches; retinal edema; striate and blot hemorrhages; retinopathy of pancreatitis has been considered to indicate multiple-organ failure and poor prognosis in severe acute pancreatitis; mechanism may be secondary to granulocyte aggregation and leukoembolization due to activated complement.

**Clinical:** Chronic pancreatitis; lipid emboli; malabsorption; vitamin A deficiency.


Papilloma (Wart; Verruca)

**General:** Cutaneous or mucosal tumor of proliferating epithelial and fibrovascular tissues; viral etiology or noninfectious.

**Ocular:** Papillary conjunctivitis; pseudopterygium; corneal opacity; epithelial keratitis; corneal vascularization; lid ulcers; lacrimal system obstruction; hemorrhages of conjunctiva, lids, and lacrimal system.

**Clinical:** Mulberry- or cauliflower-like tumors that may occur on any cutaneous or mucosal surface.

Papillon-Leage-Psaume Syndrome (Oro-Digital-Facial Syndrome; Linguofacial Dysplasia of Grob; Gorlin Syndrome; Dysplasia Linguofacialis; OFD Syndrome; Oro-Digital-Facial Dysostosis; Grob Linguofacial Dysplasia)  

**General:** Familial with strong female preponderance; transmitted as a dominant; partial trisomy has been suggested for the 6-12 (C) chromosome.

**Ocular:** Hypertelorism; displaced medial and lateral canthi; antimongoloid slanting of palpebral fissures; exotropia; see-saw winking.

**Clinical:** Clefts of jaws and tongue due to abnormalities in development of frenulum; syndactyly; polydactyly; alopecia; white, hamartomatous patches of midline of tongue; mental retardation; bradydactyly; hypoplastic nasal cartilages; seborrheic changes; dystopia canthus; pseudocleft of upper lip; alopecia; missing mandibular lateral incisors.


Papillon-Lefevre Syndrome (Hyperkeratosis Palmoplantaris with Periodontosis)  

**General:** Autosomal recessive; onset between ages 1 and 4 years.

**Ocular:** Nystagmus.

**Clinical:** Hyperkeratosis of the palms and soles associated with destruction of the periodontal ligament and premature exfoliation of the teeth; bad breath; loose teeth; loss of teeth.


Pappataci Fever (Phlebotomus Fever; Sandfly Fever)  

**General:** Viral etiology; transmitted by the sandfly Phlebotomus *papatasii*.

**Ocular:** Pick sign of conjunctiva (conjunctival injection limited to the exposed portion of the conjunctiva); uveitis; optic neuritis; papilledema; papillitis; blepharospasm; retinal venous engorgement; vitreal exudates.

**Clinical:** Fever; headaches; myalgia; pain; stiffness of the neck and back.


Paragonimiasis (Distomiasis; Endemic Hemoptysis)  

**General:** Chronic lung infection; causative agent is *Paragonimus trematode*; transmitted by eating undercooked crabs or crayfish and drinking contaminated water; affects all ages; most severe in children.

**Ocular:** Convergence paralysis; optic atrophy; lid cysts; dacroycystitis; papilledema; homonymous hemianopsia.

**Clinical:** Hemoptysis; dyspnea; fever; anorexia; weight loss; pleural effusion; pneumothorax; diarrhea; epileptic seizures; hemiplegia; leukocytosis; eosinophilia; anemia; cystic bronchiectasis; encephalitis; chronic bronchitis; intestinal or peritoneal infections; central nervous system (CNS) paragonimiasis occurs predominantly in persons under age 30 years.


Parinaud Oculoglandular Syndrome (Parinaud Conjunctiva-Adenitis Syndrome; Catscratch Oculoglandular Syndrome; Catscratch Disease; Bartonella henselae) 965

**General:** Most frequently seen in children; incubation time 7 to 10 days; caused by small pleomorphic Gram-negative bacillus; good prognosis; affects both sexes; about 90% of patients with this condition have serologic evidence of infection by Rochalimaea henselae.

**Ocular:** Conjunctivitis; retrotarsal conjunctival granulations; formation of granulomata in anterior segment about 3 mm high and 2 to 6 mm in diameter; inferior fornix usually affected; ulceration common; neuroretinitis; optic neuritis.

**Clinical:** Tender, red papule at the site of a cat scratch; regional preauricular and cervical lymphadenitis (often only one gland involved); irregular fever for 4 to 5 days and malaise; fever; parotid gland swelling.


Parinaud Syndrome (Divergence Paralysis; Subthalamic Syndrome; Paralysis of Vertical Movements; Pretectal Syndrome) 966

**General:** Various causes, including pineal tumor, supranuclear lesions, vascular lesions, inflammation, hemorrhages, midbrain lesions, lesion of posterior white commissure of pons, red nucleus, or superior cerebellar peduncle; combination of Parinaud and von Monakow syndromes is known as Gruner-Bertolotti syndrome, which consists of paralysis in upward gaze, tremors, hemiplegia, and sensory disturbances.

**Ocular:** Retraction of lids with lesion in mesencephalic gray matter and ptosis with lesions more anteriorly; paralysis of conjugate upward movement of the eye without paralysis of convergence; occasionally paralysis of upward and downward movement; spasm with convergence insufficiency; contralateral hemianopsia occurs when the lateral geniculate body becomes involved in case of infiltrating tumor; wide pupils that fail to react to light but sometimes react during accommodation (Holmes); papilledema (usually severe).

**Clinical:** Vertigo; contralateral cerebellar ataxia and choreoathetoid movement if lesion involves superior cerebellar peduncle after decussation.


Parkinson Syndrome (Paralysis Agitans; Shaking Palsy) 967

**General:** Late stages of epidemic encephalitis; present with arteriosclerosis and with manganese and carbon monoxide poisoning; widespread destruction of pigmented cells in substantia nigra.

**Ocular:** Decreased blinking; lid fluttering; blepharospasm; oculogyric crises; ocular hypotony; blepharoplegia; ptosis; nystagmus; paralysis of convergence; paralysis of lateral rectus muscle; absent or sluggish pupillary reactions to light or convergence; mydriasis or anisocoria; optic neuritis; papilledema; abnormal saccades.

**Clinical:** Slowness of movements; loss of facial expression; “cogwheel” rigidity of the arms; rhythmical tremors; drooling; shuffling gait; stooping; monotonous voice.


Parotid Aplasia or Hypoplasia (Salivary Gland Absence; Lacrimal Puncta Absence)

**General:** Autosomal dominant.

**Ocular:** Lacrimal gland aplasia; absence or severe dysfunction of lacrimal glands.

**Clinical:** Aplasia of parotid salivary glands; hemifacial microsomia; mandibulofacial dysostoses; xerostomia; rampant caries; edentulous, salivary gland dysfunction; parotid agenesis or hypoplasia; impalpable parotid gland; absence of the orifice of Stensen duct; bilateral parotid gland aplasia.


Partial Trisomy 16q Syndrome

**General:** Partial trisomy 16q with chromosome banding; rare.

**Ocular:** Narrow palpebral fissures; antimongoloid lid slant; hypertelorism; strabismus; epicanthus; congenital glaucoma; corneal edema; shallow anterior chamber; Rieger anomaly.

**Clinical:** Dry skin; periorbital edema; hydrocele; general hypotonia; low-set ears; micrognathia; hypoplastic lower lip; long philtrum; stumpy hands with short fingers; pectus excavatum.


Passow Syndrome (Bremer Status Dysraphicus; Status Dysraphicus Syndrome; Syringomyelia; Syringobulbia)

**General:** Congenital nonclosure of the neural tube; familial occurrence or may be sporadic; insidious onset in second to third decade of life.

**Ocular:** Enophthalmos; ptosis; rotatory nystagmus; heterochromia iridis; anterior uveitis; corneal anesthesia; neuroparalytic keratitis; paralysis of third, fifth, sixth, and seventh cranial nerves; Horner syndrome; anisocoria; papilledema; optic atrophy; zonular cataract (see Horner Syndrome).

**Clinical:** Anesthesia over area of first division of trigeminal nerve; facial hemiatrophy; facial nerve paralysis; muscular weakness; cervical ribs; kyphoscoliosis; spina bifida; unilateral numbness of fingers; loss of deep reflexes; insensitivity to pain and temperature in affected areas; neurogenic bladder.


Patterned Dystrophy of Retinal Pigment Epithelium

**General:** Autosomal dominant; probably there is a migration of pigment granules in the pigment epithelium, resulting in a specific configuration.
Ocular: Reticular fishnet-like dystrophy; macroticular (spider shaped); butterfly-shaped pigment dystrophy of fovea.

Clinical: None.


Pediculosis and Phthiriasis

General: Infestation of lice on head, body, or pubic area.

Ocular: Conjunctivitis; keratitis; infestation of lice or nits glued to shafts of eyelashes and eyebrow.

Clinical: Pruritus; skin excoriations; impetigo; pyoderma with lymphadenitis and febrile episodes.

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Clinical: Pruritus; skin excoriations; impetigo; pyoderma with lymphadenitis and febrile episodes.


Pelizaeus-Merzbacher Disease (Aplasia Axialis Extracorticalis Congenita; Sudanophilic Leukodystrophy)

General: Rare; subdivision of diffuse cerebral sclerosis predominantly involving the white matter of CNS; onset in infancy or childhood; X-linked recessive; male members affected through normal-appearing carrier mothers; abnormal myelin/myelin sheath structure secondary to abnormal gene on the X chromosome.

Ocular: Lateral, rotatory, or vertical nystagmus or nonrhythmic wandering eye movements; visual impairment from occipital lobe involvement; tapetoretinal degeneration characteristic of retinitis pigmentosa; attenuated arterioles; optic atrophy; papilledema.

Clinical: Retarded development; gait instability, ataxia; intention tremor and athetosis; hearing and speech disturbances; atrophic disturbances; spastic paralysis; weight gain.


Pemphigus Foliaceus (Cazenave Disease)

General: Attacks individuals of any race, age, or sex; high incidence in Brazil; characterized by bullous skin lesions resulting in generalized exfoliation.

Ocular: Exfoliative or bullous lesions of lid and conjunctiva; pannus; infiltration of cornea and iris; cataract.

Clinical: Cutaneous manifestations that progress to scaling, crusted patches; simulates lupus erythematosus and exfoliative erythroderma.

**Pemphigus Vulgaris**

**General:** Primarily in middle-aged people; prognosis varies, from poor to chronic; generalized bullous eruption; blistering autoimmune disease that affects the skin and mucous membranes; association between particular HLA-DR4 and pemphigus vulgaris has been reported.

**Ocular:** Conjunctival bullae; catarrhal conjunctivitis; scarring and adhesions of conjunctiva.

**Clinical:** Cutaneous blisters, which may be clear, pustular, or hemorrhagic.


**Pendred Syndrome (Sporadic Goiter with Deafness)**

**General:** Autosomal recessive; defect in thyroxine biosynthesis.

**Ocular:** Retinal pigmentary degenerative changes; pendular nystagmus; bull's-eye-type macular degeneration.

**Clinical:** Thyroid enlargement; sensorineural hearing loss; mental retardation; thyroid carcinoma.


**Pericentric Syndrome (pericentric Inversion of Chromosome 11)**

**General:** Etiology unknown; leukocyte chromosomes show a pericentric inversion of chromosome 11.

**Ocular:** Strabismus; hypertelorism; congenital glaucoma; aniridia; corneal disease; epicanthic folds.

**Clinical:** Microcephaly; broad nasal bridge; arched palate; hyperextensibility of elbows; left hand shows single transverse crease, and right hand shows three palmar creases radiating from radial border.


**Pericocular Metastatic Tumors (Ocular Metastatic Tumors)**

**General:** Neoplasms that develop from malignant cells and are carried from a primary site of malignancy.

**Ocular:** Retinal detachment; retinal hemorrhages; enophthalmos; exophthalmos; proptosis; rubeosis iridis; uveitis; papilledema; orbital hemorrhages; hyphema; paralysis of extraocular muscles; secondary glaucoma.

**Clinical:** Metastasis in the bloodstream and lymphatic system common; tumors of the lung or breast metastasize to globe; neoplasms that most commonly metastasize to the orbit are neuroblastosomas of suprarenal medulla and retroperitoneal ganglia; Wilms tumor may involve the orbit.


Peroneal Muscular Atrophy (PMA; Neuropathy, Hereditary Sensorimotor, with Upper Motor Neuron, Visual Pathway, and Autonomic Disturbance) 979

**General:** Peroneal muscular atrophy with involvement of other parts of nervous system; autosomal dominant; upper motor neuron and visual pathway lesions.

**Ocular:** Visual pathway lesions; ptosis; irregular pupils; iris atrophy; lack of response to light or near vision.

**Clinical:** Distal weakness and muscle atrophy; absent ankle jerks; foot drop; stocking-type sensory loss; diminished sweating in distal limbs.


Pertussis (Whooping Cough) 980

**General:** Causative agent *Haemophilus pertussis* (*Bordetella pertussis*); not all patients who develop pertussis encephalopathy are children.

**Ocular:** Conjunctivitis; severe cortical blindness; papilloedema; choroiditis; retinal ischemia; ocular muscle palsies; hemorrhages of eyelids, conjunctiva, orbit, anterior chamber, and retina; chronic papilloedema; optic neuritis; retinal and vitreous hemorrhages, and even intracranial and subarachnoid hemorrhage associated with increased intrathoracic and intraabdominal pressures during coughing.

**Clinical:** Respiratory tract infection; nasal discharge; cough ending with a loud crowing; inspiratory noise (the "whoop"); thick mucoid sputum; soreness over trachea; ulcer of glottis; vomiting; tetany; encephalopathy; cortical blindness.


Peters Anomaly 981

**General:** Autosomal recessive; may be morphologic entity with several eye syndromes, including Rieger syndrome, Mietens syndrome, and fetal alcohol syndrome; may be due to a developmental field defect, a contiguous gene syndrome, or a defective homeotic gene controlling development of the eye and other body structures.

**Ocular:** Corneal opacification; lenticulocorneal adherence; iris adhesions; glaucoma; cataract; narrow lid fissures; colobomatous microphthalmia; persistent hyperplastic primary vitreous; retinal detachment; iris nodules.

**Clinical:** Short-limbed dwarfism; broad face; thin upper lip; hypoplastic columella; hypospadias; cleft lip and palate; craniofacial abnormalities; congenital heart disease; horseshoe kidney; polycystic kidneys; Wilms tumor; mental retardation; external ear anomalies; camptodactyly.


Petzetakis-Takos Syndrome (Phlyctenular Keratoconjunctivitis) 982

**General:** Malnutrition; lack of hygiene.

**Ocular:** Superficial keratitis; palpebral edema; cornea hyperesthesia; photophobia; blepharospasm; decreased pupillary response; xerophthalmia.

**Clinical:** Lymph node hypertrophy.
### Peutz-Touraine Syndrome (Peutz-Jeghers Syndrome)

**General:** Recognized in infants; autosomal dominant; gastrointestinal polyps; jejunal polyps are consistent feature; these lesions are benign, melanin spots of the lips; buccal mucosa and digits represent second part of syndrome.

**Ocular:** Brownish speckled dots along border of skin and mucosa of eyelids; freckles similar to those seen on eyelids may appear on conjunctiva, primarily adjacent to limbus and along area of lid fissure; pigment speckles of sclera and iris; brown-pigmented corneal spot.

**Clinical:** Brownish to black dotlike pigmentation similar to that seen on eyelids is present in the perinasal, perioral, and periorbital regions, fewer on fingers and toes at birth; polyposis of gastrointestinal tract (primarily small intestine) with associated bleeding; potential for malignant transformation in second decade of life.


Reid JD. Intestinal carcinoma in the Peutz-Jeghers syndrome. *JAMA* 1974; 229:833.


### Pfeiffer Syndrome

**General:** Congenital craniosynostotic syndrome with a high rate of mortality shortly after birth.

**Ocular:** Severe proptosis (also described as extreme exophthalmic midface hyperplasia).

**Clinical:** Cloverleaf skull, elbow ankylosis, broad thumbs, and/or broad halluces; variable additional abnormalities including pulmonary problems, brain abnormalities, and prematurity that frequently lead to early death.


### Photosensitivity and Sunburn

**General:** Enhanced responsiveness to natural or artificial nonionizing electromagnetic radiation; photosensitivity induced by exogenous agents accounts for an increasing portion of the total undesirable effects caused by environmental chemicals.

**Ocular:** Photo-keratoconjunctivitis; conjunctivitis; erythema of periorbital skin; photophobia; lacrimation; blepharospasm.

**Clinical:** Erythema of skin; edema; vesiculation of skin; fever; nausea; chills; delirium; irregular pigmentation of skin.


### Pierre-Robin Syndrome (Robin Syndrome; Micrognathia-Glossoptosis Syndrome)

**General:** Etiology unknown; manifestations at birth; pathogenesis based on arrested fetal development; history of intrauterine disturbance in early pregnancy (25% of cases); also increased incidence in offspring of mothers age 35 years or older; pathogenesis is thought to be incomplete development of the first brachial arch, which forms the maxilla and mandible.

**Ocular:** Microphthalmos; proptosis; ptosis; high myopia; glaucoma; cataract (rare); retinal disinsertion; megalocornea; iris atrophy; blue sclera; esotropia; conjunctivitis; distichiasis; vitreoretinal degeneration; retinal detachments.
Clinical: Micrognathia; cleft palate; glossoptosis; cyanosis; facial expression birdlike with flat base of nose and high-arched deformed palate with or without cleft; difficulty breathing.


**Pigmentary Ocular Dispersion Syndrome (Pigmentary Glaucoma)**

**General:** Polygenic inheritance; onset at average age 52 years; distribution of pigment in chamber angle; atrophy of posterior iris epithelium; most commonly affects young Caucasian males with myopia; mechanism is likely to be mechanical rubbing between iris pigment epithelium and packets of lens zonules, resulting in aqueous flow obstruction secondary to accumulation of pigment granules in the trabecular meshwork.

**Ocular:** Myopia; glaucomatous field changes; ocular hypertension; iris translucency; abnormal number of iris processes; insertion of iris anterior to scleral spur; pigmenta\tion of posterior trabecular meshwork, grades 3 to 4; Krukenberg spindles; presence of pigmentation on equatorial border of lens capsule; glaucomatous cupping and myopic optic nerve changes.


**Pillay Syndrome (Ophthalmomandibulomelic Dysplasia)**

**General:** Autosomal dominant; both sexes affected.

**Ocular:** Cornal opacities.

**Clinical:** Temporomandibular fusion; obtuse mandibular angle; short forearms.


**Pilodental Dysplasia with Refractive Errors (Euhidrotic Ectodermal Dysplasia; Trichodental Dysplasia with Hyperopia)**

**General:** Autosomal recessive; damage to structures arising from the ectoderm.

**Ocular:** Hyperopia; astigmatism.

**Clinical:** Hypodontia; abnormally shaped teeth; scalp hypotrichosis; disorders of pigmentation of the hair; follicular hyperkeratosis on the trunk and limbs; intensified delineation and reticular hyperpigmentation of the skin of the nape; broadening of bridge and dorsum of the nose.


**Pinta (Nonvenereal Treponematosis)**

**General:** Caused by spirochete *Treponema carateum*; infectious; contagious; found in Mexico, Central America, West Indies, and the northern countries of South America; caused by an organism that is morphologically and antigenically identical to the causative agent of venereal syphilis.

**Ocular:** Hypopigmentation of eyelid.

**Clinical:** Cutaneous lesions with marked pigmentary changes; chronic relapsing course.


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**PISK (Pressure Induced Intralamellar Stromal Keratitis)**

**General:** Associated with LASIK post operative patients

**Ocular:** Elevated IOP; ocular discomfort; blurred vision; stromal keratitis


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**Pituitary Dysfunction and Chorioretinopathy (CPD Syndrome; Chorioretinopathy and Pituitary Dysfunction)**

**General:** Race, characterized by chorioretinopathy, trichosis, and pituitary dysfunction.

**Ocular:** Long lashes; bushy eyebrows; severe early-onset chorioretinopathy.

**Clinical:** Growth retardation; sexual infantilism; hypothyroidism; mental retardation; low birth weight; cerebellar ataxia.


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**Pituitary Gigantism Syndrome (Gigantism Syndrome; Launois Syndrome)**

**General:** Increased production of growth hormone due to hyperplasia of the eosinophilic cells and chromophobe adenoma of the anterior pituitary gland; onset age 8 to 10 years; pituitary adenomas represent 10% to 15% of intracranial neoplasms; 40% of nonfunctioning tumors are prolactinomas; growth hormone-secreting tumors are the next most common type.

**Ocular:** Field defects according to extent and situation of the pituitary adenoma; optic nerve atrophy (partial).

**Clinical:** Gigantism; enlarged skull with prominent chin and forehead; retarded skeletal growth; delayed puberty; muscle weakness; headache; perspiration; joint pain; mental retardation; pallor; smooth skin; scanty facial and body hair; small penis and testes; high-pitched voice; large limbs, hands, and feet.


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**Pituitary Necrosis Syndrome (Postpartum Hypopituitarism Syndrome; Sheehan Syndrome; Simmonds-Sheehan Syndrome)**

**General:** Etiology unknown; vascular occlusion of one of the vessels supplying the anterior lobe of the pituitary during childbirth; characterized by various degrees of anterior and/or posterior pituitary dysfunction due to pituitary necrosis after obstetric shock or hemorrhage.
Ocular: Hypotrichosis of eyebrows; loss of eyelashes; dry and scaly skin of the lids; visual loss due to vascular insufficiency; uveal depigmentation.

Clinical: Reduced sweating with dry skin; listlessness and lethargy; stupor; myxedema; premature aging; cutaneous hypopigmentation; reduced and sparse axillary and pubic hair; genital atrophy; menstrual irregularity; amenorrhea; thyroiditis.


**Pityriasis Rubra Pilaris (Kaposi Disease [2]; Devergie Disease; Hebra Disease; Tarral-Besnier Disease; Lichen Ruber; Lichen Ruber Acuminatus; Pityriasis Pilaris)** 995

**General:** Abnormal keratinization of unknown etiology; hereditary and acquired forms have been described in the literature; hereditary form tends to be less severe and more limited in extent.

**Ocular:** Papules on bulbar conjunctiva; keratitis; ectropion; pannus; corneal ulceration.

**Clinical:** Cutaneous manifestations; erythema; follicular papules.


**Plague (Bubonic Plague; Pneumonic Plague)** 996

**General:** Infectious disease of animals (principally wild animals and rodents) that is transmitted through the bite of infected ectoparasites; causative agent is *Yersinia pestis*.

**Ocular:** Inflammatory infiltrate of lids; subconjunctival hemorrhages; chemosis; staphyloma of sclera; keratitis; corneal abscess; iridocyclitis; chorioiditis.

**Clinical:** Abrupt onset; chills; fever; rapid, thready pulse; painful, enlarged lymph nodes in bubonic plague; headache and productive cough in pneumonic plague; meningitis; occasional patients infected with *Y. pestis* become septic and die with bacteremia without developing any detectable lymphadenitis (septicemic plague).


**Plasma Lecithin Deficiency (Cholesterol Acyltransferase Deficiency)** 997

**General:** Autosomal recessive.

**Ocular:** Corneal stromal opacities comprising small gray dots in central and peripheral areas; retinal hemorrhages; disk protrusion; dilated veins.

**Clinical:** Storage of lipid materials in various tissues.


**Plateau Iris Syndrome** 998

**General:** Rare; occurs in younger age group; presumably due in part to an anterior insertion of the iris; pupillary block is not a significant part of the mechanism leading to angle closure.
Ocular: Spontaneous or mydriasis-induced angle closure despite a patent iridectomy; anterior chamber is of normal depth axially and the iris plane is flat, but a peripheral roll of iris can close the angle either when the pupil dilates spontaneously or after mydriatic drugs are administered.

Clinical: Nausea; vomiting.


Plummer-Vinson Syndrome (Sideropenic Dysphagia Syndrome; Paterson-Brown-Kelly Syndrome; Waldenstrom-Kjellberg Syndrome)

General: Deficiency of vitamin B complex and iron; female; onset middle age; pathogenic mechanism may be related to a frameshift mutation in the human apolipoprotein A-I gene.

Ocular: Reduced tear formation; pale conjunctiva; dry eyes; retinal hemorrhages; papilledema.

Clinical: Dysphagia for solid food with main difficulties originating in the upper portion of the esophagus; glossitis and gastritis; anemia; atrophy of mucous membranes; dystrophy of the fingernails (koilonychia); fatigue.


Pneumococcal Infections (Streptococcus Pneumoniae Infections)

General: Gram-positive diplococcus Streptococcus pneumoniae; some strains are encapsulated while others are not; ocular infections usually are caused by the encapsulated strains; conjunctivitis and corneal scarring produced in an animal model have been attributed to a hemolytic cytolytic exopeptidase.

Ocular: Hypopyon; conjunctivitis; keratitis; corneal ulcer; endophthalmitis; dacryocystitis; uveitis; orbital cellulitis; secondary glaucoma; ophthalmia neonatorum.

Clinical: Upper respiratory infection; chills; sharp pain in hemithorax; cough with sputum production; fever; headache; gastrointestinal symptoms.


Poison Ivy Dermatitis (Rhus Dermatitis; Poison Oak Dermatitis; Poison Sumac Dermatitis)

General: Direct contact or airborne contact from burning of plant.

Ocular: Keratitis; chemosis; blepharospasm; pustules of eyelids.

Clinical: Dermatitis; linear lesions; erythema; pruritus.

Poland-Möbius Syndrome

**General:** Rare congenital disorder that is a combination of Möbius syndrome and Poland anomaly; has been proposed that Möbius syndrome, the Poland anomaly, and the Klippel-Feil defect all result from a transient interruption during the sixth week of gestation in the development of the subclavian artery and its branches.

**Ocular:** Bilateral sixth nerve paralysis; no movement of upper lid in horizontal gaze; no globe retraction during adduction; vertical nystagmus; diplopia; chronic keratitis; corneal ulcer.

**Clinical:** Paresis of the sixth and seventh cranial nerves; bilateral absence of pectoralis muscle and ipsilateral hand and digit anomalies; chronic drooling; speech difficulties; masklike facial expression.


Poliomyelitis (Infantile Paralysis)

**General:** Acute viral infection characterized by varying degrees of neuronal injury, with special localization in the anterior horns and motor nuclei of the brainstem.

**Ocular:** Diplopia; nystagmus; paralysis of third, fourth, and sixth nerves; paresis of seventh nerve; papilledema; visual agnosia; Homer syndrome; pupillary paralysis; optic neuritis; ophthalmoparesis; transient visual loss; internuclear ophthalmoplegia; papillary disturbances, spasm of near reflex.

**Clinical:** Flaccid paralysis of many muscle groups; death from asphyxia and involvement of vital centers in the brainstem.


Polycythemia Vera (Erythema; Erythrocytosis Megalosplenica; Myelopathic Polycythemia; Vaquez Disease; Vaquez-Osler Syndrome; Cryptogenic Polycythemia; Polycythemia Rubra; Splenomegalic Polycythemia)

**General:** Increased number of red blood cells; myeloproliferative disorder.

**Ocular:** Conjunctival vascular engorgement; dilated tortuous retinal veins; retinal hemorrhages; optic disk edema; central retinal vein occlusion; visual field defects; visual hallucinations; diplopia.

**Clinical:** Elevated red blood cells; systemic vascular congestion; leukocytosis; thrombocytosis; central nervous system involvement; splenomegaly; hepatomegaly; bleeding diathesis; gingival/mucosal bleeding; ecchymosis; epistaxis; neurologic abnormal dizziness; vertigo; ataxia.


Polymyalgia Rheumatica

**General:** Affects older patients; usually Caucasian women; relationship between polymyalgia and temporal arteritis remains uncertain; in polymyalgia rheumatic a alone there is no arteritis per se, and this syndrome is not associated with blindness or other neurologic or cardiovascular sequela; many patients with temporal arteritis have polymyalgia rheumatic a as part of their symptoms or as the presenting symptom.

**Ocular:** Amaurosis fugax; acute unilateral or bilateral visual loss; ischemic optic neuritis; optic atrophy; papilledema; retinal hemorrhages; cotton-wool spots; central retinal artery occlusion; palsy of extraocular muscles.
Clinical: Pain and stiffness in the neck, shoulders, and hips; rapid erythrocyte sedimentation rate; prompt response to corticosteroids; weakness.


### Polymyositis-Dermatomyositis (Dermatomucomyositis; Neuromyositis; Polymyositis Gregarina; Wagner-Unverricht Syndrome)

**General:** Autoimmune disease; etiology unknown; variable symptoms according to prevalence of skin (dermatomyositis) or muscular (polymyositis) involvement; association with neoplastic disease; prevalent in females; onset in childhood before age 10 years and in adults predominant in fourth to sixth decades; both children and adults are affected by this disease, but the prognosis is better for the childhood forms of disease.

**Ocular:** Violent discoloration of eyelids; conjunctivitis; episcleritis; anterior uveitis; nystagmus; exophthalmos; cotton-wool spots; retinal edema; retinal hemorrhage; venous engorgement.

**Clinical:** Erythema involving face, forearms, and upper back; muscle weakness, especially shoulder and pelvic girdles; dysphagia; respiratory difficulty; malaise; fever; tachycardia.


### Pompe Disease (Generalized Glycogenosis)

**General:** Absence of acid maltase; type II glycogen storage disease with decreased acid maltase resulting in elevated levels of lysosomal glycogen; inheritance is autosomal recessive.

**Ocular:** Cortical blindness.

**Clinical:** Anorexia; retardation of growth; cyanosis; dyspnea; convulsions; death; enlarged tongue; large heart; hypotonicity.


### Popliteal Pterygium Syndrome (PPS)

**General:** Rare autosomal dominant disorder similar to Van der Woude syndrome (the two are possibly allelic).

**Ocular:** None.

**Clinical:** Popliteal webbing; cleft lip; cleft palate, lower lip pits; syndactyly; genital and nail abnormalities; severe flexure contract at the knee; equinus foot; velar pterygium.

Porokeratosis

**General:** Autosomal dominant; prominent in males; onset in early childhood; characterized by scaly papules that enlarge to form gyrate lesions.

**Ocular:** Conjunctivitis; lesions of cornea.

**Clinical:** Squamous cell carcinoma within lesions; lesions may occur on oral mucosa, palms, or soles.


Porphyria Cutanea Tarda

**General:** Disorder of porphyrin metabolism; highest incidence in Bantu population; both sexes affected; onset between ages 40 and 60 years; insidious onset; autosomal dominant; light-sensitive dermatitis in later adult life; associated with excretion of large amounts of uroporphyrin in urine.

**Ocular:** Synophrys; keratitis; palsies of third and seventh cranial nerves; scleromalacia perforans; optic atrophy; retinal hemorrhages and cotton-wool spots; macular edema; pterygium; brownish pigmentation in conjunctiva and lid margin.

**Clinical:** Cutaneous manifestations are solar hypersensitivity, vesiculobullous lesions, ulcerations, severe scarring, and hypertrichosis; erythrodermia.


Posner-Schlossman Syndrome (Glaucatomaticyclitic Crisis)

**General:** High intraocular tension lasting from hours to several weeks and recurring at varying frequencies; low-grade, intermittent, nongranulomatous inflammation; in one series of patients, HLA-BW54 was present in 41% of patients.

**Ocular:** Slight blurring of vision and colored halos during episodes of high intraocular tension; high intraocular pressure (unilateral); glaucomatocyclitic crisis (benign and usually unilateral); enlarged pupil; anisocoria; absence of ciliary or conjunctival injection; only trace of aqueous flare; no posterior synechiae; chamber angle open; heterochromia iridis; keratic precipitates may be present.

**Clinical:** Allergy; associated with gastrointestinal disease (peptic ulcers).


Posterior Iris Chafing Syndrome

**General:** Complication following intraocular lens implantation attributed to sulcus-fixed posterior chamber lenses, decentration of the lens, traumatic insertion, movement of the lens during dilation and constriction of the pupil, or difficulty with positioning of the lens.
Ocular: Iris transillumination defects; recurrent microhyphemas; pigment dispersion glaucoma; pigment deposition in trabecular meshwork; iris pigment atrophy.

Clinical: None.


### Posterior Uveal Bleeding Syndrome

**General:** Most commonly affects middle-aged black females; also observed in white or Asian people

**Ocular:** Multiple recurrent pigment epithelial detachments; posterior uveal bleeding; polyps in the peripapillary macula and extramacular areas

**Clinical:** Hypertension; detected with indocyanine green angiograph


### Posthypoxic Encephalopathy Syndrome (Posthypoxic Syndrome; Parietooccipital Syndrome)

**General:** Hypoxia secondary to carbon monoxide poisoning, high altitude, complication during anesthesia, hypoglycemia, or cardiac failure, causing widespread demyelination in the parietal lobes, including the optic radiations.

**Ocular:** Nystagmus; nuclear ophthalmoplegia; visual hallucinations; partial cerebral blindness (predominant defects in the sphere of psychic elaboration rather than in primary visual perception); complete cortical blindness; central scotomata; pupillary paresis; retinal atrophy; optic atrophy.

**Clinical:** Confusion; irritability and agitation; alexia; disorientation (mainly spatial); muscle spasm.


Hoyt WF, Walsh FB. Cortical blindness with partial recovery following acute cerebral anoxia from cardiac arrest. *Arch Ophthalmol* 1958;60:1061.


### Poststreptococcal Uveitis Syndrome

**General:** Associated with bilateral nongranulomatous ocular inflammation; seen in individuals over 40 years of age.

**Ocular:** Vitritis; focal retinitis; optic disk swelling; multifocal choroiditis


### Postvaccinial Ocular Syndrome

**General:** Immediate contamination and inoculation or as a delayed reaction; incubation time 3 days.

**Ocular:** Orbital cellulitis; pustules and vesicles on the eyelids (vesicles appear grayish with purulent discharge); lids swollen and red with swelling of preauricular and postauricular lymph nodes; ophthalmoplegia; mild anterior uveitis; conjunctival and corneal vesicles similar to the lid lesions; corneal marginal ulcers (eventually may lead to corneal perforation and loss of the eye); chorioretinitis (occasionally); central serous retinopathy; central retinal vein thrombosis; panophthalmitis; neuritis; ocular vaccinia may mimic signs of herpes simplex virus, varicella-zoster virus, and acanthamoeba keratitis.

**Clinical:** Postvaccinal encephalitis; severe local purulent skin reaction; severe headache; fever; malaise.


Potter Syndrome (Renal Agenesis Syndrome; Renofacial Syndrome)

**General:** Unknown etiology; may be severe form of the trisomy 18 syndrome; results from prolonged oligohydramnios of any cause.

**Ocular:** Hypertelorism; pronounced epicanthal folds extending down the cheeks; antimongoloid slant of palpebral fissure.

**Clinical:** Flat bridge of the nose; low-set ears; facial deformities; micrognathia; pulmonary hypoplasia; cystic dysplasia of kidney to agenesis; oligohydramnios; clubbing of hands and feet; spina bifida; prominent infracanthal folds; flattened beaked nose; creased skin; positional deformities of the limbs.


**General:** Etiology unknown; dominant inheritance is suspected; predominantly seen in males; Royer syndrome is Prader-Willi syndrome associated with diabetes mellitus; non genetic condition characterized by infantile hypotonia, hypogonadism, and obesity.

**Ocular:** Strabismus; ocular hypertelorism; myopia; exotropia; glaucoma; cataracts; congenital ocular fibrosis syndrome; diabetic retinopathy.

**Clinical:** Mental retardation; short stature; muscular hypotonia; small hands and feet; obesity; cryptorchidism; hypogonadism; dental caries; clinoactyty; partial syndactyly of toes and fingers.


Pregnancy

**General:** Pregnancy results in hormonal changes that produce ocular effects; symptoms resolve at end of pregnancy term.

**Ocular:** Myopia; visual field defects; corneal edema; acute ischemic optic neuropathy; central serous retinopathy; glaucoma; ptosis; diabetic retinopathy; Krukenberg spindles; transient blindness; serous retinal detachment; retinal artery occlusion; retinal vein occlusion; disseminated intravascular coagulopathy; uveal melanoma.

**Clinical:** Nausea; headaches; hypertension; benign intracranial hypertension; preeclampsia; toxemia; fluid retention.


Primary Antiphospholipid Syndrome (PAS)

**General:** Thrombophilic disorder characterized by the presence of autoantibodies and CNS involvement, venous thrombosis, and ocular manifestations.

**Ocular:** Visual disturbances (visual obscurations, amaurosis fugax, diplopia, homonymous field defects, and scintillating scotoma due to migraine); two forms of retinopathy: cotton-wool spots with or without hemorrhages and vasoocclusive ocular disease.

**Clinical:** Many patients will experience CNS involvement in the form of either a stroke or a transient ischemic attack; visual disturbances are largely due to CNS rather than ocular ischemia.


Progressive External Ophthalmoplegia and Scoliosis

**General:** Rare; isolated muscle dystrophic involvement of extraocular muscles; onset in childhood or early adulthood; slowly progressive.

**Ocular:** Horizontal gaze paralysis; pendular nystagmus; ptosis; orbicularis oculi weakness.

**Clinical:** Scoliosis; facial myokymia; contracture of facial muscles.


Anderl H. Free vascularized groin fat flap m hypoplasia and hemiatrophy of the face (a three year observation). J Maxillofac Surg 1979; 7:327-332.


Progressive Foveal Dystrophy (Central Retinal Pigment Epithelial Dystrophy)

**General:** Autosomal dominant; onset late in the first decade of life.

**Ocular:** Progressive foveal dystrophy; pigmentary changes and drusen of the macula; normal electroretinogram; subnormal electrooculogram.

**Clinical:** Generalized aminoaciduria; increased glycine levels.


Progressive Inherited Retinal Arteriolar Tortuosity with Retinal Hemorrhages (Familial Retinal Arteriolar Tortuosity with Retinal Hemorrhages)

**General:** Autosomal dominant; tortuosity increases most dramatically during adolescence and progresses throughout adulthood.

**Ocular:** Tortuosity of the retinal arterioles; spontaneous retinal hemorrhages.

**Clinical:** None.


### Progressive Intracranial Arterial Occlusion Syndrome (Taveras Syndrome) 1024

**General:** Children and young adults; caused by previous trauma and atheroma; occlusive endarteritis; predilection for the circle of Willis; occurs mostly in Japanese and Caucasians.

**Ocular:** Unilateral ptosis; defective optokinetic nystagmus; visual agnosia; amaurosis fugax; abnormally high ophthalmodynamometric readings may be the first correct diagnostic sign even before arteriography reveals the vascular pathologic condition.

**Clinical:** Progressive intracranial arterial occlusion with both internal carotid arteries involved; memory loss; muteness; localized numbness; crying spells; catatonic states and episodes, staring; seizures.


### Progressive Systemic Sclerosis (Scleroderma; Systemic Scleroderma) 1025

**General:** Chronic connective tissue disease of unknown etiology; chronic and usually progressive disorder; typical onset is in third to fifth decade; ratio of women to men is 4:1; primary sites of pathology are the arterioles and capillaries of affected organs.

**Ocular:** Marginal corneal ulcers; shortened fornices of the conjunctiva; ptosis; cotton-wool patches of retina; papilledema; retinal hemorrhages; cicatrization of conjunctiva and cornea; blepharitis; blepharospasm; thready, tenacious yellow-white conjunctival discharge; hypertrophy of lacrimal gland; episcleritis; ocular myositis; Sjögren syndrome; uveitis; vitreal haze; keratitis sicca; decreased corneal sensation; iritis; ischemic choroidopathy; iris sectorial atrophy; blepharophimosis; heterochromia; keratoconus; central retinal vein occlusion; branch retinal vein occlusion.

**Clinical:** Vascular insufficiency; Raynaud phenomenon; malaise; weight loss; stiffness; fever; polyarticular arthritis; diffuse edema of the hands; calcinosis; esophageal involvement; sclerodactyly; telangiectasia; esophageal stricture; renal failure; diffuse interstitial fibrosis.


### Protein C Deficiency 1026

**General:** Autosomal recessive; congenital; heterozygous individuals predisposed to recurrent venous thrombosis; homozygous individuals may develop widespread thrombotic complications in neonatal period; reduced clotting ability; neonatal protein C deficiency also may be acquired and transient in ill preterm babies.

**Ocular:** Vitreous opacities; vitreous hemorrhages; retinal detachment; cataract; shallow anterior chamber; leukocoria; prominent iris vessels; iris synechiae to lens; bilateral involvement; retinal hemorrhages; cavernous sinus thrombosis.

**Clinical:** Hematomas; epistaxis; prolonged bleeding; hydrocephalus; *Candida* sepsis; subarachnoid hemorrhage; pulmonary embolism; thrombotic hemorrhagic gastrointestinal and genitourinary mucosal infarcts.


### Proteus Infections 1027

**General:** Gram-negative bacilli found in water, soil, and decaying organic substances.

**Ocular:** Conjunctivitis; keratitis; corneal ulcers; endophthalmitis; panophthalmitis; dacryocystitis; gangrene of eyelid; uveitis; hypopyon; paralysis of seventh nerve.
**Clinical:** Cutaneous infection after surgery; usually occurs as a secondary infection of the skin, ears, mastoid sinuses, eyes, peritoneal cavity, bone, urinary tract, meninges, lung, or bloodstream; meningitis; intracranial subdural and epidural empyema; brain abscess; intracranial septic thrombophlebitis affecting cavernous/lateral sinuses.


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**Proteus Syndrome**

**General:** A harmartoe neoplastic disorder with variable clinical manifestations

**Ocular:** Myopia; band keratopathy; cataract; vitreous hemorrhage; chorioretinal mass; serous retinal detachment

**Clinical:** Thickening of the bones of the external auditory meatus and cranial fossa; enlargement of the left internal auditory meatus; deformities of the feet and toes.


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**Proximal and Distal Click Syndrome of the Superior Oblique Tendon (Simulated Superior Oblique Tendon Syndrome)**

**General:** Produced by quick head movements; caused by adhesions (secondary to trauma and inflammation) or frontal sinus surgery; proximal click adhesions in front to trochlea; distal click adhesions behind trochlea; associated with Brown syndrome.

**Ocular:** Decreased elevation in adduction; downshoot of the affected eye on adduction; overaction of the tethered inferior oblique after cutting superior oblique tendon; widening of palpebral fissure on adduction; diplopia.

**Clinical:** Rheumatoid arthritis.


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**Pruritus**

**General:** Unpleasant sensation perceived in the skin that elicits the response of scratching; causes may be physiologic or pathologic.

**Ocular:** Vesicles, urticaria, and eczematization of lid.

**Clinical:** Urticaria; hives; macular erythema; vesicles; eczematization of lid; skin excoriations; secondary infection; lichenification.


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**Pseudoexfoliation Syndrome**

**General:** Prevalent over age 70 years; rare before age 40 years; unilateral involvement in 40% to 50% of cases; asymmetry of severity in bilateral cases; most common in Caucasians, especially from Iceland and Scandinavian countries; pseudoexfoliation fibers were identified in autopsy tissue specimens of skin, heart, lungs, liver, and cerebral meninges; consistently associated with connective tissue components, i.e., fibroblasts, collagen and elastic fibers, myocardial tissue and heart muscle cell.

**Ocular:** Gray or white fluffy material deposited in particles, flakes, or sheets on anterior surface of iris, ciliary body, posterior surface of cornea, pupillary margin, lens, and trabecular meshwork; increased pigmentation of trabecular meshwork; zonular dialysis; displaced or dislocated lens; anterior chamber depth asymmetry; preoperative phacodonesis; glaucoma; cataract.

**Clinical:** None.

**Pseudo-Foster Kennedy Syndrome (1032)**

**General:** Mimic disease of Foster Kennedy syndrome consisting of ischemic optic neuropathy and glioma.

**Ocular:** Optic disk pallor; visual field defects.

**Clinical:** Slow, progressive, decreased vision.


**Pseudo-Graefe Syndrome (Fuchs Sign) (1033)**

**General:** Misdirection of regenerating oculomotor nerve (cranial nerve III) fibers to other muscles after injury, aneurysm, tumor, exophthalmic goiter, tabes, anterior poliomyelitis, or vascular lesions of the brainstem.

**Ocular:** Elevation of the upper lid in downward gaze; lagging in upper lid movement on downward gaze (Graefe sign).

**Clinical:** None.

Bender MB. The nerve supply to the orbicularis muscle and the physiology of movements of the upper lid, with particular reference to the pseudo-Graefe phenomenon. Arch Ophthalmol 1936; 15:21.

**Seabright-Bantam Syndrome; Albright Hereditary Osteodystrophy (1034)**

**General:** Etiology unknown; autosomal dominant; more common in females (2:1); present from birth; kidney and skeleton fail to respond to parahormone; if patients receive parathyroid extract, their kidneys fail to respond with phosphate diuresis; genetic form of hypoparathyroidism resulting from end-organ resistance to parathyroid hormone; resulting hypocalcemia is responsible for many of the clinical features of this syndrome.

**Ocular:** Strabismus; blue sclera; punctate cataracts (white opacities and polychromatic cortex); papilledema; hypertelorism; keratitis; scleral and choroidal calcifications; blepharospasm; cataracts.

**Clinical:** Short stature; short metacarpals; short limbs; round face with short neck; decalcification of teeth; obesity; fat, stubby hands; tetany with positive Chvostek and Trousseau signs; atypical seizure disorder.


**Pseudo malignant Glaucoma Syndrome (1035)**

**General:** Related to obstruction of aqueous flow either by residual anterior hyaloid or by fibrin and other inflammatory debris at the level of the ciliary body/zonular apparatus following vitrectomy.

**Ocular:** Forward movement of lens-iris diaphragm; elevation of intraocular pressure; axial shallowing of anterior chamber; hydration of vitreous cavity.

**Clinical:** None.

Pseudomonas Aeruginosa Infections

**General:** Gram-negative rod with secondary contaminant of superficial wounds; *Pseudomonas* organisms produce a variety of enzymes that cause pathologic changes, including hemolysins and exotoxins as well as a glycocalyx that increases adherence.

**Ocular:** Hypopyon; conjunctivitis; keratitis; ulcerative abscess of cornea; endophthalmitis; panophthalmitis.

**Clinical:** Local tissue damage and diminished host resistance, which may occur in ear, lung, skin, and urinary tract.


Pseudopseudophthalmoplegia Syndrome (Roth-Bielschowsky Syndrome)

**General:** Supranuclear lesion in the temporal lobe.

**Ocular:** Paralysis of lateral gaze in one direction; vestibular nystagmus in which the fast phase is absent on the ipsilateral side but the slow phase is present.

**Clinical:** Basal ganglia or tectum lesion.


Pseudopapilledema (Optic Nerve Head Drusen)

**General:** Autosomal dominant; incidence in males and females is approximately the same; two thirds of cases are bilateral; visual acuity usually unaffected; may cause slowly progressive visual field defect.

**Ocular:** Elevation of optic disk; drusen; injected conjunctiva; associated with retinitis pigmentosa, subretinal, subretinal pigment epithelium hemorrhages (rare).

**Clinical:** None.


Pseudophakic Pigment Dispersion Syndrome

**General:** Caused by rubbing of peripheral iris on lens zonules with iris plane and posterior chamber lenses.

**Ocular:** Transillumination of iris defects; pigment granules in aqueous humor; pigment dusting of anterior iris surface; band of pigment in filtration portion of trabecular meshwork; glaucoma.

**Clinical:** None.


Pseudoprogeria Syndrome

**General:** Rare; autosomal recessive; absent eyebrows and eyelashes with mental retardation.

**Ocular:** Glaucoma; absence of eyelashes and eyebrows.

**Clinical:** Progressive spastic quadriplegia; microcephaly; small, beaked nose; cervical spinal cyst; occipital cranium bifidum occultum; mental retardation.

Pseudo-Pseudo-Foster Kennedy Syndrome

**General:** Mimic disease of Pseudo-Foster Kennedy Syndrome consisting of ischemic optic neuropathy and meningioma.

**Ocular:** Optic disk pallor, visual field defects.

**Clinical:** Slow, progressive, decreased view.


Psittacosis (Ornithosis)

**General:** Infectious disease transmitted from birds to man; causative agent is *Chlamydia psittaci*; poultry, pigeons, and parrots prominent carriers.

**Ocular:** Lid edema; corneal ulcers; scleritis; ophthalmoplegia; uveitis; acute focal retinitis; stellate retinopathy; cataract.

**Clinical:** Fever, malaise; headache; wheezing; intracranial hypertension.


Psoriasis (Psoriasis Vulgaris)

**General:** Chronic skin disease of unknown etiology; both sexes affected; onset at any age; disease peaks at puberty; strong human leukocyte antigen (HLA) association resulting in heritable disease susceptibility.

**Ocular:** Desquamative psoriatic plaques of lids resulting in madarosis, trichiasis, or ectropion; corneal plaques; xerosis, symblepharon; keratitis; chronic corneal ulceration; phthisis bulbi; iritis.

**Clinical:** Thick, dry, elevated red patches of skin covered with coarse silvery scales that usually affect areas of skin not exposed to sun, such as scalp, sacrum, elbows, and knees; positive association with Sjögren syndrome and keratitis sicca.


Psoriatic Arthritis

**General:** Chronic skin disease of unknown etiology; both sexes affected; onset at any age; disease peaks at puberty.

**Ocular:** Conjunctivitis; iritis; keratitis; uveitis.

**Clinical:** Rash; spondylitis; inflammatory bowel disease; diarrhea; degenerative disease of spine.


Pterygium of Conjunctiva and Cornea

**General:** Autosomal dominant; more frequent in people who work outdoors; occurs late in life.

**Ocular:** Wing-shaped thickening in the conjunctiva, usually nasal, in the interpalpebral fissure area; alterations in corneal topography can cause a reduction in visual acuity.

**Clinical:** None.

Puckering Syndrome 1046

**General:** Disturbance of vitreous with vitreous retraction.

**Ocular:** Epiretinal membrane formation following vitreous bleed, total retinal detachment, detachment of the macula, multiple retinal operations, multiple perforations, loss of formed vitreous at operation, or posterior vitreous separation from trauma to the eye or whiplash injury.

**Clinical:** Neck injury with whiplash.


Pugh Syndrome 1047

**General:** May be misdiagnosed as UGH (uveitis, glaucoma, hyphemas) syndrome; unrelated to the presence of an intraocular lens.

**Ocular:** Pseudouveitis; glaucoma; hyphemas as in UGH syndrome, but also neovascular membrane covering the iris; central retinal vein occlusion.


Pulmonary Insufficiency 1048

**General:** Elevated CO₂ levels.

**Ocular:** Tortuosity of conjunctival and retinal vasculature; retinal hemorrhages; papilledema; retinal edema.

**Clinical:** Vascular decompensation; elevation of cerebrospinal fluid pressure; polycythemia; chronic respiratory disease.


Pupil, Egg-Shaped 1049

**General:** Autosomal dominant; rare.

**Ocular:** Oval pupils; enlarged pupils; pupils react poorly to constricting stimuli.

**Clinical:** None.


Pupillary Membrane, Persistent 1050

**General:** Autosomal dominant.

**Ocular:** Remnants of pupillary membrane persist as strands and other irregular tissue in pupil; congenital cataract; corneal edema; Rieger syndrome; keratoconus.

**Clinical:** None.


Purtscher Syndrome (Fat Embolism Syndrome; Traumatic Retinal Angiopathy; Traumatic Liporrhagia; Valsalva Retinopathy of Duane; Duane Retinopathy)

General: Most frequently seen in accidents associated with sudden rise in blood pressure and congestion in the head and chest; presence of fat embolism may be the causative factor; neurovascular changes in retina referred to as traumatic retinal angiopathy; several mechanisms have been proposed, including compressive trauma and posttraumatic fat embolism; most likely mechanism appears to be leukocyte aggregation by activated complement factor 5 (C5A), which can occur in diverse conditions such as trauma, acute pancreatitis, and connective tissue disease.

Ocular: Retinal and preretinal hemorrhages over entire fundus; cotton-wool exudates, mainly posterior aspect; retinal edema; posterior and macular serous detachment; venous congestion and engorgement; papilledema; usually bilateral, although unilateral causes have been reported.

Clinical: Multiple fractures (mainly extensive crushing); lung congestion; dyspnea; lymphorrhagia; pancreatitis; scleroderma; dermatomyositis; lupus erythematosus; childbirth.


Pyoderma Vegetans (Dermatitis Vegetans)

General: Occurs in both sexes and all ages; frequently seen with preexisting eczema or infective dermatitis; nonspecific skin reaction; chronic; malnutrition and alcoholism are contributing factors.

Ocular: Lid lesions; keratitis.

Clinical: Chronic granulomatous growth with epithelial hyperplasia often with pustules, ulcers, multiple abscesses, and fistulae; lesions of mouth and nose.


Pyostomatitis

General: Reaction of chronic inflamed skin due to secondary infection; body resistance leads to folding of mucous membrane and development of a verrucous surface.

Ocular: Conjunctivitis; blepharitis.

Clinical: Early lesions are small pustules that can develop to large, weeping inflammatory plaques with foul odors; massive crust may form granulations of buccal mucosa, hand, soft palate, lips, and gingiva.


Q Fever

General: Acute rickettsial infection caused by *Coxiella burnetii*; at least 11 serotypes of this organism are capable of causing human infection; elevated inflammatory response results in granulomatous formation.

Ocular: Conjunctivitis; gangrene of eyelids; retinal hemorrhages; perivasculitis; episcleritis; optic neuritis; uveitis; papilledema; nystagmus; ocular motor nerve pareses; Miller-Fisher syndrome.

Clinical: Fever; severe headache; tissue necrosis; pneumonia; self-limited fever; endocarditis; hepatitis.

Radiation Injury (Gamma Rays; Infrared Rays; Microwaves; Radio Waves; X-Rays)  1055

**General:** Electromagnetic radiation can cause ionization in biologic tissues.

**Ocular:** Hyperemia of conjunctiva; corneal ulcer; punctate keratitis; keratoconjunctivitis sicca; blepharitis; ectropion; entropion; madarosis; poliosis; depigmentation of eyelids; uveitis; atrophy of lacrimal gland; cataracts; true exfoliation of lens capsule; orbital necrosis; retinal hemorrhage; macular degeneration; macular holes; neovascularization of retina; glaucoma; macular edema; retinal microvascular changes; optic nerve edema and hemorrhage; atrophy.

**Clinical:** Thermal burns of any part of the body; necrosis; carcinomas; edema.


Raeder Syndrome (Paratrigeminal Paralysis; Horton Headache; Histamine Cephalalgia; Ciliary Neuralgia; Cluster Headache; Periodic Migrainous Neuralgia)  1056

**General:** Interruption of sympathetic fibers about the carotid artery and involvement of the fifth nerve; meningioma and aneurysm of the internal carotid artery most frequent causes; prominent in males; possible pathogenetic mechanism of this condition is an ischemic injury of the gasserian ganglion.

**Ocular:** Mild enophthalmos; mild ptosis (unilateral); epiphora; scotoma possible; hypotonia; unilateral miosis; increased tear secretion; periorcular pain; Homer syndrome.

**Clinical:** Facial pain; occasionally weakness of the jaw muscles; headaches (V-region); hypertension; associated inflammatory processes are not infrequent.


Raeder JG. “Paratrigeminal” paralysis of oculo-pupillary sympathetic. *Brain* 1924; 47:149.


Raymond Syndrome (Raymond-Cestan Syndrome; Cestan [2] Syndrome; Pontine Syndrome; Disassociation of Lateral Gaze Syndrome)  1057

**General:** Lesion involving the pyramidal tracts as they traverse the pons; posterior longitudinal bundle and medial lemniscus may be involved; tumor and vascular thromoses are common causes; can be caused iatrogenically after neurosurgical procedures.

**Ocular:** Ipsilateral abducens palsy; paralysis of lateral conjugate gaze.

**Clinical:** Contralateral hemiplegia; anesthesia of the face, limbs, and trunk.

Raynaud Disease (Symmetrical Gangrene; Symmetrical Asphyxia) 1058

**General:** Primary, or idiopathic, form of paroxysmal digital cyanosis; possible abnormality of sympathetic nervous system; occurs in females between ages 15 and 40 years.

**Ocular:** Spasm of retinal arteries; papillitis; retrobulbar neuritis; amaurosis fugax; cotton-wool spots; retinal hemorrhages; transient corneal opacification.

**Clinical:** Intermittent attacks of pallor or cyanosis in fingers, precipitated by cold or occasionally by emotional upsets; atrophy of terminal fat pads and digital skin; gangrenous ulcers.

**References:**

Rebeitz-Kolodny-Richardson Syndrome 1059

**General:** Etiology unknown; possibly metabolic failure at cellular level; occurs in late middle age; neural achromasia with corticodentatonigral degeneration.

**Ocular:** Paralysis of ocular muscles.

**Clinical:** Clumsiness, slowness of movement of left limbs; severe impairment in control of muscular movements; postural abnormalities; tremor; severe contractures; ataxia; severe contractures; dysphagia; speech impairment; Babinski sign.

**References:**

Refsum Syndrome (Heredopathia Atactica Polyneuritiformis Syndrome; Phytanic Acid Oxidase Deficiency; Phytanic Acid Storage Disease; Refsum-Thiebaut Syndrome) 1060

**General:** Autosomal recessive; disorder of lipid metabolism; interstitial hypertrophic polyneuropathy; delamination of myelin sheaths; onset usually between ages 4 and 7 years; caused by deficiency of phytanic acid hydroxylase.

**Ocular:** Progressive external ophthalmoplegia; night blindness; visual field constriction; pupillary abnormalities; corneal opacities; retinal degeneration beginning in macula; retinitis pigmentosa; cataracts.

**Clinical:** Spinocerebellar ataxia; deafness (progressive); polyneuritis-like effect on limbs; central nervous system degeneration; ichthyosis; sensory changes; wasting of extremities; complete heart block; relapses and remissions in adolescence; normal intelligence.

**References:**

Reimann Syndrome (Hyperviscosity Syndrome) 1061

**General:** Frequently found in association with Waldenström syndrome, other hyperglobulinemias, and occasionally with myeloma, reticulum sarcoma, and other tumors; increase in y-globulin or other dysproteinemias that affect blood viscosity (see Bing-Neel Syndrome; Waldenström Syndrome); leukemia.

**Ocular:** Nystagmus; tortuosity of conjunctival vessels; sludging phenomenon with aggregation of intravascular erythrocytes; retinal vascular tortuosity and sludging; retinal hemorrhages and exudates; micro aneurysms of various degrees and sizes; central retinal vein occlusion.

**References:**
Clinical: Mucous membrane; hemorrhages; headaches; paraesthesia; ataxia; heart failure and low pulse pressure; partial loss of hearing; anorexia; vertigo; dyspnea; syncope; convulsions; peripheral edema; sausage-shaped veins.


Reiter Syndrome (Fiessinger-Leroy Syndrome; Conjunctivo-Urethro-Synovial Syndrome; Idiopathic Blennorreal Arthritis Syndrome; Polyarthritis Enterica)

General: Etiology unknown; males; onset ages 16 to 42 years; probably a combined infectious/autoimmune pathogenetic mechanism; reactive arthritis probably associated with infection with many different species of microorganisms; HLA-B27 confers disease susceptibility to infection.

Ocular: Sterile mucopurulent conjunctivitis, usually bilateral; photophobia; epiphora; iritis; keratitis; uveitis; paralysis of extraocular muscles; optic neuritis; secondary glaucoma; hypopyon; hyphema.

Clinical: Skin erythema; genital ulcerations; urethritis with discharge; cystitis with dysuria, abacterial pyuria, and hematuria; arthritis with pain, swelling, heat, and effusion; fever; weight loss; fatigue; malaise; fever; diarrhea; oral mucosal lesions; arthralgia.


Relapsing Fever (Recurrent Fever)

General: Acute infectious disease caused by Borrelia transmitted by lice; characterized by recurrent bouts of fever separated by relatively asymptomatic periods; there is an endemic form of rheumatic fever transmitted by tick vectors and spirochetes of the genus Borrelia.

Ocular: Extraocular muscle paralysis; uveitis; interstitial keratitis; hypopyon; conjunctivitis; optic nerve atrophy; subconjunctival and retinal hemorrhages; ptosis; mydriasis; retinal venous occlusion.

Clinical: Toxemia and febrile paroxysms separated by afebrile periods.


Relapsing Polychondritis (Jaksch Wartenhost Syndrome; Meyenburg-Altherz-Vehlinger Syndrome; Von Meyenberg II Syndrome)

General: Episodic, yet generally progressive; onset usually in middle life; possibly caused by lysosomal labilizing factor of endogenous or exogenous toxic nature or immunologic reactions; possible association with Reiter syndrome.

Ocular: Conjunctivitis; corneal ulcer; exophthalmos; panophthalmitis; phthisis bulbi; proptosis; optic neuritis; papilledema; retinal detachment; blue sclera; episcleritis; scleromalacia; vitreous opacity; cataracts; nyctagmus; retinal artery thrombosis; keratoconjunctivitis sicca; secondary glaucoma; scotoma; uveitis; paresis of third or sixth nerve; conjunctival mass (salmon patch); chorioretinitis.

Clinical: Destruction of cartilage and eventual replacement with connective tissue; polyarthritis; chondritis; tracheal collapse; bronchial collapse; anemia; liver dysfunction; death; malaise; fever; dyspnea; changes in pitch of voice; hearing impairment; vertigo; deformed ears; aortic valve insufficiency.


Renal Failure

**General:** Absence of renal function.

**Ocular:** Cotton-wool spots; retinal edema; optic disk edema; conjunctival calcium deposits; band keratopathy; cortical blindness; severe retinopathy is more likely to be found in patients with renal insufficiency; patients undergoing hemodialysis are at increased risk of elevated intraocular pressure, particularly in eyes that have undergone vitrectomy.

**Clinical:** Hypertension; azotemia; hypervolemia; metabolic disturbances; hyponatremia; hypercalcemia.


Renal Transplantation

**General:** Ocular complication due to systemic immunosuppressive drugs used to prevent rejection of the transplanted kidney.

**Ocular:** Cataracts; steroid-induced glaucoma; cytomegalovirus retinitis; fungal endophthalmitis; conjunctivitis; diffuse anterior scleritis with possible association with OKT3 monoclonal antibody therapy for renal transplant rejection; typical herpetic dendritic keratitis.

**Clinical:** Renal failure; kidney transplant.


Rendu-Osler Syndrome (Rendu-Osler-Weber Syndrome; Hereditary Hemorrhagic Telangiectasia; Babington Disease; Goldstein Hematemesis; Osler Syndrome [2])

**General:** Etiology unknown; autosomal dominant in Jews; repeated epistaxis begins in childhood; gastrointestinal hemorrhages with melena and hematemesis manifest in middle and later life.

**Ocular:** Star-shaped angiomas of the palpebral conjunctiva; intermittent filamentary keratitis; small retinal angiomas and occasionally retinal hemorrhages; subconjunctival hemorrhages; small retinal arteriovenous malformations; bloody tears; conjunctival telangiectasias.

**Clinical:** Epistaxis; hematuria; melena; angioma of the pharynx and oral and nasal mucosa; angiomas on lips, face, and upper extremities; cyanosis; polycythemia.


Reticular Degeneration of Pigment Epithelium (RDPE)

**General:** Etiology unknown; associated with age-related macular degeneration.
**Ocular:** Hypopigmentation of retinal pigment epithelium interspersed with hyperpigmented lines forming a coarse netlike pattern of irregular polygons; multiple drusen of peripheral fundus; choroidal nevus; central retinal vein occlusion.


**Reticular Pigmentary Retinal Dystrophy of Posterior Pole (Sjögren Disease) 1069**

**General:** Autosomal recessive; characterized by peculiar network of black-pigmented lines in posterior pole of retina.

**Ocular:** Fishnet-like knots on posterior pole of retina; drusen.

**Clinical:** None.


**Reticulum Cell Sarcoma (Non-Hodgkin Lymphoma) 1070**

**General:** Autosomal recessive; large-cell lymphoma with chronic inflammation with a predominance of cells in vitreous cavity; average age at time of diagnosis is 60 years; female to male ratio is approximately 2:1; 80% bilateral (frequently asymmetrical).

**Ocular:** Chronic uveitis; choriotinal lesions; mycosis fungoides; necrosis of orbital tissues; phthisis bulbi; endophthalmos; exophthalmos; exudative retinal detachment; iris neovascularization; glaucoma; branch retinal vein occlusion; macular edema; optic neuropathy; vitreous hemorrhage; partial cranial nerve III palsy; multiple retinal pigment epithelium masses.

**Clinical:** Lymphocytic hyperplasia; fever; anemia; thrombocytopenia; liver and spleen enlargement; associated with immune dysfunction states, such as acquired immunodeficiency syndrome, or following transplantation.


**Retinal Arteries, Tortuosity 1071**

**General:** Autosomal dominant; retinal vascular tortuosity is the isolated physical finding but may be associated with a variety of ocular and systemic anomalies; tortuosity predominantly found in the arteriolar tree.

**Ocular:** Retinal vascular tortuosity; foveal hemorrhage; macular and paramacular hemorrhages; retinal hemorrhages; peripapillary retinal hemorrhage; juxtapapillary retinal hemorrhage; conjunctival hemorrhage; retinal telangiectasia; asteroid hyalitis; amblyopia; myopia; hyperopia; glaucoma; Coats disease; von Hippel-Lindau syndrome; racemose aneurysms of the retina; arteriolar ectasia.

**Clinical:** Polycythemia; splenomegaly; recurrent nosebleeds; nasal telangiectasia; pulmonary emphysema; systemic hypertension; aortic coarctation; hereditary hemorrhagic telangiectasia; leukemia; macroglobulinemia; cryoglobulinemia; sickle cell disease; familial dysautonomia; Maroteaux-Lamy syndrome; Fabry disease; hypogammaglobulinemia; intracranial or facial aneurysmal abnormalities; migraine headaches; psoriasis; colitis; diabetes mellitus; hypertension.


Retinal Cone Degeneration

**General:** Autosomal dominant; diffuse cone degeneration; progressive loss of visual acuity; macular lesion has a bull's-eye appearance produced by a central area of uninvolved epithelium.

**Ocular:** Photophobia; defective color vision; loss of side vision; night blindness; macular lesion; poor central acuity and visual field scotomata closer to fixation compared to patients with retinal rod degeneration.

**Clinical:** None.


Retinal Detachment

**General:** Autosomal dominant; multilayered retinal tissue separates along an interface formed in course of embryonic development.

**Ocular:** Retinal tear or hole; retinal detachment.

**Clinical:** None.


Retinal Disinsertion Syndrome

**General:** None.

**Ocular:** Subluxation of the lens; microphthalmos; bilateral keratoconus; retinal detachment.

**Clinical:** None.


Retinal Ischemic Syndrome

**Ocular:** Bilateral conjunctival injection; iris rubeosis; posterior synechia; retinal vascular tortuosity; retinal detachment; Axenfeld’s syndrome; central retinal artery occlusion; glaucoma; corneal anesthesia; uveitis; ischemic retinopathy

**Clinical:** Associated with partial occlusion of carotid artery; diabetes, hypercholesterolemia; tetralogy of fallot


Retinal Vascular Hypoplasia with Persistence of Primary Vitreous

**General:** Bilateral congenital retinopathy characterized by retinal vascular hypoplasia and persistence of primary vitreous; etiology unknown.

**Ocular:** Buphthalmos; microphthalmia; fixed and dilated pupils; neovascularization of iris; glaucoma; cataract; white opaque fibrovascular retrolental membrane; retinal detachment; vitreous hemorrhage; retinal vascular hypoplasia.

**Clinical:** None.
Retinitis Pigmentosa, Deafness, Mental Retardation, and Hypogonadism

**General:** Autosomal recessive; similar to Laurence-Moon-Biedl-Bardet, Alstrom, and Usher syndromes, with the difference being the absence of polydactyly.

**Ocular:** Nystagmus; hyperplasia and thickening of prickle cell layer to lids or corner of eyes; myopia; retinitis pigmentosa; keratoconus.

**Clinical:** Acanthosis nigricans; multiple keloids; gynecomastia, small testes in males; oligomenorrhea; mental retardation; deafness; hypogonadism; glucose intolerance; hyperinsulinism.


Retinoblastoma

**General:** Malignant tumor arising in one or both retinas of young children, usually under the age of 2 years; usually unilateral; autosomal dominant; most common intraocular malignancy of childhood; incidence is one in 20,000 live births; origin is questionably neuroectodermal cells capable of multipotentiality; one third of patients have heritable (bilateral or have a positive family history) autosomal dominant and two thirds are sporadic; genetic transmission obeys two-mutation hypothesis of Knudson; trilateral retinoblastoma is bilateral retinoblastoma plus midline central nervous system tumor (most commonly pinealoma); most common second tumor is an osteogenic sarcoma (begins in second decade).

**Ocular:** Hyphema; hypopyon; corneal tumor; lid edema; endophthalmitis; corneal tumor; lid edema; endophthalmitis; papilledema; panophthalmitis; retinoblastoma extension into orbit and choroid; cat's-eye reflex; leukocoria; mydriasis; vitreous hemorrhage tumor seeding; esotropia; exotropia; glaucoma; visual loss.

**Clinical:** Metastasis into the lymph system, bone marrow, and subarachnoid space; basal meningitis; death.


Retinohypophysary Syndrome (Benign Retinohypophysary Syndrome; Lijo Pavia-Lis Syndrome)

**General:** Alterations of the bony structure of the sella turcica with decalcifications and osteolysis of the posterior clinoid process; all ages; more frequent in women.

**Ocular:** Superior nasal field contraction; narrowing of retinal vessels; macular edema; optic neuritis; optic atrophy; visual field defects.

**Clinical:** Glycosuria; headache; vertigo; psychic disturbances.


Retinopathy, Pigmentary, and Mental Retardation (Mirhosseini-Holmes-Walton Syndrome)

**General:** Autosomal recessive; this disorder may be the same as (or alleli) to Cohen syndrome.

**Ocular:** Pigmentary retinal degeneration; cataract; keratoconus.
Clinical: Microcephaly; severe mental retardation; hyperextensible joints; scoliosis; arachnodactyly; hypogonadism.


Retinoschisis (RS) 1081

General: Sex-linked; may not manifest until middle life.

Ocular: Intraretinal splitting due to degeneration or detachment of retina; retinal atrophy with sclerosis of the choroid; cystic maculopathy.

Clinical: None.


Retinoschisis, Acquired (Acquired Retinoschisis [RS]) 1082

General: Present in 4% to 22% of normal population over age 40 years; splitting in the outer plexiform layer; occasionally in the inner nuclear layer; generally asymptomatic.

Ocular: Slowly progressive; may cause retinal detachment when there are breaks in both outer and inner layers of retinoschisis.

Clinical: None.


Retinoschisis, Autosomal Dominant 1083

General: Autosomal dominant; degenerative abnormal splitting of retinal sensory layers.

Ocular: Loss of retinal function; retinal degeneration; macular degeneration.

Clinical: None.


Retinoschisis, Congenital 1084

General: X-linked recessive; nearly always found in males.

Ocular: Retinal splitting usually occurs in the nerve fiber layer; slow progression; frequently affects the macula; associated with vitreous hemorrhage; strabismus; nystagmus; retinal folds radiating from the fovea; macular pigmentary mottling; retinal detachment (rare complication).

Clinical: None.


Retinoschisis of Fovea

**General:** Autosomal recessive.

**Ocular:** Foveal dystrophy; rod-cone dystrophy; nyctalopia; hyperopia; paramacular tapetal sheen reflex.

**Clinical:** None.


Retrorenal Fibroplasia (RLF; Retinopathy of Prematurity)

**General:** Bilateral disease seen primarily in premature infants with immature retinal vessels; excessive use of oxygen responsible for the majority of cases, but disease is seen despite oxygen restrictions or even when no oxygen supplementation is used; known factors that correlate with degrees of retinopathy of prematurity are low birth weight, short gestational age, length of time with supplemental oxygen, length of time on a mechanical ventilator; role of excessive light in newborn nurseries also has been proposed.

**Ocular:** Anterior or posterior synechiae; neovascularization of iris; pallor of optic disk; dragged disk; attenuated vessels; retinal detachment; dilation of veins; retinal hemorrhage; retrolental mass; vascular tortuosity; vasoconstriction of retina; retinal pigmentary changes; vitreous haze; vitreous traction; vitreous hemorrhages; cataract; leukocoria; myopia; shallow anterior chamber; opaque retrolental membrane; ciliary body drawn anteriorly; ciliary process around dilated pupil; absent pupillary reflexes; keratoconus; associated strabismus; amblyopia.

**Clinical:** Low birth weight; prematurity.


Retroparotid Space Syndrome (Villaret Syndrome; Posterior Retroparotid Space Syndrome)

**General:** Lesions (traumatic, inflammatory, tumors) involving cranial nerves IX to XII and the cervical sympathetic (see Jugular Foramen Syndrome [Vernet Syndrome]).

**Ocular:** Enophthalmos; ptosis; lagophthalmos; epiphora; miosis; may produce sympathetic overactivity resulting in increased sympathetic outflow (i.e., pupillary dilation, widened palpebral fissure, and facial sweating).

**Clinical:** Homolateral paralysis cranial nerves IX to XII, with dysphagia and loss of taste in posterior third of the tongue; dysphonia; paralysis of sternocleidomastoid and trapezius muscles; paralysis cranial nerve VII occasionally.


Reye Syndrome (Acute Encephalopathy Syndrome)

**General:** Etiology unknown, although some relation to ingestion of aspirin with febrile illnesses, especially varicella and influenza, have been reported; both sexes; onset 6 months to 10 years; acute metabolic encephalopathy largely affecting children and adolescents; pathogenesis is controversial, although there is new evidence for a generalized defect in intramitochondrial enzyme processing resulting in lowered ratio of adenosine triphosphate to adenosine diphosphate.

**Ocular:** Cortical blindness; dilated pupils with absent or sluggish reaction to light; papilledema.

**Clinical:** Respiratory infections with recovery between 3 and 21 days; vomiting after recovery from infection; dyspnea; hypotonia; coma; convulsions; fever; flexion of elbows and hands.
Rhabdomyosarcoma

**General:** Most common malignant orbital neoplasm of childhood; usually occurs before age 10 years; more commonly seen in males; rarely may develop in adults; shows evidence of striated muscle differentiation; has been divided into three histopathologic types: embryonal, alveolar, and pleomorphic.

**Ocular:** Choroidal folds; corneal edema; exposure keratitis; rhabdomyosarcoma of orbit or extraocular muscles; decreased motility; proptosis; papilledema; orbital edema; enlarged optic foramen; erosion of bony walls of orbit; pupil irregularity; epiphora; glaucoma; visual loss; nasolacrimal duct obstruction; conjunctival mass.

**Clinical:** Metastasis to the lymph system, bone marrow, and lungs; headaches.


RHE Syndrome (Retino-Hepato-Endocrinologic Syndrome)

**General:** More common in females.

**Ocular:** Total color blindness; thinning of retinal vessels with atrophy; optic disk pallor; poor photopic vision; progressive cone dystrophy.

**Clinical:** Degenerative liver disease; endocrine dysfunction; hypothyroidism; diabetes mellitus; repeated abortions or infertility; elevated blood creatine phosphokinase.


Rheumatic Fever

**General:** Streptococcal infection; mechanisms may involve immune cross-reactivity between bacterial heat shock proteins and similar proteins in normal human tissues.

**Ocular:** Lid edema; characteristic arborizations of conjunctiva; subconjunctival hemorrhages; episcleritis; scleritis; tenonitis; uveitis; retinal detachment; central retinal artery occlusion; optic neuritis.

**Clinical:** Hematuria; proteinuria; proliferative glomerulonephritis; microembolization; calcific emboli; sore throat; fever; chorea; erythema marginatum; disseminated encephalomyelitis.


Rheumatoid Arthritis (Adult)

**General:** Systemic disease of unknown cause; more common in women (3:1); thought to have a strong autoimmune pathogenesis with positive immunoglobulins M, G, and A directed against the Fc portion of immunoglobulin G.

**Ocular:** Sjögren syndrome; episcleritis; scleritis; keratitis; corneal ulcers; corneal perforation; uveitis; motility disorders; dry eyes; posterior scleritis (rare).

**Clinical:** Synovitis; stiffness; swelling; cartilaginous hypertrophy; joint pain; fibrous ankylosis; malaise; weight loss; vasomotor disturbance.


Rhinoceroma (Klebsiella Rhinoscleromatis)

**General:** Chronic granulomatous disease; Gram-negative bacillus; cicatricial deformities; chronic progressive granulomatous infection of the upper airways caused by the bacterium *Klebsiella rhinoscleromatis*.

**Ocular:** Conjunctivitis; chronic dacryocystitis; lid inflammation.

**Clinical:** Granulomas affecting nose and upper respiratory tract causing sclerosis and deformities; airway obstruction; leprosy; paracoccidioidomycosis; sarcoidosis; basal cell carcinoma; Wegener granulomatosis; also may occur in immunocompromised human immunodeficiency virus patients.


Rieger Syndrome (Axenfeld-Rieger Syndrome; Dysgenesis Mesodermalis Corneae et Irides; Dysgenesis Mesostromalis; Axenfeld Posterior Embryotoxon-Juvenile Glaucoma)

**General:** Autosomal dominant; neural crest abnormality; 50% of patients develop glaucoma.

**Ocular:** Microphthalmia; congenital glaucoma; iris hypoplasia; deformed and acentric pupil; anterior synechiae; aniridia; microcornea; corneal opacities in Descemet membrane parallel to the limbus; dislocated lens; optic atrophy; cataract; strabismus; ptosis; hypertelorism; keratoconus; posterior embryotoxon; broad iris processes to embryotoxon; iris stromal hypoplasia; corectopia; polycoria; secondary glaucoma.

**Clinical:** Face wide; hypodontia; underdeveloped maxilla; teeth deformities; myotonic dystrophy; facial anomalies; maxillary hypoplasia, protrusion of the lower lip, broad, flat nose; dental anomalies include absent teeth, piglike incisors, and decreased crown size; hypospadias.


Rift Valley Fever

**General:** Acute viral infection transmitted by mosquito that occurs in regions of Africa.

**Ocular:** Central serous retinitis; central scotoma; macular scarring; retinal vascular occlusion; uveitis; retinitis; choroiditis.

**Clinical:** Fever; chills; severe headache; muscle and joint pain; leukopenia; bradycardia.

Riley-Day Syndrome (Congenital Familial Dysautonomia)  1096

**General:** Autosomal recessive; occurs in Ashkenazi Jewish population; impaired catechol metabolism; manifested in first few days of life; characterized by developmental loss of neurons from the sensory and autonomic nervous systems.

**Ocular:** Congenital failure of tear production; corneal anesthesia; neuroparalytic keratitis; keratitis sicca; corneal ulcers; optic atrophy.

**Clinical:** Excessive salivation; failure to thrive; recurrent respiratory infections; diarrhea; insensitivity to pain; spontaneous fractures; pandysautonomia; orthostatic hypotension; gastrointestinal paresis; decreased fungiform papillae on the tongue.


Riley-Smith Syndrome (Bannayan-Riley-Ruvalcaba Syndrome)  1097

**General:** Etiology unknown; possibly heterozygous condition (single autosomal gene); macrocephaly-pseudopapilledema-multiple hemangiomas; noted at birth; has been proposed that there is overlap with the Bannayan-Zonana syndrome and the Ruvalcaba-Myhre syndrome; autosomal dominant inherited condition.

**Ocular:** Pseudopapilledema; prominent Schwalbe line; visible corneal nerves; ocular hypertelorism; strabismus; amblyopia.

**Clinical:** Macrocephaly; subcutaneous hemangiomas; pulmonary infections; mild-to-severe mental retardation; skin hemangiomas; hypotonus; gastrointestinal polyps; pigmented maculae on the skin.


Ring Chromosome 6 (Aniridia, Congenital Glaucoma, and Hydrocephalus)  1098

**General:** Rare disorder associated with various congenital anomalies; autosomal dominant with recessive sporadically reported.

**Ocular:** Microphthalmia; aniridia; congenital uveal ectropion; Rieger anomaly; congenital glaucoma; corneal clauding; prominent Schwalbe line with attached iris strands; hypopigmented fundi; hypoplasia of iris stroma; strabismus; ptosis; nystagmus; megalocornea; iris coloboma; optic atrophy; hypertelorism; antimongoloid slant of palpebral fissures; ectopic pupils; angle anomalies; posterior embryotoxon; microcornea; colobomatus.

**Clinical:** Hydrocephalus; agenesia of corpus callosum; congenital heart defects; mental retardation; low-set malformed ears; broad nasal bridge; micrognathia; short neck; hand anomalies; high-arched palate; widely spaced nipples; deformity of feet; respiratory distress syndrome; hyperbilarubinemia; hypocalcemia; anemia; seizure; bulging anterior fontanelle.


Ring D Chromosome

**General:** Variant of the chromosome 13 deletion syndrome; ring chromosomes.

**Ocular:** Hypertelorism; epicanthal folds; ptosis; microphthalmos; uveal colobomas; abnormal palpebral fissure; strabismus; retinoblastoma.

**Clinical:** Aplasia of thumbs; mental and physical retardation; trigonocephaly; malrotation of ear; micrognathia; hypoplastic nipple; widely spaced first and second toes.


### Ring Dermoid Syndrome

**General:** Autosomal dominant; usually bilateral.

**Ocular:** Dermoid choristoma; conjunctival plaques of keratinization; corneal lipid deposition; irregular corneal astigmatism; amblyopia; concomitant strabismus.


### Rinx Disease (VSX1 Mutation)

**General:** Mutation of the homeobox transcription factor gene VSX1 (RINX) Wide interpupillary distance; hypertelorism; anomalies of corneal endothelium; abnormal cone bipolar cells

**Clinical:** Empty sella turcica; posterior fossa cyst; anterior encephalocele; hypertelorism; hydrocephalus


### Roberts Pseudothalidomide Syndrome

**General:** Rare autosomal recessive disorder characterized by prenatal and postnatal growth retardation, limb defects, and craniofacial anomalies.

**Ocular:** Cataracts; glaucoma; microcornea; corneal clouding.

**Clinical:** Patients usually do not survive past I month; patients often are mentally retarded.


### Robinow-Silverman-Smith Syndrome (Achondroplastic Dwarfism; Mesomelic Dwarfism; Robinow Dwarfism)

**General:** Autosomal dominant; both sexes affected; present from birth.

**Ocular:** Hypertelorism; epicanthal folds.

**Clinical:** Dwarfism; shortened forearms; bulging forehead; depressed nasal bridge; hypoplastic mandible; small, upturned nose; micrognathia; crowded teeth; penile hypoplasia.


### Rochon-Duvigneaud Syndrome (Superior Orbital Fissure Syndrome)

**General:** Inflammatory, traumatic, tumor, or vascular lesions such as meningioma of the sphenoid, carotid aneurysm, and arachnoiditis; infections originating in the maxillary sinus.

**Ocular:** Mild exophthalmos; lid edema; partial or complete ophthalmoplegia (III, IV, and VI); decreased corneal sensitivity; papilledema; optic atrophy.

**Clinical:** Decreased sensitivity in area of nasociliary, lacrimal, frontal, and ophthalmic nerve distribution; may result from a metastatic tumor.


**Rocky Mountain Spotted Fever**

**General:** Acute systemic disease caused by *Rickettsia rickettsii* transmitted by a wood tick or dog tick.

**Ocular:** Conjunctivitis; optic atrophy; cotton-wool spots; scotoma; uveitis; optic neuritis; paralysis of accommodation; paralysis of extraocular muscles; retinal vascular occlusion; vitreal opacity; hypopyon; anterior uveitis with fibrin clots.

**Clinical:** Fever; chills; headache; muscle aches; rash.


**Rollet Syndrome (Orbital Apex-Sphenoidal Syndrome)**

**General:** Lesion in the apex of the orbit (neoplastic, hemorrhagic, or inflammatory) involving the third, fourth, and sixth cranial nerves, the ophthalmic branch of the fifth sympathetic fibers when they pass through the sphenoidal fissure, and the optic nerve; manifestations vary greatly with extent of lesion; pain is frequent early sign; orbital fissure syndrome and sphenocavernous syndrome are similar; sudden onset.

**Ocular:** Exophthalmos; ptosis; hyperesthesia or anesthesia of the upper lid; ophthalmoplegia (partial or complete); wide pupil with loss of reaction on accommodation; neuralgic pain in the region of the ophthalmic branch of cranial nerve V; anesthesia of the cornea; papilledema; optic neuritis; optic atrophy; diplopia; herpes zoster ophthalmicus.

**Clinical:** Hyperesthesia or anesthesia of the forehead; inflammation of cavernous sinuses; meningoencephalitis.


**Romberg Syndrome (Parry-Romberg Syndrome; Progressive Hemifacial Atrophy; Progressive Facial Hemiatrophy; Facial Hemiatrophy)**

**General:** Autosomal dominant; irritation in the peripheral trophic sympathetic system; onset in the second decade; both sexes affected.

**Ocular:** Enophthalmos; outer canthus lowered; absence of nasal portion of eyebrow; ptosis; paresis of ocular muscles; miosis; iritis; iridocyclitis; heterochromia iridis; keratitis; neuroparalytic keratitis; cataracts; choroiditis; Fuchs heterochromic cyclitis; retinal vascular abnormalities; association with Coats syndrome and exudative stellate neuroretinopathy; scleral melting.

**Clinical:** Atrophy of soft tissue on one side of the face, including tongue; trigeminal neuralgia and/or paresthesia; alopecia and poliosis not uncommon.


Rosenberg-Chutorian Syndrome

**General:** Recessive inheritance; inheritance is considered X-linked semidominant.

**Ocular:** Optic atrophy.

**Clinical:** Polyneuropathy; neural hearing loss.


Rosenthal-Kloepefer Syndrome

**General:** Autosomal dominant; unilateral or bilateral; onset in early childhood; hyperplasia and furrowing of skin, especially of face and scalp, beginning about the fourth decade of life; etiology unknown; affects both sexes.

**Ocular:** Prominent lateral aspect of the supraorbital arch of the frontal bone; corneal leukomas (initially superficial stromal infiltrations that develop to thick and dense corneal opacities).

**Clinical:** Acromegaly; thickening of the bony skull; prominent lower jaw; skin changes (with verticis gyrata); abnormal ridges and creases of the palms.


Rothmund Syndrome (Rothmund-Thomson Syndrome; Telangiectasia-Pigmentation-Cataract Syndrome; Ectodermal Syndrome; Congenital Poikiloderma with Juvenile Cataract)

**General:** Autosomal recessive; more common in females (2:1); Werner syndrome in adults has certain similarities to this syndrome; inflammatory phase progresses to atrophy and telangiectasia; onset at age 3 to 6 months.

**Ocular:** Eyebrows may be sparse or absent; hypertelorism; cilia sometimes are diminished or absent; trichiasis; epiphora; cataracts (anterior subcapsular, posterior stellate, or perinuclear type); corneal lesions; retinal hyperpigmentation; keratoconus; strabismus; epibulbar dermoids.

**Clinical:** Poikiloderma; hypogonadism; hypomenorrhea; head deformity (enlarged with depressed nasal bridge as well as microcephaly); small stature, with short or malformed distal phalanges; aplasia cutis congenita (congenital absence of skin in one or more areas); alopecia.


Roussy-Cornil Syndrome

**General:** Etiology unknown; sporadic hypertrophic neuropathy; onset in second to third decade of life or later; progressive; occasionally, remissions and exacerbations.

**Ocular:** Sluggish pupils; disturbed vision.

**Clinical:** Nerves palpable and tender; peripheral atrophy; tendon reflexes diminished or abolished; scoliosis; muscle fasciculations; ataxia; lancinating pains.
**Roy Syndrome**

**General:** Long history of smoking tobacco.

**Ocular:** Cataract, usually unilateral, posterior subcapsular, and cortical.

**Clinical:** Emphysema; cardiac disorder; cancer.


**Roy Syndrome II**

**General:** Long history of smoking tobacco.

**Ocular:** Cataract; nuclear cataract may be cortical or posterior subcapsular cataract.

**Clinical:** Emphysema; cardiac disorder; cancer.


**Rubella Syndrome (Congenital Rubella Syndrome; German Measles; Gregg Syndrome)**

**General:** Rubella infection of the mother during first trimester of pregnancy; ocular disease is the most commonly found abnormality in patients with congenital rubella syndrome (75%); multiorgan disease is common (greater than 75%); no significant association has been found between gestational age and time of maternal infection and incidence of individual ocular conditions.

**Ocular:** Nystagmus; glaucoma; corneal haziness; cataracts; retinal pigmentary changes; appearance and central distribution of lesions are quite distinguishable from retinitis pigmentosa; retinopathy is not progressive and has little, if any, effect on vision; waxy atrophy of optic disk; conjunctivitis; megalocornea or microcornea; buphthalmos; microphthalmos; uveitis; iris atrophy; spherophakia; strabismus.

**Clinical:** Low birth weight; diarrhea; pneumonia; urinary infection; hearing loss; heart disease; hepatosplenomegaly; mental retardation; inguinal hernias; ataxia; cardiac abnormalities.


**Rubinstein-Taybi Syndrome**

**General:** Inheritance polygenic or multifactorial; rare.

**Ocular:** Antimongoloid slant of lid fissure; epicanthus; long eyelashes and highly arched brows; strabismus; myopia; hyperopia; iris coloboma; cataract; optic atrophy; ptosis; retinal detachment.

**Clinical:** Motor and mental retardation; broad thumbs and toes; highly arched palate; allergies; heart murmurs; anomalies of size, shape, and position of ears; dwarfism; cryptorchidism.


Rud Syndrome

**General:** Etiology unknown; immature nerve cells and decreased number of cells; Betz cells in the motor cortex show chronic chromatolysis; excess of oligodendroglia in the frontal cortex; relationship to tuberous sclerosis and neurofibromatosis; X-linked inheritance has been reported.

**Ocular:** Retinal pigmentary degeneration.

**Clinical:** Epilepsy; infantilism; idiocy; congenital ichthyosis; muscular atrophy; male hypogonadism.


Rud E. Et Tilfaelde af Infantilsms med Tetani, Epilepsy, Polyneuritis, Ichthiosis og Anaemi of Pernicios Type. *Hospitalstidende* 1927; 70:525.


Russell Syndrome

**General:** Onset between 3 months and 2 years; caused by tumors of the anterior portion of the thalamus (usually astrocytoma), optic chiasm, midcerebellar region, and midline ependymoma; erosion under the anterior clinoid processes that causes a characteristic J-shaped sella in lateral skull films.

**Ocular:** Lid retraction; nystagmus (horizontal, vertical, or rotatory); homonymous hemianopsia; optic nerve atrophy.

**Clinical:** Extreme emaciation; euphoria; pale skin.


Sabin-Feldman Syndrome

**General:** Etiology unknown; similar to toxoplasmosis; results of toxoplasma dye and complement fixation tests are negative; onset in early infancy.

**Ocular:** Microphthalmia; strabismus; fixed pupils; posterior lenticous; microcornea; chorioretinitis or atrophic degenerative chorioretinal changes; optic atrophy.

**Clinical:** Cerebral calcifications (infrequent); convulsions (frequent); microcephaly; hydrocephalus.


Saint Anthony Fire (Erysipelas)

**General:** Acute localized inflammation of the skin and subcutaneous tissue; erysipelas is a febrile infection of the skin and subcutaneous tissue, most commonly caused by *Streptococcus*, characterized by the acute onset of a red, indurated expanding plaque that nearly disappears with the use of antibiotics; sometimes caused by *Staphylococcus*.

**Ocular:** Conjunctivitis; blepharitis; elephantiasis and gangrene of lid; ptosis; dacryocystitis; cellulitis of orbit; keratitis; panophthalmitis; uveitis; eyelid involvement.

**Clinical:** Edema; fever; rigor; vesicles; tenderness; headache; vomiting; localized pain.


Saldino-Mainzer Syndrome

**General:** Autosomal recessive; Leber congenital amaurosis associated with familial juvenile nephronophthisis and cone-shaped epiphysis of the hands; similar to Senior-Loken syndrome, with the difference being cone-shaped epiphysis.

**Ocular:** Tapetoretinal degeneration; retinal atrophy; Leber congenital amaurosis; retinitis pigmentosa.

**Clinical:** Nephronophthisis; cone-shaped epiphyses of hands and feet; flared ribs; hypoplastic pelvis; brachydactyly; hyperparathyroidism; osteomalacia; osteopetrosis; renal failure.


Sandhoff Disease (Gangliosidosis Type 2 [GM2])

**General:** Hereditary cerebromacular degeneration-sphingolipidoses; onset by age 6 months; autosomal recessive inheritance; enzyme defect; caused by deficiency of hexosaminidases A and B; defect localized to chromosome 5 (5q13).

**Ocular:** Cherry-red spot of macula; visual loss; clinically identical to Tay-Sachs disease.

**Clinical:** Motor retardation; doll-like facies; dementia; hyperacusis; frequent respiratory infections; variable hepatosplenomegaly.


Sandifer Syndrome (Hiatal Hernia-Torticollis Syndrome)

**General:** Inheritance not known; males affected; hiatal hernia.

**Ocular:** Strabismus (not related to existing torticollis).

**Clinical:** Rotation of the head to one shoulder with stretching of the neck (more pronounced during eating and reading); epigastric pain associated with vomiting, primarily in infancy; malnutrition; hiatal hernia; asthenia.


Sands of the Sahara Syndrome (Diffuse Lamellar Keratitis)

**General:** Interface inflammation after laser in situ keratomileusis (LASIK).

**Ocular:** Interface inflammation after LASIK is a rare but potential sight-threatening complication; syndrome presents 1 to 5 days after LASIK; affected patients often complain of decreased or cloudy vision, foreign body sensation, and photophobia; symptoms may be mild or severe; cause of the interface debris is unknown, but microkeratome material is implicated.


Sanfilippo-Good Syndrome (Heparitinuria; Mucopolysaccharidosis III; MPS III) 1124

**General:** Autosomal recessive; excess urinary excretion of heparitin sulfate (see Hunter Syndrome; Hurler Syndrome; Maroteaux-Lamy Syndrome; Morquio Syndrome; Scheie Syndrome). Lack of a β-galactosaminidase-like enzyme causing accumulation of glycolipids, acid mucopolysaccharides, and their precursors; both sexes affected; death occurs by second decade in the majority of cases; autosomal recessive; divided into type A (with decreased levels of heparan sulfatase) and type B (with decreased levels of N-acetyl-α-D-glucosaminidase).

**Ocular:** Night blindness; slight narrowing of retinal vessels; pigment deposits in the fundi; bushy eyebrows; coarse eyelashes; acid mucopolysaccharide deposits in cornea, iris, lens, and sclera; retinal degeneration; optic nerve atrophy.

**Clinical:** Mental deficiency progressing to severe degrees within a few years; seizures; gargoyl e features very mild; dwarfism; stiff joints; hepatosplenomegaly; hirsutism; mitral valve insufficiency.


Savin Syndrome 1125

**General:** Congenital ichthyosis combined with urticarial manifestations.

**Ocular:** Nodular thickening in parenchyma of cornea.

**Clinical:** Dry, scaly skin; atopic dermatitis; pruritus.


Scalded Skin Syndrome (Toxic Epidermal Necrolysis; Ritter Disease; Toxic Epidermal Necrolysis of Lyell; Staphylococcal Scalded Skin Syndrome; Lyell Syndrome; Epidermolysis Acuta Toxica; Toxic Epidermal Necrosis) 1126

**General:** Generalized exfoliative dermatitis frequently affecting neonates and resulting from an initial focal staphylococcal infection (i.e., staphylococcal ophthalmia neonatorum); toxic epidermal necrolysis usually refers to manifestation in the adult secondary to a drug reaction but affects all ages; immunopathogenetic mechanisms probably initiated with drug-skin binding with aberrant immune responses, including complement and immunoglobulin G deposition with the epidermis and mucosa; recent reports suggest that patients with the acquired immunodeficiency syndrome (AIDS) are at higher risk for developing mucocutaneous reactions, such as toxic epidermal necrosis; mortality rate approximately 30%.

**Ocular:** Necrotic areas of lids, conjunctiva, and cornea; symblepharon; loss of corneal epithelium; corneal ulcer; leukemia; perforation of globe; abolition of lacrimal secretion; conjunctival chemosis; blepharitis; entropion; periorbital swelling; trichiasis; distichiasis; fornix shortening.

**Clinical:** Widespread reddening and tenderness of the skin followed by the exfoliation of large areas of skin; in children, erythema starts usually around the mouth and spreads over the entire body within hours, followed by blisters and large exudative lesions; fever; shock.

Scaphocephaly Syndrome  1127

**General:** Craniofacial dysostoses with failure in the development of the primitive mesoderm; facial features result from premature fusion of the sagittal cranial suture; males more commonly affected (4:1).

**Ocular:** Shallow orbits; proptosis; nystagmus; exotropia; aniridia; cataract; papilledema; optic atrophy; aniridia; dislocated lens.

**Clinical:** Long anteroposterior head diameter; short transverse diameter of the head; increased intracranial pressure; flat forehead with absent superciliary arches; prominent nose; mental retardation.


Schafer Syndrome (Tyrosine Transaminase Deficiency; Richer-Hanhart Syndrome; Keratosis Palmoplantaris Syndrome)  1128

**General:** Etiology unknown; dominant form manifested as an ectodermal dysplasia with disseminated follicular keratosis and leukokeratosis of the oral mucosa; recessive form also involves cornea; oculocutaneous syndrome; autosomal recessive phenotype associated with tyrosine transaminase deficiency.

**Ocular:** Dustlike, randomly distributed corneal lesions in the lower portion of the cornea; herpetoid corneal lesions; cataract; herpetiform corneal ulcers.

**Clinical:** Keratosis of palms and soles; pachyonychia; alopecia; microcephaly; dwarfism; oligophrenia; painful punctate keratoses of digits, palms and soles.


Schumann Syndrome (Besnier-Boeck-Schaumann Syndrome; Boeck Sarcoid; Sarcoidosis)  1129

**General:** Etiology unknown; theories include tuberculosis, hypersensitivity to pine pollen, virus infection; affects blacks most often; chronic course with spontaneous re- missions (see Heerfordt Syndrome); hilar or paratracheal nodes with erythema nodosum known as Lofgren syndrome; onset most often in middle and old age; ocular involvement in 20% to 25% of all cases.

**Ocular:** Orbital granulomatous mass; bony defects; cutaneous and subcutaneous nodules; myogenic palsy; lacrimal gland adenopathy; decreased tear formation; secondary glaucoma; granulomatous uveitis with iris nodules, cells, and flare; mutton fat keratitic precipitates; keratitis sicca; vitreous floaters; band-shaped keratitis; complicated cataract; inflammatory retinal exudates; "candle wax drippings"; optic nerve atrophy; neuritis; eyelid nodules; ocular nerve enlargement (granuloma).

**Clinical:** Lymphadenopathy; hilar nodes; fatigue; cystic, punched-out or reticulated changes in small bones (mainly hands and feet); muscle wasting; contractures; weakness in legs and arms.


Scheie Syndrome (Mucopolysaccharidosis IS; MPS IS; MPS V; Mucopolysaccharidosis V)

General: Autosomal recessive; chondroitin sulfate B excreted in excess in the urine; formerly MPS V (see Hurler Syndrome; Hunter Syndrome; Sanfilippo-Good Syndrome; Morquio Syndrome; Maroteaux-Lamy Syndrome). Both sexes affected; deficiency of α-L-iduronidase; increased urinary dermatan and heparan sulfate; fibrous long-spacing collagen on histopathologic examination; least severe form of mucopolysaccharidosis.

Ocular: Night blindness; fields may show general constriction; ring scotomata; diffuse corneal haze to marked corneal clouding (progressive); bushy eyebrows; coarse eyelashes; optic atrophy; anisocoria; cataracts; proptosis; acid mucopolysaccharide deposits in the iris and sclera; tapetoretinal degeneration; glaucoma.

Clinical: Normal intelligence; broad facies; thickened joints; aortic valvular disease; psychosis; claw hand; carpal tunnel syndrome; excessive body hair; progressive juxtaarticular cystic lesions.


Schiller Disease (Encephalitis Periaxialis Diffusa)

General: Lesions situated in the subcortical white matter (area 17); occurs in males; any age; etiology unknown; possibly toxic, infectious, or abiotrophic neural defects.

Ocular: Nystagmus; extraocular muscle palsy, either nuclear or supranuclear; hemianopsia (in occipital lobe involvement); optic nerve, chiasm, and tract involvement can lead to blindness; papilledema; optic neuritis; optic atrophy; central scotomata.

Clinical: Progressive spastic paralysis; progressive mental deterioration; irritability and peevishness; deafness if the temporal lobe becomes involved; tremor; dullness; characteristically bilateral lesions in the brain.


Schistosomiasis (Bilharziasis)

General: Parasitic infection by Schistosoma mansoni.

Ocular: Hyphema; parasite in anterior chamber; granuloma of conjunctiva, lid, lacrimal gland, and orbit; uveitis; keratitis; iritis; retinitis; dacyrooadenitis; cataract; posterior uveitis; choroiditis; blepharitis; optic atrophy.

Clinical: Dermatitis; fever; allergic reaction; short-lasting hepatosplenomegaly, inflammation, granuloma formation, and fibrosis; meningitis; cor pulmonale.


Schizophrenia

General: Organic brain syndrome due to degeneration or toxic, infectious, or metabolic conditions; acute or insidious onset; disturbances of thinking, mood, and behavior; etiology unknown.

Ocular: Miosis; ocular hallucinations; associated with retinitis pigmentosa in one pedigree; abnormal smooth-pursuit eye movement; visual perceptual dysfunction: low amplitude of accommodation, esophoria or exophoria, and vergenceduction suppression.
Clinical: Paranoia; depression; fear; anxiety; catatonia; delusions; hallucinations; misinterpretation of reality; lack of will or enthusiasm; hypochondriasis; preoccupation with own thoughts; meaningless repetitive speech; meaningless repetitive motions; sensorineural deafness; mental retardation associated in one pedigree.


Schomberg Disease

General: Blood dyscrasias; associated with thrombocytopenia.

Ocular: Subconjunctival hemorrhage.

Clinical: Petechiae may occur in any tissue; bleeding from any orifice.


Schomberg Syndrome (Dwarf-Cardiopathy Syndrome)

General: Consanguinity and familial occurrence; etiology obscure.

Ocular: Blepharophimosis; epicanthal folds; pseudoptosis.

Clinical: Dwarfism (proportionate); congenital heart disease.


Schwartz Syndrome

General: Glaucoma associated with retinal detachment; caused by inflammation of trabecula or pigment granules obstructing outflow; photoreceptor outer segments identified in the aqueous humor of patients with this syndrome are thought to play a role in the elevation of intraocular pressure.

Ocular: Secondary open-angle glaucoma; retinal detachment; uveitis; myopia; blepharophimosis; long eyelashes; microcornea.

Clinical: Small stature; myotonia; expressionless facies; joint limitation in hips; dystrophy of epiphyseal cartilage, vertical shortness of vertebrae, short neck; low hairline.


Matsuo N, et al. Photoreceptor outer segments in the aqueous humor of patients with this syndrome are thought to playa role in the elevation of intraocular pressure.


Sclerocornea

General: Autosomal dominant; feature of cornea plana.

Ocular: Malformation of cornea; indistinct limits of cornea and sclera.

Clinical: Found in a patient with monosomy 21; may be found in association with hypertelorism, syndactyly, ambiguous genitalia, and epidermolysis bullosa dystrophica.

Sebaceous Gland Carcinoma

General: Ocular adnexa contains various sebaceous glands from which carcinomas may arise; predilection for the upper lids but may involve both lids; usually in older age groups; slight female preponderance.

Ocular: Blepharitis; madarosis; meibomianitis; sebaceous carcinoma of lids or orbit; orbital edema; proptosis; conjunctivitis; superficial keratitis; lacrimal gland tumor.

Clinical: Metastasis to preauricular or cervical lymph nodes, or submandibular area.


Seborrheic Dermatitis

General: Genetically determined structural and functional abnormality of the skin that produces an inflammatory reaction and dryness.

Ocular: Scaling and pruritus of lids and lateral angles of eyes; chronic blepharitis; conjunctivitis; keratitis; iritis; keratomalacia in severe forms; scaling of the eyebrows and glabella; honey-colored crusting along rim of the eyelids; keratinaceous debris around eyelashes.

Clinical: Eruption consists of an erythematous scaling dermatitis with a distribution predisposed to involve the forehead at the hairline.


Seckel Syndrome (Bird-Headed Dwarf Syndrome; Virchow-Seckel Dwarfism; Ateliosis; Intrauterine Growth Retardation; Low-Birth-Weight Dwarfism; Nanocephalic Dwarfism; Primordial Dwarfism)

General: Autosomal recessive; syndrome shows variations in phenotypic appearance; both sexes affected; present at birth.

Ocular: Widely spaced eyes; incomplete eyebrow; strabismus; horizontal nystagmus; bilateral macular coloboma with pigmentation and umbilicated appearance; disk hypoplasia.

Clinical: Dwarfism; cranial deformity (bird head); developmental anomalies with short arms and clawlike hands; skeletal anomalies with narrow chest and beading of ribs; facial deformities with hypoplasia of maxilla and mandible; beaklike protrusion of central face; sparse hair; absent thumb; malformation of genitourinary tract and rectum; birdlike malformation of the face and further abnormalities; cardiac anomalies; hypophyseal hypoplasia; association with Legg-Calve Perthes disease.


Second Eye Syndrome

General: Associated with the second cataract surgery within one month of the first.

Ocular: Increased pain during the second surgery


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### Senior Syndrome (Senior-Loken Syndrome; Tubulointerstitial Nephropathy Syndrome; Loken-Senior Syndrome; Renal-Retinal Syndrome; Renal Dysplasia and Retinal Aplasia)

**General:** Autosomal recessive trait; pleiotropic gene with variable expression; death before adulthood by renal failure with uremia; both sexes affected; onset in early childhood.

**Ocular:** Visual loss progressing to complete blindness; progressive tapetoretinal degeneration that becomes manifest in early infancy; retinitis pigmentosa; ruberosis iridis; bilateral or unilateral cataracts; spherophakia; anterior lenticonus; hydropthalmos; corneal opacities; pallor of optic disk; narrowed arterioles; bone spicule formation.

**Clinical:** Nephropathy (tubulointerstitial type); growth and mental retardation; nocturia; thirst; polyuria; short stature; hearing loss; brain maldevelopment; convulsions; osteomalacia; deafness; nephronophthisis; osteomalacia; arterial hypertension; aortic insufficiency.


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### Senter Syndrome (Keratitis-Ichthyosis-Deafness Syndrome; KID Syndrome; Ichthyosiform Erythroderma, Corneal Involvement, and Deafness)

**General:** Autosomal recessive.

**Ocular:** Corneal involvement.

**Clinical:** Ichthyosiform erythroderma; deafness; hepatomegaly; hepatic cirrhosis; glycogen storage; short stature; mental retardation; hepatitis.


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### Shaken Baby Syndrome (Battered-Baby Syndrome; Battered-Child Syndrome; Child Abuse Syndrome; Silverman Syndrome)

**General:** Associated with parental abuse or accidents.

**Ocular:** Exophthalmos with orbital hemorrhages; lid hematoma; lid edema; secondary glaucoma; hyphema; vitreous hemorrhages; retinal exudates and hemorrhages (Berlin edema); choroidal atrophy; retinal detachment; papilledema; optic nerve sheath hemorrhage; preretinal, intraretinal, and subretinal hemorrhages; optic disk edema; choroidal hemorrhage.

**Clinical:** Soft tissue bruises; multiple fractures of long bones, ribs, and skull; pharyngeal bruising, subdural hematoma; seizures; failure to thrive; vomiting associated with lethargy or drowsiness; respiratory irregularities; coma or death; intracranial hemorrhage.


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### Shamberg Disease

**General:** Common benign skin disorder; self-limiting cutaneous vasculitis.
**Ocular:** Discrete foci of retinal periphlebitis associated with localized intraretinal hemorrhages and exudates; retinal and cutaneous vasculitides wax and wane concurrently; retinal vasculitis is self-limiting cause of visual disturbance and requires no therapy.

**Clinical:** Purpuric, erythematous patches appear on trunk and extremities; mononuclear perivascular infiltrate of involved skin.


**Shigellosis (Bacillary Dysentery)**

**General:** Caused by *Shigella*; frequently passed through food and via food handlers; more commonly seen in countries with poor sanitation; evidence suggests that the ability of shigellae to invade and multiply within the corneal epithelium is similar to the invasion in the intestinal epithelium.

**Ocular:** Scleroconjunctivitis; severe uveitis; conjunctival xerosis.

**Clinical:** Fever; abdominal pain; diarrhea; intestinal perforation; toxic megacolon; dehydration; there has been one case reported of an association with the Klüver-Bucy syndrome.


**Short Syndrome**

**General:** Autosomal recessive; short stature; hyperextensibility; hernia; ocular depression; Rieger anomaly; teething delay.

**Ocular:** Sunken eyes; ocular depression; Rieger syndrome; glaucoma.

**Clinical:** Short stature; hyperextensibility of joints; hernia; low birth weight; teething delay; delayed speech development; deafness; diabetes mellitus.


**Shy-Drager Syndrome (Orthostatic Hypotension Syndrome; Shy-McGee-Drager Syndrome)**

**General:** Etiology unknown; gradual onset; adults; progressive degeneration of the nervous system.

**Ocular:** External ophthalmoplegia; iris atrophy; ocular sympathetic and parasympathetic insufficiency (alternating Homer syndrome, cholinergic supersensitivity, decreased lacrimation, and corneal hypesthesia).

**Clinical:** Orthostatic hypotension; rigidity; tremor; adiadochokinesia; wasting of muscles; mental retardation; impotence; dysphagia; bilateral vocal cord paralysis; neurogenic bladder; anhydrosis; extremity weakness and paresthesias; dizziness; abnormal postural balance.


**Shy-Gonatas Syndrome**

**General:** Unknown etiology; similar to Hunter and Refsum syndromes; accumulation of lipids in muscles simulates gargoylism; present from birth.
Ocular: Mild proptosis; hypertelorism; ptosis; external ophthalmoplegia (progressive); concentric visual field constriction; keratopathy with possible corneal ulcer; lattice-like white opacities in the area of Bowman membrane; retinal pigmentary degeneration (atypical retinitis pigmentosa) with difficulties with night vision.

Clinical: Weakness of extremities (proximal); myopathy and neuropathy; cerebellar ataxia.


**Siegrist Syndrome (Pigmented Choroidal Vessels)**

**General:** Rare; more common in females (2:1); malignant hypertension; onset in advanced age.

**Ocular:** Exophthalmos; granular pigmented spots in the choroid, fairly uniform, following the course of larger choroidal vessels with extension radially toward the periphery; changes related to arteriosclerotic choroidal changes and seen following chorioretinitis of pregnancy and albuminuric choroiditis; Elschnig spots.

**Clinical:** Hypertension; albuminuria.


**Silent Sinus Syndrome**

**General:** Spontaneous enophthalmos and hypoglobus associated with a small, ipsilateral maxillary sinus.

**Ocular:** Enophthalmos; hypoglobus.

**Clinical:** Patients usually undergo painless progressive sinking of the eye.


**Silver Syndrome (Congenital Hemihypertrophy)**

**General:** Muscular hypertrophy of one side of face; etiology unknown; reported association of this condition with a small deletion in chromosome 13.

**Ocular:** Café-au-lait spots of the lid.

**Clinical:** Broad forehead; small triangular face; inverted V-shaped mouth; genitourinary abnormalities; precocious puberty; medullary sponge kidney; Wilms tumor.


Simmonds Syndrome (Hypopituitarism Syndrome)  1153

General: Anterior pituitary gland destroyed by various causes, such as hemorrhage, infarction, injuries, or postparturition infections; females; late form of Simmonds syndrome is Snapper-Witts with achlorhydria and subacute combined degeneration, and hypochromic or hyperchromic anemia; onset during postpubertal period.

Ocular: Loss of eyebrow; loss of eyelashes; central scotomata; diabetic retinopathy tends to improve after development of this syndrome; optic nerve atrophy (descending type).

Clinical: Weight loss and generalized weakness (progressive); anorexia; amenorrhea; dry skin and brittle nails; hypotension with bradycardia; anemia; psychosis; loss of libido.


Sipple Syndrome (Multiple Endocrine Neoplasia 2 or 2A; Multiple Endocrine Adenomatosis 2 or 2A; Familial Chromaffinomatosis; Multiple Neuroma; Pheochromocytoma-Thyroid Medullary Carcinoma; PCT; MEN2 or MEN2A; MEA2 or MEA2A)  1154

General: Autosomal dominant; sporadic types have been described; both sexes affected; genetic mapping has assigned the genes responsible for these tumors to the pericentromeric region of chromosome 10.

Ocular: Prominent corneal nerves (rare).

Clinical: Association of medullary thyroid carcinoma and pheochromocytoma; parathyroid tumors; neurofibromas; diabetes mellitus; diarrhea.


Sjögren Syndrome (Gougerot-Sjögren Syndrome; Secretoinhibitor Syndrome; Sicca Syndrome)  1155

General: Etiology unknown; autosomal recessive; occurs in women over age 40 years; failure of the lacrimal and conjunctival glands to maintain adequate secretion; similarities exist with Mikulicz syndrome (see Mikulicz-Radecki Syndrome); insidious onset; associated with collagen disorders; Epstein-Barr virus infection.

Ocular: Blepharoconjunctivitis; tears show no lysozyme; keratoconjunctivitis sicca; superficial corneal ulcers; thready, tenacious, yellow-white discharge of the conjunctiva; hypertrophy of lacrimal gland; decreased tear secretion with cellular and mucous debris in tear film; cicatrization of cornea and conjunctiva.

Clinical: Dryness of mouth and other mucous membranes; enlarged salivary glands; dysphagia; painless swelling of joints; polyarthrosis; dental cavities; vaginitis; laryngitis; rhinitis sicca; hepatomegaly; focal myositis; alopecia; splenomegaly.


Sjögren-Larsson Syndrome (Oligophrenia Ichthyosis Spastic Diplegia Syndrome)  1156

General: Rare; autosomal recessive; consanguinity; loss of neurons and gliosis throughout gray matter; autosomal recessively inherited disorder characterized by the triad of congenital ichthyosis, spastic diplegia or tetraplegia, and mental retardation.

Ocular: Hypertelorism; ichthyosis of lid; chorioretinitis with macular and perimacular pigment degeneration or bright, glistening intraretinal dots; atypical retinitis pigmentosa; blepharitis; conjunctivitis; keratitis; tan/white areas of retinal pigment epithelium loss; maculopathy.

Clinical: Oligophrenia idiocy; ichthyosis (congenital); spastic disorders; epilepsy; speech defect.
Skew Deviation Syndrome (Hertwig-Magendie Syndrome) 1157

**General:** Vascular deficiency involving the brainstem and middle cerebral peduncle; observed after encephalitis, tumors of the cerebellum, and lesions of the labyrinth; this is a vertical strabismus due to a supranuclear lesion; bilateral and alternating skew deviation suggests midbrain or caudal medullar lesion.

**Ocular:** Alternating paresis of elevators of one eye combined with depressor paresis of the other eye or alternating hyperphorias; in skew deviation the eyes point in diagonally opposite directions; this effect is more pronounced in the eye contralateral to the lesion.

**Clinical:** Cerebellar ataxia.

Sleep Apnea (Obstructive Sleep Apnea) 1158

**General:** Interruption of normal breathing during sleep secondary to airway obstruction; life threatening.

**Ocular:** Papilledema; optic disk edema; floppy eyelid syndrome.

**Clinical:** Obesity; hypertrophic tonsils and adenoids; excessive daytime sleepiness; snoring with periods of silence; memory loss; headache; increased intracranial pressure; personality changes; oscillopsia; pickwickian syndrome; chronic disequilibrium; increased intracranial pressure.

Slit Ventricle Syndrome 1159

**General:** Self limited episodes of shunt malfunction associated with a small or unchanged ventricular system; rare.

**Ocular:** Esotropia; nystagmus; optic atrophy

**Clinical:** Hydroencephalus; headache; nausea and vomiting; altered levels of consciousness

Sluder Syndrome (Sphenopalatine Ganglion Neuralgia Syndrome; Lower Facial Neuralgia Syndrome) 1160

**General:** Irritation of the sphenopalatine ganglion; attacks of pain last from minutes to days (see Charlin Syndrome).

**Ocular:** Severe orbital pain; increased lacrimation during episodes of pain.

**Clinical:** Unilateral facial pain, mainly root of nose, orbit, and mastoid area; episodes of headaches; nasal congestion.


Smallpox (Variola)  1161

**General:** Highly contagious cutaneous disease caused by viral infection.

**Ocular:** Conjunctivitis; keratitis; corneal ulcer; hypopyon; endophthalmitis; congenital corneal clouding; albinotic spots on iris; choroiditis; vitreous opacities; papillitis; extraocular muscle palsies; entropion; dacryocystitis; chorioretinitis; optic neuritis; and vesicles of the eyelid; preauricular adenopathy; eyelid ulcerating pustules; several conditions predispose to the spread of vaccinia, including eczema, hypogammaglobulinemia, steroid therapy, and AIDS.

**Clinical:** Fever, headache, and vomiting prior to appearance of the rash on the face, upper trunk, and down to the extremities.


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Smith Syndrome (Facio-Skeleto-Genital Dysplasia)  1162

**General:** Autosomal recessive; more common in males.

**Ocular:** Ptosis; antimongoloid slant; epicanthus.

**Clinical:** Microcephaly; high-arched palate; large, low-set ears; mental retardation; broad nose; hypoplastic mandible; pedal syndactyly.


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Smith-Lemli-Opitz Syndrome (Cerebrohepatorenal Syndrome)  1163

**General:** Autosomal recessive; similarities with trisomy 18 syndrome; prognosis poor, with death in early infancy (see Zellweger Syndrome); onset in fetal life; prevalent in males; reduced myelination in the cerebral hemispheres, cranial nerves, and peripheral nerves secondary to a defective cholesterol biosynthesis.

**Ocular:** Joining of the eyebrows (synophrys); ptosis (bilateral); pronounced epicanthal folds; strabismus; nystagmus; cataract; optic nerve demyelinization.

**Clinical:** Mental retardation; microcephaly; hypertonia; low-set ears; high-arched palate; failure to thrive; vomiting; hypospadias; cryptorchidism; metatarsus adductus.


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Smith-Magenis Syndrome (SMS)  1164

**General:** Mental retardation, physical dysmophia, and behavior abnormalities due to a deletion at chromosome 17p11.2.

**Ocular:** High myopia; retinal detachment; iris anomalies (absent collarette, nasal corectopia, stromal dysplasia); microcornea; strabismus; iris nodules called Wolfflin-Kruckmann spots.

**Clinical:** Wolfflin-Kruckmann spots may be confused with Brushfield spots, which are seen only in Down syndrome patients.


Sneddon Disease 1165

**General:** Livedo reticularis; neurologic abnormalities; labile hypertension; apparently autosomal dominant inheritance characterized by a rare potentially severe, arterioocclusive disorder; probably an immunologically mediated disorder leading to the migration and proliferation of smooth cells of small arteries, resulting in partial or complete narrowing of the vessel lumen.

**Ocular:** Central retinal artery occlusion; visual loss; optic atrophy; visual field defect; cherry-red macula.

**Clinical:** Diffuse headaches; hemihypesthesia; transient aphasic attack; hemianopsia; reticular blue discoloration of skin; hypertension; transient global amnesia; livedo reticularis; progressive neurologic deterioration; multiple ischemic cerebrovascular episodes; renal cell carcinoma.


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Snowflake Vitreoretinal Degeneration 1 1166

**General:** Autosomal dominant; very small yellow-white dots on the retina.

**Ocular:** Fibrillar vitreous degeneration; thickening of cortical vitreous; optically empty vitreous cavity; vitreous hemorrhage; posterior vitreous detachment with collapse; retinal detachment; retinal hemorrhage; retinal holes; marked retinal pigmentation; obliterated retinal vessels; sheathed retinal vessels; preretinal retraction; chorioretinal atrophy; corneal opacities; myopia; hyperopia; astigmatism; amblyopia; cataract; glaucoma.

**Clinical:** None.


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Snuff-Out Syndrome (Snuff Syndrome) 1167

**General:** Rare; sudden vision loss following ocular or laser surgery in patients with advanced glaucoma; etiology unknown but probably involves several factors, including unrecognized increased intraocular pressure, sudden hypotony, nerve injury, or retrobulbar anesthesia.

**Ocular:** Loss of central fixation; reduction in visual acuity; reduction in visual field; cataract; glaucoma.

**Clinical:** None.


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Sorsby I Syndrome (Hereditary Macular Coloboma Syndrome) 1168

**General:** Autosomal dominant; related to Laurence-Moon-Bardet-Biedl and Biemond syndromes; apical dystrophy of the extremities and bilateral macular colobomata; both sexes affected; onset from birth (see Laurence-Moon-Bardet-Biedl Syndrome; Biemond Syndrome).

**Ocular:** Hypermetropia; nystagmus; bilateral macular colobomata with various degrees of pigmentation but sharply lined borders.

**Clinical:** Distal dystrophy of the hands and feet; rudimentary or absent index fingernails; absence of big toe; cleft palate.

Sorsby II Syndrome (Sorsby Macular Dystrophy)  1169

**General:** Both sexes affected; onset in third and fourth decades of life.

**Ocular:** Retinal hemorrhages; retinal exudates; chorioretinitis; macular dystrophy.

**Clinical:** None.


Sorsby II Syndrome (Sorsby Macular Dystrophy)  1169

**General:** Both sexes affected; onset in third and fourth decades of life.

**Ocular:** Retinal hemorrhages; retinal exudates; chorioretinitis; macular dystrophy.

**Clinical:** None.


Sorsby III Syndrome (Sorsby Fundus Dystrophy)  1170

**General:** Both sexes affected; onset in fifth decade of life; autosomal dominant; mutations in the tissue inhibitor of metalloproteinases-3 have been associated with this condition; condition has been genetically linked to chromosome 22q 13-qter.

**Ocular:** Retinal hemorrhages; retinal exudates; retinal pigmentary deposits; choroidal atrophy; choroidal neovascularization; abnormal color vision; generalized fine granularity of the retinal pigment epithelium and peripheral iris transillumination.

**Clinical:** None.


Sotos Syndrome (Cerebral Gigantism)  1171

**General:** Idiopathic disturbance of the diencephalon; etiology unknown; cerebral gigantism in childhood, Russell syndrome, and total lipodystrophy are related forms of the same entity (see Russell Syndrome).

**Ocular:** Hypertelorism; antimongoloid lid aperture; high refractive error (hyperopia); nystagmus; strabismus.

**Clinical:** Acromegaly; large skull with frontal bossing; mental retardation; incoordination; abnormal excessive growth, mainly during first 2 years of life.


Spanlang-Tappeiner Syndrome (Keratosis Palmoplantaris and Corneal Dystrophy Syndrome)  1172

**General:** Autosomal dominant; etiology unknown; occurs in both sexes; onset age 5 to 20 years; linear palmar and diffuse plantar keratosis with dystrophy of the cornea.

**Ocular:** Corneal opacities (yellowish, tongue-shaped, and not always involving the center of the cornea).

**Clinical:** Hyperkeratosis of palms and soles; nail dystrophy; hyperhidrosis.


Sparganosis  1173

**General:** Infestations in humans by the plerocercoid larvae of the cestodes *Spirometra mansoni* and *Diphyllobothrium*; usually occurs in tropical countries.
Ocular: Conjunctivitis; blepharospasm; orbital cellulitis; ptosis.

Clinical: Marked eosinophilia; intraventricular hemorrhage; hydrocephalus; a case of sparganosis in the spinal canal with partial block has been reported.


Spasmus Nutans Syndrome

General: Etiology unknown; both sexes affected; onset between ages 6 and 18 months; disappears during sleep; aggravated by cold weather; spontaneous disappearance by age 3 or 4 years; it is not clear whether head nodding is a compensatory mechanism to control the nystagmus or an involuntary movement of pathologic origin.

Ocular: Bilateral nystagmus; attempt at gaze fixation intensifies manifestations.

Clinical: Rhythmic movements of head in upright position.


Spastic Paraplegia, Optic Atrophy, Dementia

General: Autosomal dominant.

Ocular: Pallor of optic disk; constricted visual fields; optic atrophy; visible deficit in retinal fiber layer; deficit in color vision; slight decrease in visual acuity; pupillary reflex sluggishness to light.

Clinical: Dementia; spastic paraparesis; stiff gait; increased deep tendon reflexes; bilateral extensor plantar responses; euphoria; pseudobulbar speech; incontinence.


Spastic Paraplegia, X-Linked (SPPX)

General: X-linked; early onset; slow progression and long survival with eventual involvement of the cerebellum, cerebral cortex, and optic nerves.

Ocular: Nystagmus; optic atrophy; poor vision; cataracts; convergent strabismus; red-green color vision defects.

Clinical: Spastic paraplegia; atethosis; mental retardation; dysarthria, ankle clonus; clubfeet; slow speech; spasticity of the legs; hyperactive reflexes; bilateral Babinski signs; toe walking; bilateral pes cavus; knee clonus; upward plantar reflexes; urinary incontinence; recurrent urinary infection; hematuria; progressive spastic gait disorder; hyperreflexia.


Spastic Quadriplegia, Retinitis Pigmentosa, Mental Retardation

General: Autosomal recessive; consanguineous parents.

Ocular: Granular pigmented retina; pale optic disk; retinal degeneration; exotropia; miotic pupils; ptosis; nystagmus; small optic disk; retinitis pigmentosa.
**Spatial Visualization Aptitude**

**General:** Sex-linked; more prevalent in males.

**Ocular:** Aptitude for visualizing space.

**Clinical:** None.

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**Sphenocavernous Syndrome**

**General:** Lesion in the cavernous sinus; similar to the superior orbital fissure syndrome (Rochon-Duvigneaud) and orbital apex syndrome (see Rochon-Duvigneaud Syndrome).

**Ocular:** Proptosis; edema; paresis of cranial nerves III, IV, and VI (paralysis of the abducens nerve precedes paralysis of the oculomotor nerve, because the abducens is situated between the internal carotid artery and the cavernous sinus wall); conjunctival edema.

**Clinical:** Paresis of the first (sometimes second and third) division of cranial nerve V; sinusitis.

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**Sphenomaxillary Fossa Syndrome (Pterygopalatine Fossa Syndrome)**

**General:** Malignant tumor, second division of which involves the sphenopalatine fossa, causing paralysis of cranial nerve V; similar to Trotter syndrome with unilateral deafness, mandibular pain, defective mobility of the palate, and trismus.

**Ocular:** Infraorbital anesthesia; optic nerve atrophy.

**Clinical:** Maxillary neuralgia with pain in the upper teeth; mandibular pain; displaced jaw toward involved side because of pterygoid muscle paralysis; deafness (middle ear, ipsilateral).

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**Spider Bites**

**General:** Venom of several different spiders can cause systemic poisoning in humans.

**Ocular:** Conjunctivitis; subconjunctival hemorrhages; conjunctival chemosis; lid edema; lid gangrene; necrosis of lid; ptosis; pupil constriction; retinal cyanosis; visual loss.

**Clinical:** Localized itching; vesicle and necrosis of tissue; secondary infection; abdominal rigidity; headache; sweating; nausea; facial congestion.

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Spina Bifida (Rachischisis) 1182

**General:** Defect of the bony spinal canal without defect of cord or meninges; myelocoele sac containing meninges may protrude; failure of neural tube of embryo to close and separate from surface ectoderm.

**Ocular:** Anophthalmos; microphthalmos; choroidal coloboma; aplasia of retinal ganglion cells and optic nerve; macular aplasia; Homer syndrome; strabismus; lateral rectus palsy; papilledema; optic nerve atrophy.

**Clinical:** Progressive motor, sensory, vasomotor, and trophic disturbances; hydrocephalus.


Spinocerebellar Atrophy with Pupillary Paralysis 1183

**General:** Autosomal dominant; rare.

**Ocular:** Absence of pupillary reaction to light or convergence.

**Clinical:** Spinocerebellar atrophy.


Spinocerebellar Degeneration and Corneal Dystrophy (Corneal Cerebellar Syndrome; Corneal Dystrophy with Spinocerebellar Degeneration) 1184

**General:** Autosomal recessive; consanguineous parents.

**Ocular:** Corneal opacification; thickened Descemet membrane; degeneration of pannus; congenital cataracts; myopia; tilted optic disks.

**Clinical:** Mental retardation; progressive cerebellar abnormalities with variable dorsal column lesions; upper motor neuron involvement; histologic muscle abnormalities.


Spongy Degeneration of the White Matter (Canavan Disease; Van Bogaert-Bertrand Syndrome) 1185

**General:** Neurologic disorder of childhood; Jews; familial; autosomal recessive; this is a severe leukodystrophy caused by the deficiency of aspartoacylase (ASPA) and accumulation of N-acetylaspartic acid; a missense mutation recently was identified in the human ASPA coding sequence from patients with this disorder.

**Ocular:** Optic atrophy; nystagmus; strabismus; roving eye movements.

**Clinical:** Progressive megalocerephaly; psychomotor deterioration; death.


Sporadic Cretinism (Congenital Hypothyroidism)

**General:** Variable, from complete lack of thyroid function to reduced function because of enzyme defects; endemic in particular areas (Crete, Beotia, Alpine Valley); affects both sexes; occurs at birth; normal physical and mental development possible with correct treatment.

**Ocular:** Nystagmus; piglike eyes.

**Clinical:** Excessive weight; lethargy; facies with heavy expression; large tongue; open mouth; drooling; yellowish tint on cheeks; hypothermia; altered tone of voice; persistent neonatal jaundice; protuberant stomach; umbilical hernia; dry skin; coarse hair; failure to thrive; poor appetite; constipation; cardiomegaly; slow pulse; delayed sexual development; dwarfism; imbecility; reported coexistence with the CHARGE association (bilateral papillary coloboma, congenital heart disease, dysmorphic ears, sensorineural deafness, psychomotor retardation, cryptorchidism, facial palsy, and vesicoureteral reflux).


Sporotrichosis

**General:** Chronic fungal infection caused by *Sporothrix schenckii*; lesion usually occurs on exposed skin and is characterized by nodules or pustules that may develop into small ulcers; infectious agent usually gains entrance into the skin by traumatic implantation of soil or plant materials; disseminated sporotrichosis is uncommon, usually occurring in alcoholics or immunosuppressed patients.

**Ocular:** Conjunctivitis; keratitis; corneal ulcer; blepharitis; endophthalmitis; iris atrophy; dacryocystitis; osteitis; periostitis; scleritis; erosion of bony walls of the orbit.

**Clinical:** Enlargement of regional lymph nodes; pulmonary lesions; granulomas in the joints and genitourinary system.


Sprengel Syndrome (High Scapula Congenita)

**General:** Etiology unknown; nonprogressive.

**Ocular:** Hypertelorism.

**Clinical:** One scapula short in vertical axis and wider in transverse and closer to the midline than the other scapula; scoliosis; torticollis; vertebral malformations.


Stanesco Syndrome (Stanesco Dysostosis Syndrome; Osteochondrosis-Osteopetrosis)

**General:** Autosomal dominant; present from birth; both sexes affected.

**Ocular:** Exophthalmos.

**Clinical:** Small stature; brachycephaly; depression at frontoparietal sutures; narrow maxilla; small mandible; crowded teeth; exostoses; fractures.


### Stannus Cerebellar Syndrome 1190

**General:** Vitamin B (riboflavin) deficiency.

**Ocular:** Nystagmus; increased lacrimation; asthenopia; blepharitis; angular conjunctivitis; iris nodules; periligamental vasodilation and pigmentation; corneal vascularization; superficial- diffuse keratitis; epithelial edema and corneal opacities; cataracts; brownish retinal patches.

**Clinical:** Muscular asthenia; hypotonia; ataxia; dysdiadochokinesia; mucocutaneous lesions resembling monilial intertrigo and glossitis.


### Staphylococcus 1191

**General:** Gram-positive coccus *Staphylococcus aureus*; most common cause of suppurative infection in humans; more common in patients with a previous disorder, such as diabetes, thyroid disease, renal failure, or malnutrition; although most *S. aureus* isolates from other sources are encapsulated, capsules have not been noted in ocular isolates.

**Ocular:** Uveitis; hypopyon; conjunctivitis; keratitis; cellulitis of lid; meibomianitis; ptosis; blepharitis; endophthalmitis; dacryocystitis; increased intraocular pressure; orbital periostitis.

**Clinical:** Tissues hypertonic, edematous, and painful; lesion liquefies, forming creamy yellow pus; fever; nausea; vomiting; cough; dyspnea; abdominal pain; diarrhea; bloody stools; dehydration; shock.


### Stargardt Disease (Juvenile Macular Degeneration) 1192

**General:** Onset between ages 8 and 14 years; variable appearance in different families.

**Ocular:** Heredomacular dystrophy; bilateral lesions showing some degree of symmetry; chorioretinal heredodegeneration; abnormal color vision.

**Clinical:** Possible association with neurologic deficits, including spastic tetraparesis and cerebellar involvement.


### Steele-Richardson-Olszewski Syndrome (Progressive Supranuclear Palsy) 1193

**General:** Nerve cell degeneration centered in the brainstem; resemblance to Lhermitte pyramidopallidal syndrome and to Jakob disease with dementia and rigidity; onset in the sixth decade of life; prominent in males.

**Ocular:** Supranuclear ophthalmoplegia affecting chiefly vertical gaze, especially downward.

**Clinical:** Pseudobulbar palsy; dysarthria; dystonic rigidity of neck and upper trunk; axial rigidity; bradykinesia; pyramidal signs; parkinsonism; frontal lobe-type dementia.


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Stevens-Johnson Syndrome (Dermatostomatitis; Erythema Multiforme Exudativum; Syndrome Mucocutaneo-Ocular; Baader Dermatostomatitis Syndrome; Mucosal-Respiratory Syndrome; Fuchs [2] Syndrome; Mucocutaneous Ocular Syndrome) 1194

**General:** Etiology unknown; affects all ages; most frequently seen in first and third decades of life; prevalent in males; drugs are the most commonly identified etiologic factor in this condition.

**Ocular:** Hypopyon; iritis; keratitis; corneal ulcers; keratoconjunctivitis sicca; chemosis; conjunctivitis; widespread fibrinoid necrosis of conjunctival vessels; blepharitis; endophthalmitis; thphisis bulbi; uveitis; cataracts; pannus; optic neuritis; keratoconus; adenoviral conjunctivitis has been reported to have precipitated Stevens-Johnson syndrome; orbital cyst may be a complication.

**Clinical:** General malaise, headaches, chills, and fever; severe skin and mucous membrane eruptions (erythema multiforme); dorsa of hands and feet are most frequently affected; rhinitis; balanitis; vulvovaginitis; urethritis (nonspecific); cystitis; patients with AIDS are at higher risk of developing Stevens-Johnson syndrome.


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Stickler Syndrome (Hereditary Progressive Arthroophthalmopathy) 1195

**General:** Autosomal dominant; onset in childhood; severe and debilitating connective tissue disorder inherited as an autosomal dominant syndrome with a variable phenotype; linkage analysis has provided statistical evidence for linkage of collagen type II (COL2A1) gene with this syndrome in some but not all families.

**Ocular:** Phthisis bulbi; glaucoma; chronic uveitis; keratopathy; complicated cataracts; chorioretinal degeneration; total retinal detachment during first decade of life; myopia; giant retinal tears.

**Clinical:** Bony enlargement of joints with abnormal development of the articular surfaces and premature degenerative changes; hypermobility of joints with abnormality in connective tissues supporting the joints; possible skeletal deformities.


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Strachan Syndrome (Tropical Nutritional Neuropathy)

**General:** Possibly nutritional vitamin A deficiency, chronic cyanide poisoning, and/or infectious agents.

**Ocular:** Optic atrophy; blurred vision, usually bilateral and symmetric; retrobulbar neuritis; scotoma; decreased vision; bilateral and symmetrical central or cecocentral scotomata; loss of color vision due to selective lesion of the maculopapillary bundles.

**Clinical:** Ataxic neuropathy; sensorineural deafness; high prevalence of goiter.


Strachan H. On a form of multiple neuritis prevalent in the West Indies. *Practitioner (London)* 1897; 59:477-484.

Straw Peter Syndrome (Slovenly Peter Syndrome; Struwwelpeter Syndrome)

**General:** Etiology unknown; combination of organic and environmental factors; affects 5% to 20% of the general school population; in preschool-aged children, manifested as clumsiness, abnormal activity level, and disorganized thought process; in school-aged children, manifested as distractibility and learning disability.

**Ocular:** Strabismus; impaired visual perception.

**Clinical:** Hyperkinesia; restlessness; fidgetiness; disorganized thought process; hypokinesia; impulsivity; poor coordination of fingers; motor awkwardness; impaired auditory perception.


Streptococcus (Scarlet Fever)

**General:** Gram-positive bacteria that can invade any tissue.

**Ocular:** Conjunctivitis; corneal ulcer; blepharitis; scarlatinial rash of lid; erysipelas dermatitis of lid; gangrene of lid; endophthalmitis; propotosis; dacryocystitis; optic neuritis; orbital cellulitis; uveitis; hypopyon; secondary glaucoma; paralysis of extraocular muscles; infectious crystalline keratopathy; scleritis.

**Clinical:** Pharyngitis; impetigo; scarlet fever; pneumonia; bacteremia; rheumatic fever; glomerulonephritis.


String Syndrome

**General:** Following encircling operations or circular diathermy, chemosis, and excess protein exudation into the anterior chamber; onset between postoperative days 4 and 19; suture causing vascular obstruction; configuration of eye is a predisposing factor.

**Ocular:** Necrosis of iris; necrosis of ciliary body; corneal ring abscess; iritis; deep anterior chamber; propotosis; lid edema; chemosis of conjunctiva; ocular hypertension; iris assumes a green color; retinal detachment.

**Clinical:** None.


Strumpell-Leichtenstern Syndrome (Acute Hemorrhagic Encephalitis)

**General:** Etiology viral, postvaccinal, drug-induced, or allergic; both sexes affected; onset at all ages but prevalent in children.

**Ocular:** Optic atrophy; acute retinal necrosis syndrome.

**Clinical:** Fever; convulsions; mental dullness; delirium; coma; ataxia; neck rigidity; tachypnea; myoclonus; aphasia; acute disseminated encephalomyelitis.

Sturge-Weber Syndrome (Meningocutaneous Syndrome; Vascular Encephalotrigeminal Syndrome; Neuro-Oculocutaneous Angiomatosis; Encephalofacial Angiomatosis; Encephalotrigeminal Syndrome) 1201

**General:** Trisomy 22 or partial trisomy inheritance. Variations include Jahnke syndrome (neuro-oculocutaneous angiomatosis without glaucoma), Schirmer syndrome (oculocutaneous angiomatosis with early glaucoma), Lawford syndrome (oculocutaneous angiomatosis with late glaucoma and no increase in volume of globe), and Mille syndrome (oculocutaneous syndrome with choroidal angioma but no glaucoma).

**Ocular:** Unilateral hydrophthalmos; secondary glaucoma (late) conjunctival angioma (telangiectases); iris decoloration; neovoid marks or vascular dilation of the episclera; glioma; serous retinal detachment; choroidal angioma; deep anterior chamber angle; port-wine stain of eyelid; buphthalmos; optic nerve cupping; anisometropia; increased corneal diameter; enophthalmos; exophthalmos; optic atrophy; choroidal hemangioma; anterior chamber angle vascularization.

**Clinical:** Vascular port-wine nevus (face, scalp, limbs, trunk, leptomeninges); acromegaly; facial hemihypertrophy; intracranial angiomatosis; convulsion; mental retardation; obesity; limb atrophy.

Subacute Bacterial Endocarditis 1202

**General:** Inflammation of the lining of the heart typically caused by Staphylococcus epidermidis.

**Ocular:** Optic neuritis; papillitis; choroiditis; conjunctival and retinal petechiae; retinal hemorrhages; Roth spots; floaters in aqueous and vitreous; rare embolic occlusion of the central retinal artery; ophthalmoplegia; papilledema.

**Clinical:** Fever; anemia; splenomegaly; heart murmur.

Subclavian Steal Syndrome 1203

**General:** Reversal of blood flow through the vertebral artery caused by stenosis of one subclavian artery proximal to the origin of the vertebral artery; arteriosclerosis; atresia of the proximal subclavian artery; aortic coarctation; subclavian artery may siphon (steal) blood from the vertebral artery; causes fluctuating symptoms of basilar artery insufficiency.

**Ocular:** Ptosis; nystagmus; temporary visual loss during activity of one arm; visual disturbances.

**Clinical:** Brachial vascular insufficiency; claudication; tingling of the arm; syncope; decrease of blood pressure in affected arm by at least 20 mm Hg; numbness; coldness; pain; facial paresthesia; headache; syncopal attacks; vertigo; intermittent claudication of the involved upper extremity.


**Submandibular, Ocular, and Rectal Pain with Flushing**

**General:** Autosomal dominant; brief, severe pain of submandibular, ocular, and rectal areas with flushing of surrounding skin.

**Ocular:** Ocular pain.

**Clinical:** Jaw aches; severe rectal pain.


**Sulfite Oxidase Deficiency**

**General:** Rare abnormality of sulfur metabolism in which there is an accumulation of S-sulfocysteine, thiosulfate, and sulfate and a deficiency of sulfite; present at birth; death occurs before age 3 years.

**Ocular:** Dislocated lens; nystagmus; ectopia lentis.

**Clinical:** Muscular rigidity; eccentric behavior; coarse face; broad nasal bridge; long philtrum (in association with Leigh syndrome).


**Sunrise Syndrome**

**General:** Occurs when the intraocular lens (IOL) optic is displaced superiorly out of the visual axis.

**Ocular:** Edge of IOL in pupil; decreased visual acuity; glare.

**Clinical:** None.


**Sunset Syndrome**

**General:** Occurs when the capsule or zonules have been sufficiently damaged to allow the posterior chamber IOL to slip gradually into the inferior vitreous in the postoperative period.

**Ocular:** Superior edge of IOL in pupil; IOL in vitreous body; retinal detachment; decreased visual acuity; glare; zonular disinsertions.

**Clinical:** None.

Superior Vena Cava Syndrome (Vena Cava Superior Syndrome)  1208

**General:** Compression or obstruction of the superior vena cava by aortic aneurysms, mediastinal neoplasms, thyroid adenoma, or carcinoma of the lung; found in males in their 50s.

**Ocular:** Glaucoma; conjunctival vasodilation; retinal hemorrhages (Valsalva retinopathy); eyelid edema; optic disk edema; engorgement of the conjunctival and episcleral vessels; periorbital edema.

**Clinical:** Cyanosis and edema of face, neck, and upper trunk with a rather sharp demarcation line (short cape edema); dysphagia; epistaxis; hoarseness; vertigo; tinnitus; may be caused by Graves disease and as a presentation of Behçet disease.


Suprarenal Sympathetic Syndrome (Pheochromocytoma Syndrome; Adrenal Sympathetic Syndrome; Adrenal Medulla Tumor Syndrome)  1209

**General:** Tumors producing increased secretion of norepinephrine and epinephrine deriving from chromaffin cells of the adrenal medulla; more common in males (3:2); symptoms occur in paroxysms or attacks; precipitated by emotional upsets; predilection for the right adrenal.

**Ocular:** Spasm of retinal arteries with associated cotton-wool exudates; flame-shaped hemorrhages; papilledema; pupillary dilation; neovascularization of retina; following removal of the tumor there may be macular scarring and optic atrophy.

**Clinical:** Hypertension; tachycardia; severe anxiety; headache; nervous tension; sweating; pallor; nausea; polyuria; polydipsia; association with neurofibromatosis and von Hippel disease.


Susac Syndrome  1210

**General:** Rare; unknown origin; characterized by the triad of encephalopathy, fluctuation hearing loss and visual loss resulting from microangiopathy of the brain, cochlea and retina.

**Ocular:** Cotton wool spot; central retinal vein occlusion;

**Clinical:** Hearing loss; encephalopathy


Sweet Syndrome (Acute Febrile Neutrophilic Dermatosis)  1211

**General:** Cause unknown; common in middle-aged women; associated with acute leukemia; acute febrile neutrophilic dermatosis; neurologic symptoms.

**Ocular:** Conjunctivitis; episcleritis; glaucoma; limbal nodules.

**Clinical:** Cutaneous eruption with fever; nondeforming, asymmetrical, large-joint arthritis; albuminuria; anemia; oral aphthae; genital ulcers; association with Behçet disease has been reported.

**Sylvestre Disease**

**General:** Dominant inheritance.

**Ocular:** Optic atrophy.

**Clinical:** Ataxia; moderate and slowly progressive hearing loss.


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**Symonds Syndrome (Otitic Hydrocephalus Syndrome; Serous Meningitis Syndrome; Benign Intracranial Hypertension; Pseudotumor Cerebri)**

**General:** Children and adolescents; protracted course; increased cerebrospinal fluid, but without increase in protein or cells.

**Ocular:** Sixth nerve palsy, ipsilateral side with otitis media; retinal hemorrhages and exudates; moderate-to-marked papilledema followed by secondary optic atrophy; unilateral or bilateral swelling of the optic nerve head have been reported; cranial nerve III and IV involvement; bilateral retinal vein occlusion.

**Clinical:** Greatly increased pressure of spinal fluid, often greater than 300 mm, without increased cells or protein; intermittent headaches; otitis media; chronic renal failure; chronic myeloid leukemia.


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**Sympathetic Ophthalmia**

**General:** Trauma or injury to one eye and later onset of inflammation in the other eye.

**Ocular:** Iridocyclitis (acute inflammation of iris, ciliary body, and anterior chamber); choroiditis; chronic persistent keratitic precipitates; posterior synechiae; phthisis bulbi; has been reported following laser cyclocoagulation.

**Clinical:** None.


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**Takayasu Syndrome (Martorell Syndrome; Aortic Arch Syndrome; Pulseless Disease; Reversed Coarctation Syndrome)**

**General:** Two types are (i) occlusive inflammatory lesion (seen in young Japanese women) and (ii) occlusive vascular disease without inflammation, associated with atherosclerosis and syphilis; onset in fifth and sixth decades; both sexes affected; can involve the aorta and its major branches as well as the coronary, hepatic, mesenteric, pulmonary, and renal arteries.

**Ocular:** Iris atrophy; cataracts; retinal microaneurysms; sausage-shaped venous dilations; reduced central retinal artery pressure; optic atrophy; cotton-wool spots; anterior segment ischemia; retinal arteriovenous shunts.

**Clinical:** Diminished or absent pulsation of arteries (head, neck, upper limbs); orthostatic syncope; facial atrophy; epileptiform seizures; intermittent claudication.

**Tangier Syndrome (Lipoprotein Deficiency; Familial High-Density Lipoprotein Deficiency; \(\alpha\)-Lipoprotein Deficiency; Fish Eye Disease)**

**General:** First seen on Chesapeake Bay Island; rare; autosomal recessive; inability to synthesize polypeptide required in the elaboration of high-density lipoprotein; cholesteryl esters are stored; occurs in both sexes; onset from childhood to fifth decade of life; disorder appears to be a variant of familial lecithin-cholesterol acyltransferase (LCAT) deficiency in which the enzyme remains partly active.

**Ocular:** Corneal infiltrates; fine, dotted stromal opacities, most marked in posterior central third of corneal stroma; wasting of orbicularis oculi muscle.

**Clinical:** Maculopapular rash; orange-yellow striped tonsils; hepatosplenomegaly; lymphadenopathy; intermittent diarrhea; bilateral motor weakness.


**Tapetal-Like Reflex Syndrome**

**General:** Rare.

**Ocular:** Ring scotoma; discrete bright yellow spots in posterior polar region deep to the retinal vessels; tapetal-like reflex and retinitis pigmentosa may be present in members of the same family.

**Clinical:** None.


**TAR Syndrome (Thrombocytopenia Absent Radius Syndrome)**

**General:** Bilateral absence of the radius and hypomegakaryocytic thrombocytopenia.

**Ocular:** May have cataracts, glaucoma, megalocornea, and blue sclera.

**Clinical:** Patients have foreshortened forearms and radically deviated hands; infrequently associated with mental retardation (7%); also may have lower extremity deformity.


**Tay-Sachs Syndrome (Norman-Wood Syndrome; Familial Amaurotic Idiocy; Gangliosidosis GM2 Type 1; Hexosaminidase Deficiency)**

**General:** Similar to ceroid lipofuscinosis; autosomal recessive; occasional dominant inheritance; onset birth to 10 months; affects Jewish females; death during first 2 years; stored ganglioside (see Cereoid Lipofuscinosis); decreased hexosaminidase A localized to chromosome 15 (15q22-15q25.1).

**Ocular:** Nystagmus; strabismus; whitish-gray macular area with cherry-red spot in the center; retinal pigmented changes with involvement of the macula occasionally may be seen instead of the typical red spot; grayish coloration of the macula is due to
the swollen ganglion cells in the perifoveal macular region; retinal vessels become narrowed; progressive ascending optic atrophy; cortical blindness by age 12 to 18 months with reactive pupils; deterioration of ocular motor function.

**Clinical:** Hyperacusis; mental retardation; convulsions; muscles, initially flaccid, becoming spastic with progression; infants normal at birth but fail to thrive after 4 to 8 months; hypotonia; death; occurs primarily in Jewish children; biochemical heterogeneity; absence of hexosaminidase A most common (type 1); absence of hexosaminidase A and B in Sandhoff variant (type 2); feeding difficulties; doll-like facies; fine hair; macrocephaly; abnormal acoustico-motor reaction.


**Temporal Arteritis Syndrome (Cranial Arteritis Syndrome; Giant Cell Arteritis; Hutchinson-Horton-Magath-Brown Syndrome)**

**General:** Etiology unknown; mainly females; mainly whites; ages 55 to 80 years; temporal artery shows inflammatory thickening; arteritis of the vessels supplying the optic nerve.

**Ocular:** Transient ptosis; partial or complete loss of vision on the affected side; retinal detachment; exudates and hemorrhages; narrowing of retinal vessels; obstruction of the central retinal artery; optic atrophy; ischemic optic neuropathy; acute decreased intraocular pressure; corneal hypesthesia; palsies of extraocular muscles; hemorrhagic glaucoma; diplopia; hemorrhages on or around the disk.

**Clinical:** Throbbing headache; hyperalgesia of the scalp; malaise; anorexia; weakness; weight loss; fever; nodular pulmonary nodules; cough; otitis with deafness.


**10q-Syndrome (10q Deletion Syndrome)**

**General:** Chromosome 10q deletion syndrome.

**Ocular:** Microphthalmia.

**Clinical:** Intrauterine growth retardation; microcephaly; truncus arteriosus type I; respiratory distress; craniofacial dysmorphism.


**Terrien Disease (Terrien Marginal Degeneration; Gutter Dystrophy; Peripheral Furrow Keratitis; Senile Marginal Atrophy)**

**General:** Rare; no known cause; 75% of patients are males from age 10 to 70 years.

**Ocular:** Usually bilateral; may be asymmetrical; peripheral, fine, yellow-white, punctate stromal opacities associated with mild, superficial corneal vascularization; progressive thinning leads to peripheral gutter formation; decrease in visual acuity; loss of Bowman membrane and anterior stromal lamella with partial replacement of these tissues by a vascularized connective tissue; fatty deposits; thin stroma; thickness changes in Descemet membrane; regular recurring attacks of pain and inflammation; keratoconus; reported association with Terrien marginal degeneration.
Clinical: None.


Terson Syndrome (Subarachnoid Hemorrhage Syndrome)

General: Spontaneous rupture of aneurysm or traumatic intracerebral hemorrhage; onset at all ages.

Ocular: Weakness of extraocular muscles; disarranged and uncoordinated gaze; severe intraocular hemorrhage; preretinal hemorrhages; peripapillary hemorrhages; papilledema secondary to optic nerve sheath hemorrhages; pigmentary changes in macula and retina; preretinal membrane formation; vitreous detachment; amblyopia; anisocoria; bilateral retinal detachments have been associated with this disorder; epiretinal membranes (sequelae).

Clinical: Sudden unconsciousness; elevated cerebrospinal fluid pressure.


Thalasselas Syndrome

General: Keratoconus-tetany-menopause.

Ocular: Spontaneous keratoconus.

Clinical: Syndrome describes an association between hormone deficiency, magnesium deficiency, allergy, keratoconus, and type B behavior.


Thanatophoric Dwarfism

General: Etiology unknown, but autosomal recessive inheritance is suspected; seen most frequently in males; onset in fetal life; death usually within first 3 days of life; possible association with drug administration has been reported.

Ocular: Exophthalmos.

Clinical: Head enlarged; small face; enlarged fontanelles; high forehead; frontal bossing; saddle nose; abdomen protuberant; thorax narrow; short rib; hypotonia; respiratory distress; cardiac failure; marked shortening of the extremities; macrocephaly.


Thelaziasis

General: Ocular infection caused mainly by the nematode Thelazia callipardon; natural habitat is the lacrimal gland of the dog.

Ocular: Uveitis; parasites in the conjunctiva, cornea, lacrimal system, and anterior chamber; corneal opacity; corneal abrasion; increased tear secretion.

Clinical: None.


Thermal Burns

**General:** May occur to any body tissue.

**Ocular:** Conjunctival necrosis; corneal ulcer; exposure keratitis; ectropion; contracture deformity of lids; lid edema; entropion; endophthalmitis; proptosis; dacryocystitis; chronic epiphora; cellulitis; corneal perforation; symblepharon.

**Clinical:** Burns of any body tissue; edema; contractures; secondary infections.


13q- Syndrome (13q Deletion Syndrome)

**General:** Chromosome 13q deletion syndrome.

**Ocular:** Retinoblastoma; telecanthus; hypertelorism; optic nerve hypoplasia; retinal dysplasia.

**Clinical:** Holoprosencephaly; abnormal lower extremity configuration; atrial septal defect; microcephaly; ambiguous genitalia; hypotonia; low-set ears; growth retardation; mild mental retardation; intestinal atresia.


Thompson Syndrome

**General:** Autosomal dominant inheritance.

**Ocular:** Congenital optic atrophy; nystagmus; blindness.

**Clinical:** None.


Thomsen Syndrome (Congenital Myotonia Syndrome; Myotonia Congenita)

**General:** Dominant; inheritance manifestations before age 5 years; prevalent in males; possibly caused by excessive production of acetylcholine neuromuscular junction; emotions and cold enhance symptoms; warmth decreases symptoms; there are two types of this disorder, an autosomal dominant and an autosomal recessive, both with the same Clinical features; has been linked to chromosome 7q35 in the region of the human skeletal muscle chloride channel gene (HUMCLC).

**Ocular:** Inability to open eyelids for a few seconds after closure; spasm of the orbicularis oculi muscle; extraocular muscle paresis.

**Clinical:** Myotonia with muscles of upper and lower extremities primarily affected; muscle hypertrophy; pronounced delay in relaxation of contracted voluntary muscles.


Thomsen AJT. Myotonia Congenita; Tonische Krampfe in Willkürlich Beweglichen Muskeln in Folge von Ererbter Physischer Disposition (Ataxia Muscularis?). *Arch Psych* 1876; 6:702.
### 3B Translocation Syndrome

**General:** Chromosomal anomaly transmitted by the female but not the male carrier.

**Ocular:** Iris coloboma; corneal opacity; proptosis; strabismus.

**Clinical:** Low birth weight; micrognathia; small ears; cleft lip and palate; cardiac defects; ventricular septal defect; atrial septal defect; absent ductus arteriosus; pulmonary arterial diverticulum; right aortic arch; absent pulmonic valve.


### 3p-Syndrome (3p Deletion Syndrome)

**General:** Chromosome 3p deletion syndrome.

**Ocular:** Blepharoptosis, telecanthus, mongoloid (down-slanting) palpebral fissures.

**Clinical:** Mental retardation, profound growth failure, characteristic facies, low birth weight, trigonocephaly, psychomotor delay, micrognathia.


### Thrombocytopenia

**General:** Decrease in platelets.

**Ocular:** Retinal hemorrhages; papilledema; visual field defects; oculomotor nerve palsy; optic nerve atrophy.

**Clinical:** Anemia; cranial nerve palsies; thrombotic thrombocytopenic purpura; intracranial hemorrhage; neuroophthalmologic signs and symptoms.


### Thygeson Syndrome (Keratitis Superficialis Punctata)

**General:** Etiology probably of viral origin; recurrence every 3 to 4 years.

**Ocular:** Punctate lesions of cornea; keratitis.

**Clinical:** None.


### Thyrocerebroretinal Syndrome (Familial Thyrocerebral Retinal Syndrome)

**General:** Autosomal recessive; renal, neurologic, and thyroid disease.

**Ocular:** Retinal hemorrhages; central vision defect; retinal edema; optic atrophy.

**Clinical:** Thrombocytopenia; chronic renal disease; colloid goiter.


### Tic Douloureux (Trigeminal Neuralgia)

**General:** Brief, sharp, unilateral facial pain that usually occurs in the middle or lower face; occurs more often in females; occurs most frequently in persons over age 40 years; right side affected more than left side.
Ocular: Ipsilateral hyperemia with the pain of conjunctiva; periorbital pain; ipsilateral lacrimation during the pain; decreased corneal sensitivity; photophobia.

Clinical: Pain triggered by chewing, swallowing, laughing, brushing teeth, shaving, or combing hair; may be present with multiple sclerosis.


### Tilted Disk Syndrome

**General:** Relatively common congenital anomaly (1% to 2% of the population) consisting of inferonasal "tilting" of the disk, with the upper and temporal portion of the disk laying anterior to the inferonasal portion.

**Ocular:** "Tilting" of the disk with associated findings of an obliquely directed long axis of the disk, inferonasal crescent, posterior staphyloma of the affected inferonasal region of the fundus, and upper and temporal emergence of the retinal vessels rather than the nasal (situs inversus); lenticular astigmatism.

**Clinical:** Patients tend to have myopic astigmatism and superotemporal or bitemporal visual field depression; patients are at increased risk for retinal serous detachment.


### Tolosa-Hunt Syndrome (Painful Ophthalmoplegia)

**General:** Symptoms last from days to weeks; attacks recur at intervals of months or years; inflammatory lesion of cavernous sinus; onset most frequent in fifth decade of life; recurrent Tolosa-Hunt syndrome has been observed in some patients.

**Ocular:** Steadily "growing" retroorbital pain; ptosis; involvement of cranial nerves III, IV, VI, and first division of V; scintillating scotoma; sluggish pupil reaction to light; corneal sensitivity diminished; optic neuritis.

**Clinical:** Inflammatory lesions of cavernous sinus.


Tolosa E. Periarteritic lesions of carotid siphon with *Clinical* features of carotid infraciliary aneurysm. *J Neurol Neurosurg Psychiatry* 1954; 17:300.

### Toluene Abuse

**General:** Caused by inhalation of toluene and other toluene-containing substances, such as glue.

**Ocular:** Jerking movements of the eyes; bilateral optic neuropathy.

**Clinical:** Progressive tremor of the limbs, trunk, and head; dizziness; slurred speech; bilateral hearing loss; mild impairment of memory and concentration; reduced magnetic resonance signal intensity in the brain; distal renal tubular acidosis.


Topless Optic Disk Syndrome  1240

**General**: Superior segmental optic hypoplasia.

**Ocular**: Relative superior entrance of the central retinal artery; thinning of the superior peripapillary nerve fiber layer; superior peripapillary scleral halo; pallor of the superior disk.

**Clinical**: Patients usually have good visual acuity and an inferior altitudinal or sector-like field defect; all reported patients either are children of mothers with type I diabetes mellitus or are Japanese.


Torre Syndrome  1241

**General**: Multiple sebaceous gland tumors of nonglabrous skin and visceral malignancy (primarily colonic).

**Ocular**: Sebaceous cell carcinoma of lid; carcinoma of caruncle; tumor of the meibomian gland.

**Clinical**: Visceral carcinoma of gastrointestinal tract, breast, and prostate.

Torre D. Multiple sebaceous tumors. *Arch Dermatol* 1968; 98:549.

Torre-Muir Syndrome  1242

**General**: Multiple sebaceous gland tumors of nonglabrous skin and visceral malignancy (primarily colonic) and keratoacanthomas; like Torre syndrome but associated with keratoacanthomas; benign skin tumors accompany and sometimes precede development of internal visceral malignancy; autosomal dominant inheritance.

**Ocular**: Sebaceous cell carcinoma of lid; carcinoma of caruncle; keratoacanthomas; tumor of the meibomian gland.

**Clinical**: Visceral carcinoma of gastrointestinal tract, breast, and prostate.

Torre D. Multiple sebaceous tumors. *Arch Dermatol* 1968; 98:549.

Touraine-Solente-Gole Syndrome (Pachydermoperiostosis; Acropachyderma; Audry II Syndrome; Brugsch Syndrome; Friedrich-Erb-Arnold Syndrome; Héhlinger Syndrome)  1243

**General**: Rare; hereditary; predominant in males; onset in puberty to third decade.

**Ocular**: Elephantiasis of the lids caused by meibomian gland cysts and connective tissue hypertrophy; ptosis.

**Clinical**: Thick and furrowed skin of forehead, face, scalp, hands, and feet; hyperhidrosis of hands and feet; increased subcutaneous secretion; enormous hands and feet; watch crystal-like nails; cylindrical arms and legs; effusions of ankles, knees, and other joints; finger clubbing; facial enlargement; periostitis; cutaneous mucinosis.

Tourette Syndrome (Gilles de la Tourette Syndrome; Brissaud II Syndrome; Caprolalia Generalized Tic; Guinon Myospasia Impulsiva)

**General:** Etiology unknown; occurs at ages 7 to 8 years; emotional trauma is frequent precipitating factor; disturbed parent-child relationship frequently encountered.

**Ocular:** Blepharospasm; oculogyric deviations; dystonic neck movements.

**Clinical:** Chorea; caprolalia; echolalia tic; blinking and facial twitching.


Toxic Lens Syndrome (Toxic Anterior Segment Syndrome; TASS)

**General:** Syndrome occurs within a few days to several weeks of implantation of an intraocular lens; with therapy, vision is restored in the majority of cases; increased incidence of disease caused by use of ethylene oxide sterilization (dry pack intraocular lenses); toxic lens syndrome may be prevented by treating the lens with sodium hydroxide and by using modern lathe-cut or compression-molded lenses with polypropylene loops; risk factors include uveitis in history, pseudoexfoliation syndrome, inadequate mydriasis at the start of surgery, problems with intraocular lens implantation, and pigment effusion during surgery.

**Ocular:** Pigment precipitation on the surface of an intraocular lens; hypopyon; vitreous opacification; chronic uveitis; secondary glaucoma.

**Clinical:** None.


Toxic Shock Syndrome

**General:** Multisystem illness caused by toxin-producing *Staphylococcus aureus* infection; occurs in all sexes and ages but most frequently in women; seen in association with tampon use; onset may be sudden.

**Ocular:** Eyelid necrosis; periorbital abscess; neonatal conjunctivitis; conjunctival hyperemia; preseptal cellulitis.

**Clinical:** Fever; rash; desquamation; hypotension; syncope; dizziness; shock; cardiovascular collapse; coma; death.


Trachoma

**General:** Most common in rural communities of the Middle East, Africa, Asia, and South and Central America; caused by *Chlamydia trachomatis*; associated with poor sanitation and medical care.

**Ocular:** Chronic keratoconjunctivitis; papillae follicles; keratitis; opacities of cornea; scars of palpebral conjunctiva; ptosis; tearing; entropion.

**Clinical:** Rhinitis; otitis media; upper respiratory tract infection.


### Transient Light Sensitivity Syndrome

**General:** Associated with femtosecond laser keratome; may be related to the pulse energy used in flap creation

**Ocular:** Transient postoperative photosensitivity


### Traumatic Encephalopathy Syndrome (Posttraumatic General Cerebral Syndrome; Postconcussion Syndrome; Punch-Drunk Syndrome)

**General:** Small focal hemorrhages within the cerebrum and/or cerebellum causing functional brain damage; minor traumatic brain injury is the most common type of traumatic encephalopathy.

**Ocular:** Nystagmus or nystagmoid ocular movements.

**Clinical:** Personality change; rigid face without expression; staggering gait; dysphonia.


### Treft Syndrome

**General:** Autosomal dominant; usually appears by age 11 years.

**Ocular:** Optic atrophy; visual loss; ptosis; ophthalmoplegia.

**Clinical:** Hearing loss by age 14 years; myopathic changes; balance difficulty.


### Triangular Syndrome

**General:** Rare; follows trauma of excessive compression during surgery, such as Faden operation.

**Ocular:** Choroidal ischemia; retinal tear; sectorial infarction of choroid; occlusion of posterior ciliary vessel with triangular chorioretinal scar.

**Clinical:** None.


### Trichinellosis (Trichinosis)

**General:** Parasite *Trichinella* enters the body by ingestion of infected meat (usually poorly cooked pork).

**Ocular:** Conjunctivitis; splinter hemorrhages of conjunctiva; paralysis of sixth nerve; exophthalmos; proptosis; uveitis; optic neuritis; papilledema; retinal hemorrhages; dyschromatopsia; scotoma; secondary glaucoma; encysted parasites in the extraocular muscles.
Clinical: Fever; urticaria; respiratory symptoms; muscle pain; myalgias and severe proximal muscle weakness; impaired coordination.


Trichomegaly with Mental Retardation, Dwarfism, and Pigmentary Degeneration of the Retina (Oliver McFarlane Syndrome) 1253

General: Autosomal recessive.

Ocular: Excessive growth of eyelashes and brow hair; pigmentary degeneration of retina; horizontal nystagmus; bilateral choroiodoretinal pigmentary degeneration; ring heterochromia of the iris.

Clinical: Bulging of occipital and frontal bones; low birth weight; dwarfism; cryptorchidism; underdevelopment of penis; frontal alopecia.


Triploidy Syndrome 1254

General: Extra set of chromosomes due to diandry or digyny; stillbirth or early neonatal death.

Ocular: Iris coloboma; microphthalmia; hypertelorism.

Clinical: Large placenta; prenatal growth deficits; large fontanelles; syndactyly; heart defects; cleft lip; genital, brain, ear, and kidney malformations; meningomyelocele; micrognathia.

Kaufman MH. New insights into triploidy and tetraploidy, from an analysis of model systems for these conditions. Hum Reprod 1991; 6:8-16.

Trisomy 8 Mosaicism Syndrome 1255

General: Chromosomally abnormal cell line with each cell containing an extra chromosome 8; other cell lines normal; both sexes affected; present from birth.

Ocular: Strabismus; hypertelorism; deep-set eyes.

Clinical: Mild-to-moderate mental retardation; low-set or malformed ears; broad, bulbous nose; palatal deformity; congenital cardiovascular disorders; hydronephrosis; cryptorchidism; poor coordination; prominent forehead; enlarged nares; full lips; cupped ears; camptodactyly of fingers and toes; reported as a nonrandom secondary change in myxoid liposarcoma.


Trisomy 18 Syndrome (E Syndrome; Edwards Syndrome) 1256

General: Chromosome 18 present in triplicate; more common in females (3:1); age of mother over 40 years; onset from fetal life.
**Ocular:** Unilateral ptosis; epicanthal folds; congenital glaucoma; corneal opacities; lens opacities; optic atrophy.

**Clinical:** Low-set ears; micrognathia; high-arched palate; prominent occiput; cryptorchidism; failure to thrive; ventricular septal defect; hypertonicity with rigidity in flexion of limbs; mental retardation; umbilical and inguinal hernias.


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### Trisomy 9q Syndrome

**General:** Congenital mental retardation syndrome due to 9p trisomy.

**Ocular:** Hypertelorism; deep-set eyes; antimongoloid (up-slanting) eyes.

**Clinical:** Mental retardation; short stature; down-turned corners of the mouth; slightly or moderately bulbous nose; moderately large ears; nail dysplasia and hypoplasias; clinodactyly; abnormal dermatoglyphs.


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### Trisomy 17p Syndrome

**General:** Trisomy 17p duplication syndrome.

**Ocular:** Hypertelorism, antimongoloid (up-slanting) palpebral fissures.

**Clinical:** Growth retardation; microcephaly; long philtrum with a thin upper lip; micrognathia; high-arched palate.


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### Trisomy 6p Syndrome (6p+ Syndrome, 6p Duplication Syndrome)

**General:** Chromosome 6p trisomy syndrome.

**Ocular:** Blepharochalasis.

**Clinical:** Low birth weight; psychomotor retardation; craniofacial abnormalities (prominent forehead, large fontanelle, wide sagittal suture, low-set and/or malformed ears); congenital heart malformation; small kidneys; proteinuria.


Rosi G, et al. Trisomy 6p22 leads to 6pter due to familial t(6;13) (p22; q34 or 33) translocation. *Hum Genet* 1979; 51:67-72.

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### Trisomy 6q Syndrome (Duplication 6q+6q Syndrome)

**General:** Chromosome 6q trisomy syndrome.

**Ocular:** Hypertelorism; mongoloid (down-slanting) palpebral fissures.

**Clinical:** Cleft soft palate; bow-shaped mouth; micrognathia; short, laterally webbed neck; microcephaly; clubbing of hands and feet; syndactyly; growth retardation; mental retardation; "carp" mouth.


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### Trisomy 10q Syndrome (10q+ Syndrome)

**General:** Chromosome 10q trisomy (duplication) syndrome.

**Ocular:** Microphthalmia; deep-set eyes; epicanthus; bilateral, enlarged, gray optic disks; distended retinal vessels; bilateral punctate yellow deposits near the macula and optic disk.
**Clinical:** Mental retardation; microcephaly; prominent forehead; upturned nose; bow shaped mouth; micrognathia; thick and flat helices of the ears; long slender limbs.


**Trisomy 13 Syndrome (Trisomy D1 Syndrome, Patau Syndrome, Reese Syndrome)**

**General:** Extra chromosome in the D group; fatal in the first few months of life; trisomy 13-15 resembles trisomy D1.

**Ocular:** Anophthalmia; microphthalmia; iris coloboma; cataracts; retinal dysplasia; optic nerve coloboma; optic atrophy; iris dysplasia; calcified lens; retinal detachment; optic nerve hypoplasia; orbital cysts.

**Clinical:** Apneic spells; developmental deficiency of the nervous system; seizures (minor motor); deafness; cleft lip and palate; hemangiomata; horizontal palmar creases; interventricular septal defects; renal abnormalities; cardiovascular changes; respiratory involvement; gastrointestinal disease; urogenital involvement; cerebral hypoplasia with hydrocephalus; mental retardation.


**Trisomy 20 Syndrome**

**General:** Trisomy 20q (duplication) syndrome.

**Ocular:** Oblique palpebral fissures, strabismus.

**Clinical:** Round face; cardiac and vertebral abnormalities; mild psychomotor retardation with poor coordination and speech impediment; anencephaly.


**Trisomy 21q- Syndrome (21q Deletion Syndrome)**

**General:** Chromosome 21q deletion syndrome.

**Ocular:** Blepharochalasis; microphthalmia with persistent hypoplastic primary vitreous.

**Clinical:** Mental and physical retardation; generalized hypertonia; high nasal bridge; micrognathia; malformed ears with preauricular pits, and overlying fingers.


**Trisomy 2q Syndrome (q33-qter)**

**General:** Associated with monosomy 9p (p24-pter); autosomal recessive or X-linked inheritance.

**Ocular:** Congenital glaucoma; hypertelorism; epicanthus.

**Clinical:** Low-set and malformed ears; short saddle nose with antverted nostrils; long, hypoplastic philtrum; thin upper lip; hypospadias; short fingers; muscular hypotonia; psychomotor retardation; clinodactyly; scoliosis; broad, flat nasal bridge; short neck; short esophagus; tubular stomach.


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Trisomy 22 Syndrome

**General:** Trisomy for chromosome 21/22; trisomy 22 may be very mild form of Down syndrome (trisomy 21).

**Ocular:** High myopia.

**Clinical:** Schizophrenia; micrognathia; large nostrils; flat occiput; hyperextension of elbows; macrocephaly; hydrocephalus; holoprosencephaly; facioauriculovertebral (Goldenhar) sequence.


Trisomy D1 Syndrome (Trisomy 13; Patau Syndrome; Reese Syndrome)

**General:** Extra chromosome in the D group; fatal in first few months of life; trisomy 13-15 resembles trisomy D1.

**Ocular:** Anophthalmia; microphthalmia; iris coloboma; cataracts; optic nerve coloboma; optic atrophy; iris dysplasia; calcified lens; retinal detachment; optic nerve hypoplasia; orbital cysts.

**Clinical:** Apneic spells; developmental deficiency of nervous system; seizures (minor motor); deafness; cleft lip and palate; hemangiomata (capillary type); horizontal palmar creases; hyperconvex fingernails; interventricular septal defects; renal abnormalities; cardiovascular changes; respiratory involvement; gastrointestinal disease; urogenital involvement; cerebral hypoplasia with hydrocephalus; mental retardation.


Tristichiasis

**General:** Autosomal dominant.

**Ocular:** Three rows of eyelashes.

**Clinical:** None.


Tritianomalous Color Blindness (Color Blindness, Partial Tritanomaly)

**General:** Sex-linked; rare; lacking blue and yellow sensory mechanisms while retaining those for red and green.

**Ocular:** Blue-yellow color blindness.

**Clinical:** None.


Tritanopia (Color Blindness; Blue Color Blindness)

**General:** Autosomal dominant; more common in males; defective blue color vision is characteristic; two amino acid substitutions in the gene encoding the blue-sensitive opsin have been detected.

**Ocular:** Lacking blue and yellow sensory mechanisms while retaining those for red and green; optic atrophy.
Clinical: None.


Tropical Pancreatic Diabetes (TPD) 1271

**General:** Secondary diabetes as a result of chronic calcific pancreatitis; limited geographically to a few tropical countries; highest prevalence in southern India; male predominance; onset at young age; associated with protein calorie malnutrition; possible cause is cassava ingestion; malnutrition has been postulated as a possible etiology.

**Ocular:** Background retinopathy; proliferative retinopathy; fibrous retinitis proliferans; microaneurysms; macular edema; hemorrhages; exudates; decreased visual acuity.

**Clinical:** Chronic pancreatitis; recurrent abdominal pain; steatorrhea.


Tuberculosis 1272

**General:** Communicable disease caused by the acid-fast bacillus *Mycobacterium tuberculosis.*

**Ocular:** Conjunctivitis; subconjunctival nodules (tuberculomas); keratitis; pannus; corneal ulcer; blepharitis; cellullitis; meibomianitis; uveitis; dacryocystitis; chronic orbital cellullitis; retinitis; scleritis; scleral perforation; hypopyon; vitreous hemorrhages; optic neuritis; optic atrophy; tuberculous panophthalmitis; choroidal tubercles; intraorbital extraocular lesions.

**Clinical:** Pulmonary infection; pyuria; hematuria; epididymitis; dysuria; flank pain; distorted calyces; productive cough.


Tunbridge-Paley Disease 1273

**General:** Onset in childhood; familial; optic atrophy and deafness seen in conjunction with juvenile diabetes mellitus.

**Ocular:** Optic atrophy; ptosis; retinal pigmentation.

**Clinical:** Hearing loss; perceptive deafness; juvenile diabetes mellitus; neurogenic bladder; Friedreich ataxia; Refsum syndrome; amnesia; epilepsy; Laurence-Moon-Biedl-Bardet syndrome.


Tuomaala-Haapanen Syndrome 1274

**General:** Unknown etiology; features similar to pseudohypoparathyroidism.
Ocular: Antimongoloid lid fissures; hypoplastic tarsus; distichiasis; nystagmus; strabismus; myopia; cataract; hypoplasia of the fovea.

Clinical: Dwarfism; short fingers and toes; wide nose bridge; small maxilla; oxycephaly; cutaneous depigmentation; alopecia; micrognathia; anodontia.


**Turner Syndrome (Turner-Albright Syndrome; Gonadal Dysgenesis; Genital Dwarfism Syndrome; Ullrich-Turner Syndrome; Bonnevie-Ullrich Syndrome; Pterygolymphangiectasia Syndrome; Ullrich-Bonnevie Syndrome)**

General: Ovarian or gonadal agenesis; 45 chromosomes with an XO sex chromosome constitution; females; rare in males; onset in childhood.

Ocular: Exophthalmos; hypertelorism; ptosis; epicanthal folds; blue sclera; corneal nebulae; cataracts; conjunctival lymphoedema; keratoconus.

Clinical: Webbed neck (pterygium colli); diminished growth; mandibulofacial disproportion; cubitus valgus; masculine chest and trunk; late appearance of pubic and axillary hair; congenital deafness; mental retardation; coarctation of aorta.


**UGH Syndrome (Uveitis-Glaucoma-Hyphema Syndrome)**

General: Caused by a defective anterior chamber lens; can be caused by toxic substance incorporated into the plastic of lens during manufacture or warped intraocular lens; syndrome may rarely occur after extra-capsular cataract extraction (ECCE) with implantation of a posterior chamber intraocular lens.

Ocular: Uveitis; glaucoma; hyphema (UGH).

Clinical: None.


**Ulcereative Colitis (Regional Enteritis; Inflammatory Bowel Disease)**

General: Chronic inflammatory disease of unknown etiology; both sexes affected; onset at all ages, most frequently between ages 20 and 40 years; usually abrupt onset; psychosomatic pathogenesis possible.

Ocular: Iritis; uveitis; episcleritis; papillomatous changes of palpebral conjunctiva; scleritis; serous retinal detachment; choroidal infiltrates; retrobulbar neuritis; papillitis; retinal pigment epithelium disturbance; choroidal folds.

Clinical: Abdominal pain; cramps; diarrhea; arthritis; weight loss; erythema nodosum; aphthous stomatitis; pallor; tenderness over colon; nutritional deficiency; carcinoma; associations with Sjögren syndrome and Takayasu disease have been reported.


**Ullrich Syndrome (Ullrich-Feichtiger Syndrome; Dyscraniopylophalangy)**

**General:** Belongs to trisomy 13-15; unknown etiology; sporadic occurrence.

**Ocular:** Microphthalmia to anophthalmia; hypertelorism; narrow lid fissures; strabismus; glaucoma; aniridia; cloudy cornea; corneal ulcers; chorioretinal coloboma.

**Clinical:** Hypoplastic mandible; broad nose; polydactyly; bicornuate uterus or septa vagina; congenital heart disease.


**Ultraviolet Radiation**

**General:** Eye and skin are the only organs of the body particularly sensitive to the nonionizing wavelengths of radiation normally present in the environment.

**Ocular:** Photokeratitis; pterygia; band keratopathy; herpes simplex keratitis; recurrent corneal erosions; discoloring of lens; retinal degeneration; cataract formation; questionable alterations to the corneal endothelium.

**Clinical:** Actinic keratosis; edema; erythema of skin, blisters of skin; depigmentation of skin; skin carcinoma.

**Unna II Syndrome (Marie-Unna Syndrome)**

**General:** Both sexes affected; rare; autosomal dominant; onset in children.

**Ocular:** Eyebrows and eyelashes missing.

**Clinical:** Loss of hair; scant growth of axillary and pubic hair, teeth, and nails; loss of hair in the eyebrows, eyelashes, and body.

**Unverricht Syndrome (Familial Myoclonia Syndrome; Lafora Disease)**

**General:** Fatal hereditary form of diffuse neuronal disease; autosomal recessive; late childhood; death within 2 to 10 years from onset of symptoms.

**Ocular:** Amaurosis; laminated Lafora bodies in ganglion cell and inner nuclear layers of the retina, either intracellular or extracellular, in inner plexiform and nerve fiber layers, and in the optic nerve.

**Clinical:** Major epilepsy; widespread myoclonus; dementia; tetraplegia; pseudobulbar palsy; generalized tonic-clonic seizure; behavioral changes; brisk tendon reflexes; cerebellar signs.
Urbach-Wiethe Syndrome (Rossle-Urbach-Wiethe Syndrome; Lipoproteinosis; Hyalinosis Cutis et Mucosae; Lipoid Proteinosis; Proteinosis-Lipoidosis) 1282

**General:** Rare autosomal recessive disorder in which hyaline material is deposited in the skin, mucous membranes, and brain; both sexes affected; onset in infancy; relatively benign progressive course; association with diabetes mellitus.

**Ocular:** Margin of eyelids may show beadlike excrescences with loss of cilia; itching of eyes; dry eyes.

**Clinical:** Skin about face covered with small, yellowish-white, waxy nodules; alopecia; hoarseness of voice at birth or within first few years of life; tongue large, thick; hyper-keratotic lesions on knees, elbows, and fingers; inability to cry; dry mouth.


Usher Syndrome (Hereditary Retinitis Pigmentosa-Deafness Syndrome) 1283

**General:** Retinitis pigmentosa associated with deaf-mutism; dominantly inherited; anatomic and metabolic condition; onset unknown (see Hallgren Syndrome).

**Ocular:** Concentric contraction of visual fields; retinitis pigmentosa with dotted, fine pigmentation in midperiphery; bone-corpseule configured pigment deposits mainly along the vessels toward the periphery; yellow-white dots in outer retina and choroid; poor night vision.


Uvea Touch Syndrome 1284

**General:** Caused by intraocular lens coming in contact with uveal tissue; seen most frequently with intracapsular cataract extraction and anterior chamber intraocular lens implant.

**Ocular:** Corneal decompensation; endothelial dystrophy; retinal edema; pigment dispersion; painful eye; disorders of motility.

**Clinical:** None.


Uveal Effusion Syndrome 1285

**General:** Congenital anomaly of sclera and, in some cases, the vortex vein; inability to transport extravascular protein across abnormal sclera; condition typically affects middle-aged men and causes recurrent, spontaneous serous retinal and ciliochoroidal detachments, often resulting in significant visual impairment.

**Ocular:** Exudative retinal detachment; sclera abnormally thick; vortex vein obstruction; idiopathic central serous chorioidopathy; vitreous cells; ciliochoroidal detachment; nanophthalmos.

**Clinical:** Viral infection; elevated blood pressure; allergic reaction; minor trauma.


Uveitis Masquerade Syndrome(s) (VMS)

**General:** Uveitis masquerade syndrome is a group of disorders that mimic uveitis; cells seen may be of noninflammatory origin or are inflammatory and secondary to another disorder.

**Ocular:** Uveitis; panuveitis; pars planitis; vitreitis; papillitis; anterior segment cells; hypopyon; vitreal infiltrates.

**Clinical:** Causes may be malignant, such as lymphoma, leukemia, retinoblastoma, melanoma, and lung cancer metastasis, or nonmalignant, such as ocular toxoplasmosis, diabetic retinopathy, hypertension, and radiation retinopathy.


Uyemura Syndrome (Fundus Albipunctatus with Hemeralopia and Xerosis)

**General:** Rare; resembles retinitis punctata albescens, fundus albipunctatus, and congenital idiopathic night blindness or Oguchi disease; avitaminosis A; affects both sexes.

**Ocular:** Night blindness; conjunctival xerosis; Bitot spots; white spots on the fundus.

**Clinical:** None.


V Esotropia Syndrome

Esotropia greater looking down by 15 prism diopters than looking up; may have underaction of superior oblique or overaction of inferior oblique; antimongoloid (downward) slant of lid fissures; may have accommodative, nonaccommodative, or paralytic esotropia components.

**Clinical:** Fusion obtained by chin depression.


V Exotropia Syndrome

Exotropia greater looking up by 15 diopters than looking down; underaction of superior oblique or overaction of inferior oblique muscles; antimongoloid (downward) slant of lid fissures.

**Clinical:** Fusion obtained by chin elevation.


Vaccinia

**General:** Laboratory virus used for vaccination against smallpox.

**Ocular:** Pustules of lids; edema of lids; conjunctivitis; orbital cellulitis; keratitis; pannus; corneal perforation; iridocyclitis; central serous retinopathy; perivasculitis; pseudoretinitis pigmentosa; ocular palsies papillitis; optic atrophy.

**Clinical:** Vesicles; pustules; erythema; fever; malaise; axillary lymphadenopathy; necrosis of skin; vaccinia gangrenosa; encephalomyelitis; drowsiness; vomiting; coma; death.

**Van Bogaert-Hozay Syndrome**

**General:** Manifest after age 3 years; similar to Rubinstein-Taybi syndrome; etiology unknown; affects both sexes.

**Ocular:** Hypertelorism; hypoplastic cilia and eyebrows; ptosis; esotropia; astigmatism; myopia. Clinical: Facial dysplasia; broad nasal bridge and zygomatic arch; flat, wide nose; arched palate; skeletal anomalies with short, thick phalangeal joints; finger and toes appear infantile; flat nasal bridge; thickened cheeks; deformed ears; micrognathia.


**Van Bogaert-Scherer-Epstein Syndrome (Primary Hyperlipidemia; Familial Hypercholesterolemia Syndrome)**

**General:** Autosomal dominant; high serum lipoprotein; both sexes affected; onset at all ages.

**Ocular:** Xanthelasma; arcus juveniles of the cornea; lipid keratopathy; cataract; retinopathy with yellowish deposits and cholesterol crystals have been reported but are more rare manifestations.

**Clinical:** Xanthelasmatosis of skin and tendons; progressive atherosclerosis; coronary insufficiency; cardiac infarcts; dementia; progressive ataxia; cerebral infarction; poly-neuropathy.


**Van Der Hoeve Syndrome (Osteogenesis Imperfecta; Osteopsathyrosis; Ekman Syndrome; Lobstein Syndrome; Spurway Syndrome; Vrolik Syndrome; Eddowes Syndrome; Brittle Bone Disease)**

**General:** Autosomal dominant.

**Ocular:** Glaucoma; blue sclera; keratoconus; cataract; optic nerve atrophy; retinopathy; retinal detachment.

**Clinical:** Brittle bones; deafness; hyperflexibility of ligaments; dental defects; developmental delay.


**Velocardiofacial Syndrome**

**General:** Autosomal dominant; anomaly of neural crest derivatives; most common syndrome of clefting; possible association with microdeletion at 22q11.
**Ocular:** Retinal vascular tortuosity; posterior embryotoxon; narrow palpebral fissures; suborbital discoloration; small optic nerves; iris nodules; cataracts; prominent corneal nerves; strabismus; hyperopia; myopia; astigmatism; anisometric astigmatism.

**Clinical:** Cleft palate; learning disability; ventricular septal defect with or without the tetralogy of Fallot; right-sided aortic arch; prominent nose; retrognathia; helical thickening; small auricles; auricular protrusion; microcephaly; small stature; inguinal or umbilical hernia; scoliosis; slender hands and digits; small vermis; small posterior fossa; developmental delay; heart malformations; late-onset psychosis.


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**Vermis Syndrome**

**General:** Medulloblastomas arising primarily in the posterior vermis but invading the fourth ventricle secondarily and compressing the bulb; frequent in children.

**Ocular:** Nystagmus; papilledema; abnormal saccades.

**Clinical:** Enlargement of the head; vomiting (early sign); stiffness and pain of the neck and shoulders; equilibratory disturbances; incoordination of the limbs.

Baker GS. Physiologic abnormalities encountered after removal of brain tumours from the floor of the fourth ventricle. *J Neurosurg* 1965; 23:338.


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**Vertebral Basilar Artery Syndrome**

**General:** "Whiplash" injury with hyperextension of the neck followed by rapid forward movement of the head or osteoarthritis of the cervical spine, cervical ribs (see Cranio-cervical Syndrome).

**Ocular:** Nystagmus (postural); internuclear ophthalmoplegia; visual deterioration; visual hallucinations may be associated with a decrease in consciousness; homonymous hemianopsia (bilateral); contralateral hemianopic visual field defect.

**Clinical:** Severe, throbbing occipital headache associated with neck pain; vertigo from ischemia of the internal auditory artery, from the temporoparietal cortex or from ischemia in the lateral tegmentum of the pons; nausea and vomiting; ataxia; hemiparesis; quadriplegia; dysarthria; dysphagia; deafness; dyslexia; atonia; confusion; coma; tremor.


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**Vesell Syndrome (Deafness-Strabismus Symphalangia Syndrome)**

**General:** Etiology unknown; possibly autosomal dominant inheritance.

**Ocular:** Strabismus.

**Clinical:** Symphalangism; hearing loss.


Vestibular Paralysis, Bilateral

**General:** Infective process bilaterally affecting the vestibular system in any site between the semicircular canals and the vestibular nuclei in the brain; usually vasculitic-type lesion.

**Ocular:** Nystagmus; no depth perception with eyes closed, no difficulty with eyes open.

**Clinical:** Staggering; inability to swim; disorientation in water; loss of hearing; vertigo.


Vincent Infection (Gingivitis; Trench Mouth)

**General:** Onset usually sudden; remission, but may reoccur; young adults.

**Ocular:** Conjunctivitis; corneal ulcer; dacyrocystitis; dacryoadenitis; orbititis; uveitis.

**Clinical:** Painful, bleeding gingiva characterized by necrosis and ulceration of gingival papillae and margins; lymphadenopathy; foul mouth odor; fever; leukocytosis.


Visual Disorientation Syndrome (Riddoch Syndrome)

**General:** Parietal lobe lesion.

**Ocular:** Visual agnosia; stereoscopic vision and central vision unimpaired; homonymous quadrantanopsia.

**Clinical:** Contralateral numbness and tingling; loss of static or postural sensation when postcentral convolution affected.

Riddoch G. Dissociation of visual perceptions due to occipital injuries, with especial reference to appreciation of movement. *Brain* 1917; 40:15.

Visual Paraneoplastic Syndrome

**General:** Loss of visual acuity and loss of visual field due to malignant disease without metastasis to visual system; most often occurs with small cell carcinoma of lung; visual symptoms may be only symptom, before neoplastic disease is diagnosed; can occur associated with cutaneous melanoma.

**Ocular:** Visual field defects; impaired color vision; narrowing of arterioles; changes in retinal pigment epithelium; orbital myositis; optic neuropathy.

**Clinical:** Endometrial carcinoma, small cell lung carcinoma.


Vitamin A Deficiency

**General:** Worldwide cause of blindness; onset in childhood; dietary vitamin A insufficiency; interference of absorption from the intestinal tract and transport or storage in the liver, as with diarrhea or vomiting; most frequent in young children.

**Ocular:** Bitot spots; conjunctival xerosis; corneal xerosis; keratomalacia with perforation; photophobia; enlarged tarsal glands; corneal ulcer; nyctalopia; hemeralopia; obstruction of the tear ducts.

**Clinical:** Diarrhea; malabsorption syndrome; follicular hyperkeratosis; lesions on buttocks, legs, and arms; xerosis of skin; tracheitis; bronchitis; pneumonia; chronic obstruction of pancreatic or biliary ducts; increased infant mortality.

**Vitiligo**

**General:** Patchy depigmentary disorder of the skin; progressive; etiology unknown; autosomal dominant; associated with Vogt-Koyanagi-Harada disease and uveitis.

**Ocular:** Posterior uveitis; depigmentation of lids, lashes, iris, and retina; retinal atrophy.

**Clinical:** Loss of pigment in hair; depigmentation of skin; halo nevi.


**Vitreocorneal Touch Syndrome**

**General:** After cataract extraction, vitreous bulges through the pupillary space, coming in contact with and finally attaching to the corneal endothelium; onset 2 to 3 weeks after cataract extraction.

**Ocular:** Area of decreased transparency of the central cornea; Descemet folds; striate keratopathy where the intact vitreous face is touching the corneal endothelium; corneal edema; iris bombe; bullous keratopathy.

**Clinical:** None.


**Vitreoretinal Skeletal Syndrome**

**General:** Associated with various systemic skeletal abnormalities; autosomal dominant.

**Ocular:** Optically empty posterior vitreous; fibrillar anterior vitreous preretinal membrane; retinal breaks.

**Clinical:** Cleft palate; hypermobility of joints; mild skeletal abnormalities; pelvic bone deformities; hearing loss.


**Vitreoretinochoroidopathy (VRCP; Autosomal Dominant Vitreoretinochoroidopathy; ADVIRC)**

**General:** Autosomal dominant.

**Ocular:** Chorioretinal hypopigmentation or hyperpigmentation; preretinal punctate opacities; retinal arteriolar narrowing and occlusion; choroidal atrophy; diffuse retinal vascular incompetence; cystoid macular edema; presenile cataracts; fibrillar condensation and moderate pleocytosis of vitreous; myopia; optically empty vitreous; lattice degeneration; retinal breaks; retinal detachment; glaucoma; spontaneous vitreous hemorrhage.

**Clinical:** None.


**Vitreous Tug Syndrome (Vitreous Wick Syndrome)**

**General:** Vitreous strand passes through the pupillary space and becomes attached or incarcerated in a corneal wound either posttraumatic or after intraocular surgery (see Irvine Syndrome); associated with endophthalmitis.

**Ocular:** Sensation of light flashes due to vitreous pull on the retina; irregular pupil; vitreous strands passing through pupil to attach to corneal wound or scar; loss of foveal reflex on ophthalmoscopic examination; circumscribed retinal edema; occasional posterior retinal detachment.
Vogt-Koyanagi-Harada Disease (Harada Disease; Uveitis-Vitiligo-Alopecia-Poliosis Syndrome)  1308

**General:** Viral infection; occurs predominantly among Italian and Japanese individuals; young adults; chronic.

**Ocular:** White lashes; secondary glaucoma; bilateral uveitis; sympathetic ophthalmitis; exudative iridocyclitis; vitreous opacities; bilateral serous retinal detachment and edema with spontaneous reattachment after weeks; depigmentation and patches of scattered pigment later; bilateral acute diffuse exudative choroiditis; papilledema; macular hemorrhage; cataracts; phthisis bulbi; poliosis; scleromalacia; intraocular lymphoma.

**Clinical:** Poliosis; vitiligo; hearing defect; headache; vomiting; meningeal irritation; reported to occur rarely in children.

Von Bekhterev-Strumpell Syndrome (Marie-Strumpell Spondylitis; Ankylosing Spondylitis; Pierre-Marie Syndrome; Bekhterev Disease; Rheumatoid Spondylitis)  1309

**General:** Variant of rheumatoid arthritis; etiology unknown; autosomal dominant; male preponderance; onset at age 20 to 40 years; although genetic background determines susceptibility to uveitis, the disease pattern suggests the possibility of random environmental triggers unrelated to the course of the underlying rheumatologic disorder.

**Ocular:** Nongranulomatous anterior uveitis; optic nerve atrophy (occasionally); hypopyon; band keratopathy; spontaneous hyphema.

**Clinical:** Spondylitis of vertebrae and sacroiliac joints; ankylosis; general arthralgia; kyphosis; scoliosis; displaced head and total rigidity of spine.

Von Economo Syndrome (Encephalitis Lethargica; Sleeping Sickness; Iceland Disease)  1310

**General:** Etiology not understood; may be caused by filterable virus; both sexes affected; onset at all ages; epidemic form.

**Ocular:** Nystagmus; strabismus; diplopia; muscle imbalance; lid retraction; homonymous hemianopsia; cortical blindness.

**Clinical:** Fever; headache; cramps; lethargy; insomnia; athetoid or choreiform movements; convulsions; depression; unsteady gait; fatigue; parkinsonism; oculogyric crisis; behavior disorder.

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Clinical: None.


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**Clinical:** Fever; headache; cramps; lethargy; insomnia; athetoid or choreiform movements; convulsions; depression; unsteady gait; fatigue; parkinsonism; oculogyric crisis; behavior disorder.

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Clinical: None.


**Von Gierke Disease (Glycogen Storage Disease Type I; Glycogenosis Type I; Glucose-6-Phosphatase Deficiency)**

**General:** Condition simulating congenital glaucoma; affects both sexes during first year of life.

**Ocular:** Corneal clouding; discrete, nonelevated, yellow flecks in macular area.

**Clinical:** Convulsions; failure to thrive; epistaxis; bleeding tendency; steatorrhea; lumbar lordosis; adiposity; hepatomegaly; enlarged kidney; renal tubular acidosis type I and kidney stones.


**Von Herrenschwand Syndrome (Sympathetic Heterochromia)**

**General:** Congenital anomaly; heterochromia with Horner syndrome; sympathetic palsy from cervical ribs, tumor of the thyroid gland, enlarged cervical lymph nodes, scars following tuberculosis, or syringomyelia in apex of the pleura.

**Ocular:** Enophthalmos; ptosis; heterochromia (ipsilateral iris); miosis; iris on the side of the sympathetic denervation usually shows subtle hypochromia.

**Clinical:** Decrease of sweating ipsilateral side of face as part of the sympathetic paralysis.


**Von Hippel-Lindau Syndrome (Retinocerebral Angiomatosis; Angiomatosis Retinae; Cerebelloretinal Hemangioblastomatosis; Lindau Syndrome; Retinal Capillary Hamartoma)**

**General:** Dominant inheritance; angiomata in the cerebellum and the walls of the fourth ventricle; young adults.

**Ocular:** Secondary glaucoma; angiomatosis of the iris; vitreous hemorrhages; tortuosity of dilated retinal artery and vein (feeder vessels); retinal exudates and hemorrhages; retinitis proliferans; angiomata of optic nerve and retina; papilledema; retinal detachment; lipid accumulation in macula; keratoconus; bilateral macular holes; choroid plexus papilloma; bilateral optic nerve hemangioblastomas.

**Clinical:** Cerebellar angiomatosis; epilepsy; psychic disturbances to dementia.


**Von Monakow Syndrome (Monakow Anterior Choroidal Artery Syndrome)**

**General:** Rupture or thrombosis of anterior choroidal artery; aneurysm; tumor.
**Ocular:** Visual field defect; hemianopia.

**Clinical:** Hemiplegia; hemianesthesia.


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**Von Recklinghausen Syndrome (Neurofibromatosis Type I; Neurinomatosis)**

**General:** Dominant inheritance activated at puberty, during pregnancy, and at menopause; strong evidence supports the existence of NFI as a tumor suppresser gene.

**Ocular:** Proptosis; displacement of the globe; pulsation of the globe; ptosis; elephantiasis of the lids; pigment spots on lids; hydrophthalmals; nodular swelling of corneal nerves; cataracts; optic atrophy; choroidal melanoma; neurofibroma of the choroid, iris, eyelid, and ciliary body; enlarged optic foramen; underdevelopment of orbital bones; cafe-au-lait spots on fundus; hamartoma of retina; congenital glaucoma; focal iris nodules; choroidal nevi; optic nerve gliomas; orbital neurofibroma; keratoconus.

**Clinical:** Cafe-au-lait skin pigmentation; fibroma molluscum; lipomas and sebaceous adenomas; schwannomas; growth abnormalities; spontaneous fractures; facial hemihypertrophy.


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**Von Reuss Syndrome (Galactosemic Syndrome; Galactokinase Deficiency; Galactosemia)**

**General:** Autosomal recessive; consanguinity; conversion of galactose into glucose is blocked, leading to galactosemia; onset after a few days or weeks of milk ingestion; deficiency of galactose-1-phosphate uridyltransferase.

**Ocular:** Searching-type nystagmus; bilateral nuclear or cortical cataracts appear clinically as oil droplets; bilateral zonular cataracts with fine punctate opacities in the lens periphery.

**Clinical:** Vomiting; refusal of food; diarrhea; weight loss; hepatomegaly with ascites; jaundice; galactosuria; aminoaciduria; dehydration; hypoglycemic crisis; failure to thrive; hypotonia; lethargy; severe mental and neurologic manifestations.


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**Von Sallmann-Paton-Witkop Syndrome (Hereditary Benign Intraepithelial Dyskeratosis; Witkop-von Sallmann Syndrome; HBID Syndrome)**

**General:** Autosomal dominant; conjunctival and oral lesions; found in whites.

**Ocular:** Foamy gelatinous plaques located in a typical horseshoe fashion at 3 and 9 o'clock positions of the perileminal area; superficial hyperemia of bulbar conjunctiva; corneal vascularization and consequent visual decrease.

**Clinical:** Thickening of oral mucosa with whitish plaques and folds of a spongy character (asymptomatic), with slow progression from birth into second decade of life.


### Waardenburg Syndrome (Van Der Hoeve-Halberstam-Waardenburg Syndrome; Waardenburg-Klein Syndrome; Embryonic Fixation Syndrome; Interoculo-Iridodermato-Auditive Dysplasia)

**General:** Irregular dominant inheritance; developmental fault in neural crest with absence of the organ of Corti, aplasia of the spiral ganglion, and pigmentary changes; no sex preference; onset at birth.

**Ocular:** Hyperplasia of the medial portions of the eyebrows; hypertelorism; blepharophimosis; strabismus; heterochromia iridis; aniridia; microcornea; cornea plana; microphthalmia; abnormal fundus pigmentation; hypoplasia of optic nerve; synophrys; poliosis; hypopigmentation and hypoplasia of retina and choroid; epicanthus; lateral displacement of inferior puncta; lenticous; underdevelopment of orbital bones; lateral displacement of inner canthi; hypopigmented iris.

**Clinical:** Congenital deafness; unilateral deafness or deaf-mutism; broad and high nasal root with absent nasofrontal angle; albinotic hair strain (unilateral); faint patches of skin pigmentation; pituitary tumor; nasal atresia; white forelock.


### Wagner Syndrome (Hyaloideoretinal Degeneration; Hereditary Hyaloideoretinal Degeneration and Palatoschisis; Clefting Syndrome; Goldmann-Favre Syndrome; Favre Hyaloideoretinal Degeneration; Retinoschisis with Early Hemeralopia)

**General:** Irregular dominant inheritance; both sexes affected.

**Ocular:** Epicanthus; nystagmus; myopia; iris atrophy; vitreous opacities with dense streaks and folds in posterior hyaloid membrane; corneal degeneration, including band-shaped keratopathy; cataracts; hyaloideoretinal degeneration (usually apparent after 15 years); narrowing of retinal vessels; pigmentary changes; type of retinal degeneration varies from case to case; retinal detachment and avascular preretinal membranes; marked choroidal sclerosis; pale optic disk; Bergmeister papilla.

**Clinical:** Palatoschisis; genua valga; facial anomalies; hypoplastic maxilla; saddle nose; hyperextensible fingers, elbows, and knees; tapering fingers.


### Waldenström Syndrome (Macroglobulinemia)

**General:** Occurs mainly in males over age 50 years; chromosomal abnormalities were reported, with most of the cells having 47 chromosomes.

**Ocular:** Sludging of conjunctival vessels; crystalline deposits in conjunctiva and cornea; keratoconjunctivitis sicca; retinal venous thrombosis; hemorrhagic glaucoma; papilledema; retinal microaneurysms; cotton-wool spots; dry eye.

**Clinical:** Adenopathy; hepatosplenomegaly; weakness; fatigue; pallor; dyspnea; weight loss; lymphoid orbital tumor.

Walker-Clodusius Syndrome (Lobster Claw Deformity with Nasolacrimal Obstruction; EEC; Ectodactyly, Ectodermal Dysplasia, and Cleft Lip/Palate) 1321

**General:** Autosomal dominant; both sexes affected; onset from birth; association with chromosome 7 abnormalities.

**Ocular:** Hypertelorism; nasolacrimal obstruction with constant epiphora; mucopurulent conjunctival discharge; keratitis; nanocanalization of the lacrimal duct.

**Clinical:** Deformities of hands and feet ("lobster claw"); absence of both index and middle fingers and second metacarpals with rudimentary third metacarpals; syndactyly; cleft palate and lips; deafness; ear malformation; renal anomalies.


Walker-Warburg Syndrome (Cerebroocular Dysplasia-Muscular Dystrophy; Warburg Syndrome; COD-MD Syndrome; Fukuyama Congenital Muscular Dystrophy; Hard + or - E Syndrome) 1322

**General:** Rare; encompassing a triad of brain, eye, and muscle abnormalities; probably autosomal recessive.

**Ocular:** Microphthalmia; cataract; immature anterior chamber angle; retinal dysplasia; retinal detachment; persistent hyperplastic primary vitreous; optic nerve hypoplasia; iris coloboma; opaque cornea; myopia; orbicularis weakness; irregular gray subretinal mottling; optic atrophy.

**Clinical:** Cerebral and cerebellar agyria-micropolygyria; cortical disorganization; gliomesodermal proliferation; neuronal heterotopias; hypoplasia of nerve tracts; hydrocephalus; encephalocele; muscular dystrophy; seizures; mental retardation; hypotonia; abnormal facies.


Wallenberg Syndrome (Dorsolateral Medullary Syndrome; Lateral Bulbar Syndrome) 1323

**General:** Occlusion of the posterior inferior cerebellar artery; onset after age 40 years; similar to Babinski-Nageotte syndrome but crossed hemiparesis is absent; nystagmus is produced by involvement of the vestibular nuclei or posterior longitudinal bundle.

**Ocular:** Enophthalmos; ptosis; spontaneous homolateral or contralateral horizontal or torsional nystagmus; miosis; Horner syndrome; skew deviation; impaired contralateral pursuit; saccadic abnormalities; gaze-holding abnormalities.

**Clinical:** Nausea; vertigo; difficulty in swallowing and speaking; ipsilateral ataxia; muscular hypotonicity; ipsilateral loss of pain and temperature sense of the face; neurotrophic skin ulcers; contralateral hypalgesia; facial weakness.


Ward Syndrome (Epitheliomatous Phakomatosis; Nevus-Jaw Cyst Syndrome) 1324

**General:** Autosomal dominant.

**Ocular:** Hypertelorism; dystopia canthorum; nevi of eyelids; congenital cataracts; congenital corneal opacities; colobomata.

**Clinical:** Basal cell nevi with multiple basolamatos nodules on face, neck, and trunk; epithelioma adenoides cysticum.
Water Intoxication Syndrome (Overhydration Syndrome)  
**General:** Administration of water in excess of kidney excretion capacity; psychogenic water drinker; may be chronic, with slow accumulation of water, or acute.  
**Ocular:** Lacrimation.  
**Clinical:** Chronic symptoms include weakness, sleepiness, apathy, anorexia, nausea, vomiting, sialorrhea, diarrhea, perspiration, behavioral changes, seizures and coma, tendon hyporeflexia, pitting edema, hemiplegia, pulmonary edema, and congestive heart failure; acute symptoms include tendon hyporeflexia, decreased attention, confusion, aphasia, muscle twitching, hemiplegia, incoordination, apathy, violent behavior, and marked muscle weakness.  

Weber Syndrome (Weber-Dubler Syndrome; Cerebellar Peduncle Syndrome; Alternating Oculomotor Paralysis; Ventral Medial Midbrain Syndrome)  
**General:** Lesion of the peduncle (crus), pons, or medulla, which interrupts the third nerve before it emerges from the peduncle and interrupts fibers in the pyramidal tract above the level of the third nuclei; hemorrhage and thrombosis; tumor of the pituitary region, extending posteriorly; also may result secondary to cerebrovascular disease.  
**Ocular:** Ptosis; homolateral third nerve palsy (usually complete); fixed, dilated pupil.  
**Clinical:** Contralateral hemiplegia; contralateral paralysis of face and tongue (supranuclear type).  

Weber-Christian Syndrome (Pfeiffer-Weber-Christian Syndrome)  
**General:** Etiology unknown; subcutaneous inflammatory lesions; occurs at any age; no gender dominance.  
**Ocular:** Secondary glaucoma; anterior uveitis; acute exudative central choroiditis.  
**Clinical:** Generalized distribution of subcutaneous nodular lesions that vary in size and are located predominantly on the trunk, arms, and legs; recurrent attacks of fever; anorexia; hepatosplenomegaly; malaise; oropharyngeal infections; myocardosis; liver cirrhosis; retroperitoneal fibrosis; ulcerative colitis; myalgia; cardiac dilatation with congestive heart failure.  

WEBINO Syndrome (Wall-Eyed Bilateral Internuclear Ophthalmoplegia)  
**General:** Differential diagnosis includes demyelinating disease, arteriosclerotic cerebrovascular disease, trauma, Arnold-Chiari malformation, syphilis, periarthritis nodosa, glioma, cryptococcal meningitis, and premature infants; usually represents midline involvement of oculomotor nucleus.  
**Ocular:** Wall-eyed internuclear ophthalmoplegia (bilateral internuclear ophthalmoplegia with exotropia).  
**Clinical:** Depends on cause; association with multiple sclerosis in young patients.
Weigener Syndrome (Wegener Granulomatosis)

**General:** Etiology unknown; occurs in fourth and fifth decades of life; persistent rhinitis or sinusitis; three characteristic features are necrotizing granulomatous lesions in the respiratory tract, generalized focal arthritis, and necrotizing thrombotic glomerulitis.

**Ocular:** Exophthalmos; lid and conjunctival chemosis; papillitis; conjunctivitis; corneal ulcer; corneal abscess; optic atrophy; optic neuritis; orbital cellulitis; episcleritis; cataract; peripheral ring corneal ulcers; ptosis; dacyrocystitis; retinal periphlebitis; cotton-wool spots; retinal and vitreous hemorrhages; ruberosis iridis; neovascular glaucoma.

**Clinical:** Severe sinusitis; pulmonary inflammation; arteritis; weakness; fever; weight loss; bony destruction; granulomatous vasculitis of the upper and lower respiratory tracts; glomerulonephritis; diffuse pulmonary infiltrates; lymphadenopathy; diffuse pulmonary hemorrhage; overlap with giant cell arteritis.


Weil Disease (Leptospirosis)

**General:** Acute severe infection caused by *Leptospira* transmitted by ingestion of food contaminated by the reservoir bacterium.

**Ocular:** Acute conjunctivitis; episcleritis; fibrinous iridocyclitis with vitreal haze; hypopyon; keratitis; pain on ocular movement; uveitis; optic neuritis; cataract; hemorrhagic retinitis; ptosis.

**Clinical:** Jaundice; fever; headaches; chills; vomiting; anemia; psychologic disturbances.


Weissenbacher-Zweymuller Syndrome

**General:** Probably neonatal expression of Stickler syndrome; autosomal dominant.

**Ocular:** Myopia; congenital glaucoma; cloudy cornea; buphthalmos; Descemet membrane tears; retinal detachment; cataracts.

**Clinical:** Flattened occiput; flat facies; low nasal bridge; anteverted nostrils; flat vascular nevus-covered glabellar area; small mouth with protruding tip of tongue; high-arched palate; short neck; limbs shorter proximally; dumbbell-shaped long bones; posterior defects in vertebral bodies thoracic region; platyspondylic evidence of coronal cleft lumbar area; deafness; small size at birth; mid face hypoplasia; parietooccipital encephalocele; hearing loss; dwarfism.


Werlhof Disease (Hemophilia and Thrombocytopenic Purpura)  
**General**: Hemorrhagic disease of unknown etiology.  
**Ocular**: Retinal hemorrhages; degeneration or severe intraocular hemorrhages with resultant retinitis proliferans.  
**Clinical**: Petechiae and ecchymoses of skin and mucous membranes; tendency to bruise; decreased level of circulating platelets with a normal clotting time.  
Werlhof PG. *Disquisitio Medica et Philologica de Variolis et Anthracibus*. Brunswick: 1735.  

Wermer Syndrome (Multiple Endocrine Neoplasia 1; MEN1; Multiple Endocrine Adenomatosis 1; MEA1; Endocrine Adenoma-Peptic Ulcer Complex; Pluriglandular Adenomatosis n)  
**General**: Autosomal dominant; high degree of penetrance; both sexes affected; onset after the first decade (see Zollinger-Ellison Syndrome).  
**Ocular**: Visual field defects secondary to pituitary adenoma.  
**Clinical**: Parathyroid adenomas or hyperplasia; pancreatic adenomas; pituitary adenomas; thyroid adenomas; adrenocortical adenomas; subcutaneous lipomas; hypoglycemic crisis; headaches; amenorrhea; diarrhea; weight loss; acromegaly; Cushing syndrome; hyperthyroidism; ulcer; cerebral aneurysm.  

Wermer Syndrome (progeria of Adults)  
**General**: Etiology unknown; recessive inheritance; consanguinity; second and third decades; possible mechanisms have been proposed to explain mutation of a gene causing inhibition of deoxyribonucleic acid (DNA) synthesis and early cellular senescence.  
**Ocular**: Absence of eyelashes and scanty eyebrows; blue sclera; juvenile cataracts; bullous keratitis; trophic corneal defects; paramacular retinal degeneration; proptosis; telangiectasia of lid; astigmatism; nystagmus; presbyopia; uveitis.  
**Clinical**: Leanness; short stature (160 cm maximum); thin limbs; short, deformed fingers; small mouth; early baldness; stretched, atrophic skin (scleropoikiloderma); telangiectasia and trophic indolent ulcers on toes, heels, and ankles; arteriosclerosis with secondary heart failure.  

Wernicke Syndrome I (Superior Hemorrhagic Polioencephalopathic Syndrome; Hemorrhagic Polioencephalitis Superior Syndrome; Encephalitis Hemorrhagica Superioris; Avitaminosis B; Thiamine Deficiency; Beriberi; Gayet-Wernicke Syndrome; Wernicke-Korsakoff Syndrome)  
**General**: Lack of vitamin B or thiamine; focal vascular lesions in the gray matter around third and fourth ventricles and sylvian aqueduct; alcoholics (adults); beriberi of all ages.  
**Ocular**: Ptosis; acute bilateral nuclear ophthalmoplegia; complete ophthalmoplegia; retinal hemorrhages; optic atrophy; optic neuritis; conjunctivitis; blepharitis; nutritional ambyopia; central scotoma; papilledema; nystagmus; absolute pupillary paralysis or Argyll Robertson pupils; accommodative palsy.
Clinical: Early prostration; lethargy; irritability; stupor; delirium; mental disturbances to Korsakoff psychosis; ataxia; tremors; peripheral neuritis; anorexia; vomiting; insomnia; perspiration; tachycardia; hallucinations; retrograde amnesia; apathy; anxiety; fear; defective concentration; cardiomyopathy.


Westphal-Strumpell Disease (Pseudosclerosis of Basal Ganglion)

General: Part of Wilson disease; mostly in men; onset at ages 11 to 25 years (see Wilson Disease).

Ocular: Brown-yellow-green ring in Descemet membrane, about 2 mm wide, which begins directly at the limbus and fades away toward the center; deposition of copper.

Clinical: Jaundice; difficulty in swallowing, speaking, and mastication; extensive muscular rigidity; coarse tremors.


West Nile Virus Infection

General: Zoonotic disease transmitted by a mosquito vector with wild birds serving as its reservoir; seen worldwide but first seen in North America in 1999

Ocular: Photophobia; chorioretinal lesions; mild vitreous inflammatory reaction; retinal hemorrhage; optic disk swelling

Clinical: Muscle rigidity; meningeal inflammation; encephalitis; poliomyelitis; acute flaccid paralysis


West Syndrome (Massive Myoclonia; Jackknife Convulsion)

General: Brain damage from trauma, anoxia, degenerative and metabolic factors and infective agents; onset in first year of life.

Ocular: Nystagmus.

Clinical: Convulsion; nodding of head; opisthotonos; mental retardation.


Weyers Syndrome (2) (Weyers IV Syndrome; Iridodental Dysplasia; Dentoideal Dysplasia; Dysgenesis Iridodentalis; Dysgenesis Mesodermalis Corneae et Irides with Digodontia)

General: Present from birth; hereditary; etiology unknown.

Ocular: Dysplasia; small perforation of iris; pupillary synechiae; microphthalmia; corneal opacity.

Clinical: Dwarfism; myotonic dystrophy; microdontia; oligodontia; hypoplasia of dental enamel.


Whipple Disease (Intestinal Lipodystrophy)

General: Multisystem disorder; prominent in males; onset between fourth and seventh decades; etiology unknown.
Ocular: Ophthalmoplegia (vertical gaze involved more than horizontal gaze); papilledema; intraocular inflammation; vitreous opacities; bilateral panuveitis; small, round grayish retinal lesions; chémosis; supranuclear ophthalmoplegia; myoclonic ocular jerks; pendular nystagmus.

Clinical: Pneumonia; pleurisy; tonsillitis; sinusitis; arthritis; fever; leukocytosis; diarrhea; malabsorption; death; dyspnea; weight loss; lymphadenopathy; polyserositis; gray pigmentation of skin; dementia; facial jerks; rhythmic movement of the mouth, jaw, and extremities.


Wildervanck Syndrome (Cervicooculoclastic Syndrome; Franceschetti-Klein- Wildervanck Syndrome; Wildervanck-Waardenburg Syndrome; Cervicooculofacial Dysmorphia; Cervicooculofacial Syndrome) 1341

General: Etiology unknown; female preponderance; similarities to Klippel-Feil syndrome; onset at birth.

Ocular: Abducens paresis; choriotica iridis.

Clinical: Deafness or deaf-mutism; torticollis with short, webbed neck; epilepsy; mental retardation; cleft palate; scoliosis; ventricular septal defect; ectopic kidney; hydrocephalus; hypoplastic thumb and growth retardation.


Williams-Beuren Syndrome (Supravalvular Aortic Stenosis; Beuren Elfin Face; Hypercalcemia Supravalvular Aortic Stenosis; Hypercalcemic Face) 1342

General: Onset at birth or early infancy; occurs in both sexes; etiology unknown; possible abnormality of vitamin D metabolism.

Ocular: Bilateral corneal opacities; hypertelorism; prominent epicanthal folds; strabismus.

Clinical: Anorexia; slow weight gain; retarded physical and mental development; elfin face; absent aortic systolic click; harsh ejection systolic murmur; tooth root hypoplasia; malocclusion; cavities.


Wilson Disease (Hepatolenticular Degeneration) 1343

General: Lesion in the putamen and lenticular nucleus; familial; occurs in first decade of life; reduced liver function; increase in copper content in tissue.

Ocular: Night blindness; golden, grayish-green, or ruby-red peripheral posterior corneal stromal deposit (Kayser-Fleischer ring); sunflower cataract (same as with retention of copper foreign body); nystagmus; extraocular muscle palsies.

Clinical: Liver cirrhosis; jaundice (early); difficulties in speaking (early); difficulties in swallowing and mastication; extensive muscular rigidity; mouth usually open with salivation ("fixed grin"); coarse tremor.

Windshield Wiper Syndrome

**General:** Following cataract extraction with a posterior chamber J loop intraocular lens; intraocular lens too short and moves from side to side with the same motion as windshield wipers.

**Ocular:** Ruptured zonules; lateral intraocular lens tilt; lateral decentration of intraocular lens; decreased visual acuity; glare.

**Clinical:** None.


Wiskott-Aldrich Syndrome

**General:** Sex-linked recessive; early infancy with death in the first decade of life; abnormal immune responses; expression of CD43 is defective in this X-chromosome-linked immunodeficiency disorder, suggesting that CD43 might have a role in T-cell activation.

**Ocular:** Periorbital hemorrhages; vesicular skin eruptions; blepharitis; lid nodules; episcleritis; scleral icterus; conjunctival hemorrhages and purulent discharge; corneal ulcers; retinal hemorrhages; papilledema; peripapillary hemorrhages.

**Clinical:** Eczema; epistaxis; purpura; hematemeses; bloody diarrhea; otitis media.


Wolf-Hirschhorn Syndrome

**General:** Partial deletion of the short arm of chromosome 4 (4p)

**Ocular:** exodeviation; nasolacrimal obstruction; foveal hypoplasia; upperlid coloboma; optic disk anomalies; microcornea; hypertelorism; nystagmus; chorioretinal coloboma

**Clinical:** Developmental delay; microcephaly; seizures; craniofacial anomalies; mental retardation; cardiac defects


Wolf Syndrome (Monosomy 4 Partial Syndrome; Chromosome 4 Partial Deletion Syndrome; Hirschhorn-Coooper Syndrome)

**General:** Partial deletion of chromosome 4 of the B group; short life expectancy; present from birth (see Cri-du-Chat Syndrome).

**Ocular:** Hypertelorism; antimongoloid slanting of palpebral fissures; ptosis; nystagmus; strabismus; iris coloboma; retinal coloboma.

**Clinical:** Microcephaly; mental retardation; seizures; ear malformations; hypoplasias; beaked nose; broad nasal root; cleft lip and palate; hypotonia.


Woody-Ghadimi Syndrome (Ghadimi-Woody Syndrome; Hyperlysineemia, Persistent)

**General:** Inherited probably autosomal dominant; affects both sexes; age of detection from infancy to early adulthood.

**Ocular:** Strabismus; ectopia lentis.
Clinical: Severe mental retardation; convulsions; laxity of joints; absence of secondary sex characteristics; hepatosplenomegaly; altered facial features.


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**Wrinkly Skin Syndrome**

**General:** Autosomal recessive; consanguinity; similar to pseudoxanthoma elasticum and Ehlers-Danlos syndrome; onset from birth.

**Ocular:** Myopia; chorioretinitis; partial optic atrophy; microphthalmia; blepharophimosis; cataract.

**Clinical:** Dry skin with many wrinkles, including the dorsal surfaces of hands and feet; many creases of palms and soles; loss of skin elasticity; no abnormal wrinkles or creases about the face; prominent venous pattern over the chest wall; mental retardation; dwarfism; kyphosis; delayed growth; small size at birth; retarded growth and development; microcephaly; craniofacial dysmorphism; skeletal anomalies.


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**X Chromosomal Deletion**

**General:** Deletion of proximal part of long arm of the X chromosome; deletion covers part of region Xq21.1-Xq21.31, the locus for choroideremia; congenital deafness; probable mental retardation.

**Ocular:** Choroideremia, translucent pigment epithelium; peripheral hyperpigmentation; diffuse choriocapillary layer and retinal pigment epithelium; decreased night vision; optic atrophy; excessive myopia; nystagmus.

**Clinical:** Congenital deafness; mental retardation; corpus callosum agenesis; cleft lip and palate; anhidrotic ectodermal dysplasia; agammaglobulinemia.


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**Xanthism (Rufous Albinism)**

**General:** Autosomal recessive; occurs in blacks.

**Ocular:** Lack of color in iris.

**Clinical:** Bright copper-red color of skin and hair.


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### Xeroderma Pigmentosum 1352

**General:** Rare autosomal recessive disorder characterized by extreme cutaneous photosensitivity; both sexes affected; onset in infancy or early childhood.

**Ocular:** Conjunctivitis; symblepharon; keratitis; corneal ulcer; blepharitis; uveitis; malignancies of conjunctiva, cornea, eyelids, and iris; ectropion; keratoconus; lid freckles; chronic conjunctival congestion; corneal opacification; bilateral pterygium; epibulbar and palpebral squamous cell corneal carcinoma.

**Clinical:** Neurologic abnormalities and cutaneous malignancies of ectodermal origin; speech disorders; spastic paralysis; convulsions; mental deficiency; gonadal hypoplasia; stunted growth; carcinoma of tongue.


### X-Linked Cone Dysfunction Syndrome 1353

**General:** X-linked pattern of inheritance; stationary cone dysfunction

**Ocular:** Myopia; visual loss; color vision abnormality


### X-Linked Mental Retardation Syndrome (XLMR) 1354

**General:** X-linked mental retardation syndrome, of which many types may exist.

**Ocular:** Glaucoma may be found in some patients; optic atrophy has been noted.

**Clinical:** Short stature; small hands and feet; seizures; cleft palate; "coarse" facial appearance; brachydactyly.


### XXXXX Syndrome (penta X Syndrome) 1355

**General:** Congenital syndrome due to aneuploidy.

**Ocular:** Epicanthal folds; hypertelorism; antimongoloid (upward slant) of palpebral fissures.

**Clinical:** Growth retardation, bilateral.


### XXXXY Syndrome 1356

**General:** 49 chromosome anomaly; characterized by mental retardation; hypoplastic male genitalia; proximal radioulnar synostosis.

**Ocular:** Upward slanting of palpebral fissures; strabismus; hypertelorism.

**Clinical:** Microcephaly; mongoloid facies; high-arched palate; cubitus valgus; in-curving fifth fingers and toes; depressed nasal bridge; nasal speech; prominent lower lip; broad chin; round face configuration; small chest; depressed sternum; wide-spaced nipples; genu valgum; flat feet; no facial or pubic hair; small testes; poor scrotal development; girdle obesity; tremor; excessive dribbling; withdrawal; irritability; proximal radioulnar synostosis; vertebral anomalies; parkinsonism.


### XYY Syndrome

**General:** Y chromosome polysomy syndrome.

**Ocular:** Colobomata.

**Clinical:** Mild mental retardation; autism; impulsiveness; aggressive behavior; developmental motor and language delays; excessive height for age; frequent antisocial behavior.


### Y Syndrome

**General:** Etiology unknown.

**Ocular:** Partial V-pattern strabismus seen only in superior gaze; a partial A-syndrome with the deviation only occurring in down gaze can be considered an inverted Y-syndrome; occurs primarily with exodeviation.

**Clinical:** None.


### Yaws (Frambesia)

**General:** Chronic infectious disease of childhood; causative agent is *Treponema pertenue*; incubation period 3 to 4 weeks; factors include poor hygiene, humidity, and scanty clothing; infectious cutaneous relapses occur during first 5 years of life.

**Ocular:** Conjunctivitis; lid granulomas; keratitis; iridocyclitis; lid ulceration; cornea leukoma; cicatrical ectropion.

**Clinical:** Skin lesion and relapsing secondary lesion of skin and bone; painful papules of soles of feet; crablike gait; fever; lymphadenopathy; nocturnal bone pain; periostitis; gummas of skin and long bones; hyperkeratoses of soles and palms; ostitis; periostitis; hydrarthrosis.


### Yellow Fever

**General:** Acute infectious disease of short duration and extremely variable severity.

**Ocular:** Lid edema; orbital pain; subconjunctival, vitreal, and anterior chamber hemorrhages; mydriasis; optic neuritis; partial optic atrophy.

**Clinical:** Fever; headache; nausea; epistaxis; relative bradycardia; albuminuria; convulsions; tongue shows red margins with white furred center; copious hemorrhages; anuria; delirium; slow pulse; jaundice; "black vomit"; hematemesis; coma; death.


### Yersiniosis

**General:** Caused by gram-negative bacilli *Yersinia enterocolitica* or *Yersinia pseudotuberculosis*.

**Ocular:** Conjunctivitis; corneal ulcer; endophthalmitis; panophthalmitis; uveitis; retinal hemorrhages; cataract; hypopyon; Parinaud oculoglandular syndrome; iridocyclitis; panuveitis; disseminated chorioretinitis.

**Clinical:** Gastroenteritis; diarrhea; vomiting; fever; acute terminal ileitis; mesenteric adenitis; arthritis; erythema nodosum.

Young-Simpson Syndrome

**General:** Rare congenital syndrome.

**Ocular:** Blepharophimosis.

**Clinical:** Congenital hypothyroidism; congenital heart defects; facial dysmorphism (microcephaly, bulbous nose, low-set ears, micrognathia); cryptorchidism in males; hypotonia; mental retardation; postnatal growth retardation.


Zellweger Syndrome (Cerebrohepatorenal Syndrome of Zellweger)

**General:** Rare; congenital; lethal disease; prevalent in females; demyelination of cerebral white matter, spinal cord, and peripheral nerves; enzymatic defects cause myelin deficiency (see Smith-Lemli-Opitz Syndrome); severe multisystem disorder resulting from defective biogenesis of the peroxisome causes death within the first year.

**Ocular:** Hypertelorism; microphthalmia; nystagmus; glaucoma; hemimydriasis; corneal opacities; cataract; narrowing of retinal vessels; pigment irregularities and areas of depigmentation; retinal holes without detachment; tapetoretinal degeneration; irregular border-lined optic disks; gray-colored disks; hypoplastic supraorbital ridges; optic atrophy.

**Clinical:** Hypotony; hepatomegaly; albuminuria; mental retardation; failure to thrive; vomiting; seizures; low birth weight; jaundice; short stature; broad nose; hypospadias; cryptorchidism; sepal defect; craniofacial dysmorphic features; high forehead; renal cyst; psychomotor retardation; hepatosplenomegaly; severe hearing impairment.


Zieve Syndrome (Hyperlipemia-Hemolytic Anemia-Icterus Syndrome)

**General:** Unknown etiology; pancreatitis; prevalent in middle-aged males with history of recent alcohol intake; insidious onset.

**Ocular:** Moderate-to-marked icterus; cloudy cornea; corneal ulcers; retinal lipemia.

**Clinical:** Anorexia; nausea and vomiting; epigastric pain; diarrhea; weight loss; fatigue; weakness; jaundice; hepatomegaly.


General: Variant of Fanconi familial aplastic anemia; recessively inherited with male linkage; consanguinity; onset between ages 5 and 13 years.

Ocular: Ectropion; chronic blepharitis; obstruction of lacrimal puncta; conjunctival keratinization; bullous conjunctivitis; epiphora; nasolacrimal duct obstruction; loss of lashes; cataract; glaucoma; strabismus; abnormal fundi.

Clinical: Congenital dyskeratosis with pigmentation of "marble" configuration or "gun metal" appearance; atrophic areas and telangiectasis; dystrophy of nails; vesicular and bullous lesions of oral cavity followed by ulceration; mucosal atrophy; leukoplakia; aplastic anemia; defect of teeth; physical and mental development may be retarded; tufts of hairs on the limbs; keratinized basal cell; papillomas on the trunk.

Zinsser F. Atrophica Cutis Reticularis cum Pigmentatione, Dystrophia Unguinum et Leukoplakia Oris. *Ikonographia Dermat (Kyoto)* 1906; 219-223.

**Ziprkowski Syndrome**

General: X-linked recessive albinism.

Ocular: Ocular albinism.

Clinical: Albinism generalized.


**Zollinger-Ellison Syndrome (Polyglandular Adenomatosis Syndrome; Multiple Endocrine Adenomatosis Partial Syndrome)**

General: Autosomal dominant; more frequent in males (2:1); etiology is islet cell adenoma of pancreas secreting a gastrin-like material; onset third to fifth decade (see Werner Syndrome).

Ocular: Scotomata according to size and position of pituitary tumors; optic nerve atrophy; papilledema; bilateral extraocular muscle metastases.

Clinical: Enteritis and/or peptic ulcers; malignant or benign tumor of islet cell of pancreas; hypersecretion; vomiting; diarrhea; polyglandular adenomatosis; endocrine involvement.
