ETIOLOGY, PATHOPHYSIOLOGY AND SYSTEMIC ASSOCIATIONS OF DUANE RETRACTION SYNDROME

DR. MEENAKSHI SWAMINATHAN
SN-ORBIS PEDIATRIC OPHTHALMOLOGY LEARNING AND TRAINING CENTRE, SANKARA NETHRALAYA, CHENNAI, INDIA
DISCLOSURE AND CONSENT

- No financial conflict of interest to disclose

- Have consent of patients to share photos for educational purpose
AUDIENCE POLL #1

- YOU ARE A/AN
  1. OPTHALMOLOGIST
  2. RESIDENT (POSTGRADUATE)
  3. FELLOW IN TRAINING
  4. ALLIED OPTHALMIC PERSONNEL (OPTOMETRIST, ORTHOPTIST, OPTHALMIC TECHNICIAN)
  5. ALLIED OPTHALMIC PERSONNEL IN TRAINING
  6. OTHERS (INCLUDING FAMILY AND FRIENDS)
CCDD (CONGENITAL CRANIAL DYSINNERVATION DISORDER)
COMPLET OR PARTIAL ABSENCE OF ABDUCTION

• RETRACTION OF GLOBE ON ADDUCTION
• NARROWING OF PALPEBRAL FISSURE ON ADDUCTION
PARTIAL DEFICIENCY OF ADDUCTION

OBLIQUE MOVEMENTS WITH ATTEMPTED ADDUCTION
UPSHOOT OR DOWNSHOOT OF THE GLOBE WITH ATTEMPTED ADDUCTION

REDUCED CONVERGENCE
OTHER NAMES?

Stilling-Türk-Duane syndrome
ALEXANDER DUANE-USA-1905
EPIDEMIOLOGY AND DEMOGRAPHICS
- 1 in 1000/1 in 10,000 in general population
- 1-5% of all strabismus cases
- Isolated unilateral finding or in conjunction with other congenital anomalies
- Females > Males
  - Susceptible genes are sex limited
  - Higher estrogen levels during embryogenesis
- Left eye > Right eye in Type 1 and 3
ETIOLOGY
MECHANICAL ANOMALIES

- Türk: abnormally tight LR which behaves like an inelastic band (1899)

- Wolff: noted a congenital anomaly (1900)

- Could this be related to birth trauma or nuclear aplasia? (1920)
MECHANICAL ANOMALIES

- Dual insertion of Medial rectus: one in the usual place and the other posteriorly, the contraction of which can cause the globe retraction

- No support found during surgery
1956 Breinin

- Studied the electrical potential generated by muscles in various gazes in DRS
- NO LR potentials in abduction; maximum LR potentials in adduction.
- Concluded that LR is receiving innervation during adduction of the eye

All studies found MR innervation to be normal

Strachan and Brown quantified this paradoxical innervation

Synergistic innervation between MR and verticals and obliques also were found

*Brit. J. Ophthal. (1972) 56, 594*

Electromyography of extraocular muscles in Duane’s syndrome

I. M. STRACHAN AND B. H. BROWN
Hollamshire Hospital, Sheffield
Metz noted slowing of abduction and adduction saccades during the saccadic velocities recording in DRS

Sixth nerve innervation to the LR was defective

Nuclear
- anomalous innervation by the oculomotor nerve
- Absent or hypoplastic abducens nucleus

Infranuclear
- Cavernous sinus where the III and VI nerve are closer anatomically
NEUROLOGIC ANOMALIES

- Presence of synkinesis along with DRS
  - Marcus Gunn Jaw winking
  - Crocodile tears
- Abducting nystagmus in the normal eye also supports brainstem anomaly
Absence of the abducens nerve in duane syndrome verified by magnetic resonance imaging

MD Cameron F. Parsa, MD P. Ellen Grant, MD William P. Dillon Jr, PhD Sascha du Lac, MD William F. Hoyt
Clinical correlation of imaging findings in congenital cranial dysinnervation disorders involving abducens nerve

Chanchal Gupta, Pradeep Sharma, Rohit Saxena, Ajay Garg, and Sanjay Sharma
Congenital innervation dysgenesis syndrome (CID)/congenital cranial dysinnervation disorders (CCDDs)
- 90% Sporadic
- 10% Familial
  - Autosomal dominant inheritance if part of a syndrome
- Twin studies
- Autosomal dominant variant: CHN1 gene

- Isolated: DURSI gene on 2q31

- Duane radial ray syndrome: SALL4 gene on chromosome 20
Expansion of the CHN1 Strabismus Phenotype

Noriko Miyake,1,2,3 Joseph L. Demer,4,5,6,7 Sberin Sbaaban,1,8,9,10 Caroline Andrews,1,8,9,11 Wai-Man Chan,1,8,11 Stephen P. Christiansen,12,15 David G. Hunter,14,15 and Elizabeth C. Engle1,2,8,9,11,14,15,16,17

Purpose. Hyperactivating CHN1 mutations have been described in individuals with Duane retraction syndrome with or without vertical gaze abnormalities. This was a study of five family members with distinctive ocular dysmotility patterns that co-segregated with a novel hyperactivating CHN1 mutation.

Results. All five clinically affected family members exhibited monocular or binocular supraduction deficits, three in the absence of Duane retraction syndrome. MRI in four affected individuals demonstrated small or absent abducens nerves in all four, small oculomotor nerve in one, and small optic nerves in three. Superior oblique muscle volume was also decreased in three of the individuals, supporting trochlear nerve hypoplasia. Strabismus segregated with the CHN1 locus and affected
Duane retraction syndrome: causes, effects and management strategies

Abstract: Duane retraction syndrome (DRS) is a congenital eye movement anomaly characterized by variable horizontaluction defects, with narrowing of the palpebral fissure and globe retraction on attempted abduction, occasionally accompanied by upshoot or downshoot. The etiopathogenesis of this condition can be explained by a spectrum of mechanical, innervational, neurologic and genetic abnormalities occurring independently or which influence each other giving rise to patterns of clinical presentations along with a complex set of ocular and systemic anomalies. Huber type I DRS is the most common form of DRS with an earlier presentation, while Huber type II is the least common presentation. Usually, patients with unilateral type I Duane syndrome have exotropia more frequently than exotropia, those with type II have exotropia and those with type III have exotropia and exotropia occurring equally common. Cases of bilateral DRS may have variable presentation depending upon the type of presentation in each eye. As regards its management, DRS classification based on primary position deviation as exotropia, exotropia or orthotropia is more relevant than Huber’s classification before planning surgery. Surgical approach to these patients is challenging and must be individualized based on the amount of ocular deviation, abnormal head position, associated globe retraction and overshoots.

Keywords: Duane syndrome, strabismus surgery, exotropia, overshoots
AUDIENCE POLL #2

Which of the following can be seen in DRS?

1. Absence or hypoplasia of abducens nucleus
2. Anomalous innervation by the oculomotor nerve
3. Hypoplasia of the trochlear nerve
4. All of the above
SYSTEMIC ASSOCIATIONS
DUANE RADIAL RAY SYNDROME OR OKIHIRO SYNDROME

- Shortened radius
- Abnormal angulation of wrist and/or thumb

https://radiopaedia.org/images/18589027
GOLDENHAR SYNDROME

- Ptosis
- Preauricular skin tags
- Epibulbar dermoid
- Deafness
- Coloboma
- Pinna defects
KLIPPEL FEIL ANOMALY

- Fusion of at least two vertebrae of neck
- Short neck
- Marcu Gunn Jaw Winking
- Micro cornea
- Optic nerve hypoplasia
- Cleft palate
- Facial asymmetry

HOLT ORAM SYNDROME

<table>
<thead>
<tr>
<th>Skeletal abnormalities of the hands and upper limbs</th>
</tr>
</thead>
<tbody>
<tr>
<td>Horner’s syndrome</td>
</tr>
<tr>
<td>Keratoconus</td>
</tr>
<tr>
<td>Morning glory disc</td>
</tr>
<tr>
<td>Cardiac anomalies</td>
</tr>
</tbody>
</table>
WILDERVANCK SYNDROME

- Klippel Feil anomaly
- Deafness
- Nystagmus
- Myelinated Nerve fiber
- Limb deformities
ARTHROGRYPOSIS MULTIPLEX CONGENITA

Multiple vertebral anomalies
- cataract
- staphyloma
- Spina bifida
- Microphthalmos

Published in 2005
THE 1960s EPIDEMIC OF ARTHROGRYPOSIS MULTIPLEX CONGENITA
Pitter, F., Williams
OCULOCUTANEOUS ALBINISM
THALIDOMIDE EMBRYOPATHY

Remember to check other systems

Paves way for future genetic studies

Children who need surgery for DRS may be difficult patients from anesthesia perspective
THANK YOU