Congenital Cataract; Current Diagnosis and Management

ORBIS Cybersight
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Thank You!

>2000 participants

• No Financial Interest in Any of the Topics Discussed.
Question

• What is your profession?
  1. General Ophthalmologist
  2. Pediatric Ophthalmologist
  3. Ophthalmic Technicians / Orthoptists
  4. Doctor in training (resident / fellows)
  5. Other – Specify
Question

• How Many Cases of Congenital Cataracts do you perform per month?
  1. 0 to 1
  2. 2 to 4
  3. 5 to 10
  4. 10 to 20
  5. >20
Question

• How Long Have you practiced in ophthalmology
  1. 1 to 5
  2. 6 to 10
  3. 11 to 15
  4. 15 to 20
  5. >20
Overview

• What is Cataract? Embryology
• Diagnosis and Management
• Timing of Surgery
• IOL?
• Preoperative consideration
• Surgical Techniques
• Postoperative considerations
Lens: Embryology

- The surface **ectoderm** over the eye field thickens into the lens placode.
Lens: Embryology

- Invaginates toward the developing optic cup, forming the lens pit.
Lens pit closes, and the resulting lens vesicle (LV) pinches off from the surface ectoderm; forming Human Lens around 4 weeks GA
29 Week GA Cataracts
Lens

• Lens transmits light with wavelengths from 390 - 1200 nm.
  – limit of visual perception (390 to 740 nm).
• Lens transparency: Architecture of lens cells and tight packing of the proteins
• At birth, lens weighs 65 mg. It grows to about 160 mg in the first decade and then more slowly to about 250 mg by 90 years of age. (4 X the weight !)
Lens Composition

• Mostly water-soluble proteins:
  – Crystallins (90% of the lens)
  – Critical structural role for transparency and refraction
  – α-crystallin 40%, β-crystallin 35%, and γ-crystallin 25%

• Lens opacity (cataracts)
  – malfunction of α-crystallin due to genetic basis or ageing

• Encoded by CRYAA on chromosome 21q22.3 and CRYAB on chromosome 11q22-q22.3
  – Also expressed in skeletal muscles, heart, kidney and brain
Growth of Eye

- Newborn eye axial length (AL)
  - AL: 16.8 mm
- In adults
  - AL: 23.6 mm
  - 40% increase!

- **Lens** also reduces its refractive power to compensate the axial length changes.
  - if not we all we would be wearing very thick myopic glasses
Growth of the Lens

- Lens changes significantly 1\textsuperscript{st} year of life to compensate for the AL.
  - > 50\% growth in axial length occurs before 1 year of age
Prevalence of Congenital Cataracts (CC)

- WHO estimated that **2.2 billion** people are visually impaired around the world, out of which **65.2 million** people are affected with cataract.

  [https://www.who.int/publications-detail/world-report-on-vision](https://www.who.int/publications-detail/world-report-on-vision)

- **14 million children** are bilaterally blind from cataracts.
Congenital cataracts: Overview

• One of the most common Treatable cause of childhood blindness
  – 1/3 of cases of blindness in infants
• 15% of blindness worldwide
• Prevalence
  – 1 / 2,000 – 1 / 10,000
• If untreated, vision loss occurs due to stimulus deprivation
How Do They Present?

- Family history
- Abnormal reflex (Leukocoria) per parents
- Fetal Ultrasound
  - Fetal orbits and lenses can be seen by ultrasound from 14 weeks
- Sudden onset in older children (DM)
- Referred on routine Vision Screening
Description of Congenital Cataract

• Unilateral vs bilateral
• Congenital vs acquired
• Partial vs complete
• Stable vs progressive
• Isolated vs assoc. with systemic dz
Etiologies: Overview

• Bilateral Cataract Causes
  – $\frac{1}{4}$ hereditary: most common AD
  – $\frac{1}{4}$ associated diseases
  – $\frac{1}{2}$ idiopathic*

• Genetic testing
Etiologies: Overview

- **Unilateral Cataracts Causes**
  - Idiopathic (most common)
  - Traumatic
  - Post Lenticonus
  - PFV/PHPV
  - Uveitis
  - ROP treatment - Laser
  - Secondary to RD/RB
- **Genetic testing: more common**
  - Idiopathic
Question

Do you routinely refer CC patients for Genetic Testing?

1. Yes
2. No
3. Certain Cases
4. This service is not readily available
Question

• When do you refer CC patients for Genetic Testing?
  1. All cases
  2. All Bilateral Cases
  3. All Unilateral Cases
  4. If negative Family H/O, No systemic disease, and Negative w/u
  5. Other – specify
Bilateral Cataracts Etiologies

- Chromosome
  - Trisomy 21, 13, 18
- Cranial facial syndromes
  - Hallerman-Streiff
  - Rubenstein-Taybi
  - Smith-Lemli-Opitz
Bilateral Cataracts Etiologies

- Intrauterine Infection
  - Rubella
  - Toxoplasmosis
  - Herpes
  - CMV
  - Varicella
  - Syphilis
- Uveitis
Bilateral Cataracts Etiologies

- **Metabolic diseases**
  - Galactosemia, Fabry, Wilson, Mannosidosis, DM, Chondrodysplasia Punctata, MPS

- **Musculoskeletal disorders**
  - Albright syndrome
  - Myotonic dystrophy
  - Sengers syndrome (hypertrophic cardiomyopathy)

- **Renal Syndrome**
  - Lowe syndrome
  - Alport syndrome
How to Evaluate?
Evaluation of Cataracts

• Family History
• Detailed history of child growth, development and systemic disorders, trauma
• Pediatric physical examination
  – hearing loss, mental retardation, development delay, kidney disease, possible chromosomal abnormalities
• Ocular Examination
  – Laterality
  – K diameter
  – Iris configuration
  – AC depth
  – Lens position
  – Cataract morphology
Evaluation of Cataracts

• Posterior Segment
  – Rule out posterior mass
  – Rule out RD
  – Rule out optic nerve stalk to lens

• IOP

• Ultrasound if no view of the posterior pole
Laboratory Studies

• NO Workup:
  – **Unilateral**: no other systemic findings
    • Complete physical exam by pediatrician
  – **Bilateral**:
    • Positive family history

– Genetic Testing becoming more common*
Workup

• Infection: TORCH profile, varicella titer (maternal rash during pregnancy), VDRL

• Metabolic:
  – Galactose metabolism:
    • Urine for reducing sugar, Galactose-1-phosphate uridyltransferase, Galactokinase
    • Urine and Plasma amino acid (Lowe)
  – Calcium (low in hypoparathyroidism)
  – Phosphorous (high in hypoparathyroidism)
  – Blood glucose, 7 dehydrocholesterol
  – Ferritin (high hyperferritinemia)
  – CXR (hypertrophic cardiomyopathy)

• Morphology based workup:
Workup

• **Geneticist** for further testing
  - Dysmorphic, Not feeding well with developmental delays
  - Congenital cataract NGS (Next Generation Sequencing) test from a commercial lab: > 70 to 80% of patients
  - Challenge: Cost of tests and the variable insurance coverage
Types of Cataracts
Anterior Polar Cataracts (APC)

- < 3mm in diameter
- Unilateral or bilateral
- Anisometropia and amblyopia
Types of Anterior Cataract

1. Dot anterior polar (aka anterior capsular). Usually benign and stable, can sometimes cause amblyopia. The capsular portion does NOT enlarge. It is the underlying anterior cortical component, if any, that grows.

2. Anterior pyramidal. Histologically this is heaped up PAS + material. The pyramid does not grow but underlying cortical component can. May be large with protrusion and sx.

3. Anterior lenticonus. anterior cortical cataract and can enlarge: Look for renal disease!
Anterior Cataract

4. Anterior subcapsular (aka anterior cortical). These can enlarge: Assoc w/ trauma, radiation, uveitis, atopic skin disease

5. PFV “traction cataract”: Eccentric anterior capsule plaques associated with persistent iris strands arising from the collarette. The strands can break leaving pigment on the lens capsule. May need removal if visually significant.
Nuclear Cataracts

- Opacities that involve the center or nucleus
- 3 mm or larger
- Irregularity of the lens fibers extend into periphery
- Bilateral in most cases
- Inherited or sporadic
- Congenital but may become dense as they grow
- Risk of glaucoma
Nuclear
Nuclear
Lamellar Cataract

- Discrete, round shape, lamellar cataracts affect 1 or more layers of the developing lens cortex surrounding the nucleus
- 5 mm or larger
- Bilateral more common
- Good prognosis
Lamellar
Posterior Subcapsular Cataract (PSC)

- PSC less common in children
- Acquired and bilateral and progressive
- Corticosteroids, uveitis, radiation exposure
- Processive
- PSC seen in NF type 2
Sectoral Cataract

- Wedge shaped cortical cataract
- Idiopathic
- Occult posterior segment tumor, blunt trauma, retinal coloboma with fibrous bands attached to the posterior lens capsule
Peripheral Vacuolar Cataract

- Premature infants
- Idiopathic
- Encountered during ROP exam
- Visually insignificant and can resolve with time
Posterior Lenticiconus

- Progressive thinning of central posterior capsule
- Bulging posterior lens capsule
- Oil droplet on retinoscopy
- Unilateral in most cases
- Lowe syndrome
- Caution during surgery!
PFV (Anterior/Posterior/Combined)

- Microphthalmia
- Glaucoma
- 90% unilateral
- Elongation of Ciliary process
- Poor prognosis
Persistent Fetal Vasculature

- PFV (PHPV): most common cause of unilateral cataract
- Idiopathic in unilateral cases
- Bilateral: Assoc with systemic and neurological abnormalities
- Spectrum of severity
  - Hyaloid vessel remnants, Mittendorf Dot, Bergmeister papilla
  - Opacity of retrolental plaque: Cartilage and fibrovascular tissue
  - Thick fibrous persistent hyaloid artery with traction of the optic n. and retina.
- RB may be considered: microphthalmos and cataracts unusual
Management
Management

- Most Anterior opacities
  - No surgery (retinoscopy)
  - Irregular astigmatism
- Younger the patient: Denser the cataract before consider sx.
- Refract to see if glasses help in older children (subluxation)
- Treat amblyopia for partial cataracts
  - Dilation or Patching
  - Watch behavior while patching
Pediatric Cataract Surgery Considerations

• Different than Adult Sx
  – Etiology may need to be determined
  – 3 per 10,000 live births: rare
  – Elastic Ant and Post capsules / Smaller
  – Visual outcome – amblyopia
  – Post op care: complicated/increased inflammation / PCO 100%
  – Multiple surgeries may be needed
  – Long term issues and management
  – Become lifelong patients
Pediatric Cataract Surgery Considerations

- Requires General Anesthesia
- Small Palpebral Fissure
- Soft Eye and leak: Close surgical incisions
- Shallow AC (1.2 mm vs. 3-4 mm)
- Poor Pupil Dilation
- Aphakic or IOL
- Posterior Capsule management/ VAO
Pediatric Cataract Surgery Impact

• Parents
  – Anxiety and fear level
  – Unknown
  – Guilt
  – Greater Psychosocial impact than the physical well-being (Multiple studies)
Pediatric Cataract Surgery Impact

- Dx and Treatment has negative long-term impact on parents and children
- HR-QoL in children/parents with CC
  - Similar to levels of Severe Heart Defects or Liver Transplants
- Psychosocial scores:
  - comparable to children undergoing Acute Lymphocytic Leukemia treatment
You Say

• “Your child has a congenital cataract and we may be able to fix it”

Parents Hear

• “There is something wrong with my baby’s eye, she may be BLIND for the rest of her life”
Potential Miscommunication

• Parents
  – Internet search
  – Neighbors/ Friends
    • “Stem cells help / Gene therapy”
    • “Bifocal implants”
    • “Laser can fix anything”
    • “Eye Transplants (Artificial Eyes)”
When to intervene?

- **Dense cataracts**
  - Urgent removal and optical correction
  - Nystagmus = poor prognosis, can resolve

- **Partial cataracts**
  - Judgment call
  - Party line: 3 mm or greater
  - Patching helpful

- **Central and posterior cataract**: more visually significant
When to intervene?

My approach:

- Central >3mm in diameter; Retinoscope
- Assoc w/strabismus
- Assoc w/ nystagmus
- Retinoscopy
  - If you can’t refract, then baby can’t see
- Direct ophthalmoscopy
  - If you can’t see, then neither can baby
Timing of Surgery
Question

• Timing of Unilateral Cataract?
  1. At Birth
  2. < 2 weeks
  3. 2 to 4 weeks
  4. 4 to 10 weeks
  5. 10 to 16 weeks
Question

• Timing of bilateral Cataract?
  1. At Birth
  2. < 2 weeks
  3. 2 to 4 weeks
  4. 4 to 8 weeks
  5. 8 to 12 weeks
Question

• Timing between Bilateral Cataracts for Sx?
  1. Same Day (change drapes)
  2. 1 week
  3. 2 weeks
  4. 4 weeks
  5. 8 weeks
Question

• At what age do you try to place IOL in congenital cataract cases? Unilateral cases
  1. Never
  2. From birth
  3. 6 mo to 11 mo
  4. 1 yo to 2 yo
  5. >2 yo
Question

• At what age do you try to place IOL in congenital cataract cases? Bilateral cases
  1. Never
  2. From birth
  3. 6 mo to 11 mo
  4. 1 yo to 2 yo
  5. >2 yo
Timing of Surgery

• Unilateral dense congenital cataract:
  – Unilateral: 4 - 10 weeks (health of the baby)
  – Risk of glaucoma with earlier cat sx IATS
    • < 4 weeks)

• Bilateral dense cataract:
  – Bilateral: 2 - 3 months
  – Short time interval between cases
  – Bilateral sx on same day not recommended
    • Cost/Risk of Anesthesia
      Travel/Location/Availability of Healthcare
IOL or No IOL? Considerations

- Unilateral vs Bilateral cataracts
- Nystagmus
  - Contact lenses difficult to fit and position
- Dry eye
  - e.g. previous radiation therapy
- Dirty or sandy living conditions
- Availability of CTL or Glasses
- Compliance and Hygiene concerns
- Behavioral issues
- Cost
- Follow up issues
IOL or No IOL? Considerations

- Children with chronic inflammatory disease
  - Active Uveitis or poorly controlled JIA iritis
- Nanophthalmos
- Microphthalmia, with corneal diameters of less than 9 mm,
  - difficulties with lens size.
IOL or No IOL? Considerations

• Less than <7 months old
  – IATS higher complication (VAO) and repeat sx
• >7 months: accepted
• Corneal diameter 9 -10 mm or greater
• Lens diameter 8 mm or greater
  – IOL lens diameter 6.0 mm
  – Cut the haptics in IOL to fit in smaller eye
Once IOL Placement Decided

- Measurement of AL and K
  - Difficult in office setting
  - Manual or automated Keratometry

- EUA needed

- Errors in measurement
  - Lack of fixation under GETA
  - A scan biometry should be performed OU
Biometry

- Conventional U/S biometry
  - Applanation vs Immersion
- Optical biometry
Contact vs Immersion

• Contact:
  – Convenient
  – Accurate with trained technicians
  – Corneal compression: 0.32 mm error rate
    • Due to soft cornea and sclera

• Immersion:
  – More accurate
  – Low risk of error
  – Cumbersome procedure
  – More time required
Once IOL Placement Decided

• Perform lensectomy
• Place IOL (in bag if possible)
• Primary posterior capsulotomy vs YAG in future
AL Error

- 1.0 mm error: 3.0 D
  - In long eyes: 1.75 D
  - In shot eyes: 3.75 D
AL Error prevention

• Well trained biometrist
• Calibrate manual keratometer/ IOL masters
• CTL out for 2 weeks before keratometry
• IOL under correction
Choosing IOL

- **PMMA**
  - Longer experience
  - Large wound

- **Silicone**
  - Future risk of retinal issues

- **Acrylic**
  - Small wound
  - SA/SN series very flexible
  - SA/SN series edge design
IOL Calculation Dilemma

• Difficult Issue: varying degrees of refractive myopic shift after surgery
IOL Calculation Dilemma

• Refractive outcomes
  – Growth of eye follows logarithmic curve with greatest growth in first 2 years of life
  – Refractive growth continues up to 20 y/o
  – AL increases from 16.8mm @ birth to 23.6 mm in adults
  – Corneal curvature decreases from 51.2D to 43.5D
## Projected Lenticular Refractive Changes

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<th>Age</th>
<th>D</th>
<th>iopter Change</th>
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<tr>
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<td></td>
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<tr>
<td>2</td>
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<td></td>
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<tr>
<td>4</td>
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<td>10</td>
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<tr>
<td>12</td>
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<tr>
<td>14</td>
<td>-2.00</td>
<td></td>
</tr>
<tr>
<td>16</td>
<td>0</td>
<td></td>
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</table>

![Graph showing the relationship between age and lenticular refractive changes](image-url)
IOL Calculation Dilemma
IOL power: Target refraction

• Problems with any formula:
  – Anticipation of ocular growth with Individual variability
  – Growth and other optical factors mean possible shift of 20 D from age 0-20 yr.
  – Lens position changes with ocular growth
IOL power: Target refraction

• Two Schools of Thought
  – Emmetropia
    • Easier to treat amblyopia
    • Deal with high myopia later
    • An implant of 28 D producing emmetropia in an 8-month-old may induce 7 D of myopia and anisometropia when the child is 3 yo.
  – Low power IOL – aim for 6-10 D hyperopia
    • Supplement with contact lenses for first year
# Commonly used Age Adjusted Target Refraction

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<tr>
<th>Age</th>
<th>Refraction</th>
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<tr>
<td>1</td>
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</tr>
<tr>
<td>2</td>
<td>+3.5 to +4.0</td>
</tr>
<tr>
<td>3</td>
<td>+2.5 to +4.0</td>
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<tr>
<td>4</td>
<td>+2.5 to +4.0</td>
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<tr>
<td>5</td>
<td>+2.0 to +3.0</td>
</tr>
<tr>
<td>6</td>
<td>+2.0</td>
</tr>
<tr>
<td>7</td>
<td>+1.5</td>
</tr>
<tr>
<td>8</td>
<td>+1.0</td>
</tr>
<tr>
<td>9 and over</td>
<td>emmetropia</td>
</tr>
</tbody>
</table>
Which Formula?

- A study comparing the predictive accuracy of four common IOL power formulas (SRK-II, SRK-T, Holladay, and Hoffer Q) in children did not reveal any significant predictive differences between the formulas.

- SRT /T in most patients
  - Barrett Universal 2

- Hoffer Q in infants <1 yo
Surgical Technique
Keep It Simple!

• “Simple can be harder than complex: You have to work hard to get your thinking clean to make it simple. But it's worth it in the end because once you get there, you can move mountains.” - Steve Jobs
KEEP IT SIMPLE!
Watch Your Posture
Surgery

- Dilation – important
- Povidone Iodine 5%
- Phenylephrine in IVF
- Tight incision with tunnel – soft eye
- Vision Blue
- I/A / Vitrector and No PHACO
- Viscoelastics*
- Triesence and Moxifloxacin (Subconjunctival and intracameral injections)
- 10.0 Vicryl to close
- Wound check: Seidel
  - No Heparin, Post-op Dilation, Air bubble post op
Honey vs. Gelatin Viscoelastistics

Dispersive versus Cohesive
Anterior Capsule

• Capsulorrhexis is much more difficult in children
• If the tear begins to extend too far to the periphery of the lens, the technique should be abandoned rather than risk an area of zonulolysis.
• Trypan Blue dye helpful
• Around 4.5 to 5.5 mm opening
  – IOL 6mm
Adult or Teenage Capsule

Direction of Pull

Younger Child’s Capsule

Direction of Pull
Adult Lens
Tips for Anterior Capsule Capsulorrhexis

• Very elastic
• Radial Forces
• High Viscosity viscoelastic
  – Healon GV
• Pull more Centrally
  – 45 to 90 degrees from intended direction
Anterior Capsule Vitrectorhexis; AC capsulectomy

- PHPV with scars
- Dense/Fibrous AC capsule
- Failed CCC
- Younger than 3 months of age
- If not comfortable with CCC
Anterior Vitrectorhexis

- Vitrector over capsule
- Start in Middle and enlarge
- 23 gauge for Both (same size)
- Corneal incisions: Watertight
Lensectomy

• Hydrodissection/ Hydrodelineation may be performed with caution
• Removal of lens cortex is required
  – Vigorous inflammatory response
  – Clean posterior surface of anterior capsule: VAO
Ant and Post Capsulectomies

- Tight Fit; **Clear Corneal Incision**
- Micro-forceps
- Vitreector:
  - Anterior capsulectomy: 150-300 cutting rate/Vacuum 300
  - Nucleus: Aspiration 30 Vacuum 100-500; IOP 30
  - Posterior capsulectomy and Vitrectomy: 1000 cutting rate/Vacuum 300
Posterior Capsule Management

- 100% opacify even with Acrylic materials
- Primary posterior capsulotomy vs Capsulectomy
- Leave PC intact age of 4; YAG later
- Problem: Recurrence is common after YAG
Posterior Capsule Management

• 4 yo or older
  – Leave PC intact : YAG

• < 4 yo
  – Posterior capsulotomy (PC Vitrectorhexis or PCC)

• Approach
  – Anterior approach after IOL placement
  – Pars plana approach after IOL placement
3.5 - 4.5 mm
IOL Choices

• MN60, SA60AT and SN60AT
  – Acrylic
  – One piece
  – 2.4 mm corneal incision
  – Flexible that Unfold slowly
IOL Placement with Open PC
Video of Surgery
PCC before IOL
Lueke Cannula AC maintainer
Post-Op management

- Intra-ocular injection: Triesence 4mg / Moxifloxacin
- Depot subconjunctival steroid / Moxifloxacin
- Topical steroids every 4X day for 1 week
- Taper over 3 weeks
- No IOL capture
- No Miostat /AT
- Eye Shield at Night X 4 weeks
  - Glasses during daytime.
YAG if Clinic procedure not possible
Post Op Management

• Long Term F/U
• Refraction,
  – CTL and Glasses
• Amblyopia Treatment
• Strabismus
• Nystagmus
Visual Rehab

- **Bilateral Aphakia:**
  - Glasses or CTL
  - < 4 yo: Overcorrect for intermediate distance
  - 4 yo or older: Bifocals

- **Unilateral Aphakia:**
  - CTL and later IOL if possible
Vision Outcomes

• Wide Range
• Unilateral cases: worse prognosis
• 6 yo outcome in various studies:
  – Unilateral: 20/200
  – Bilateral: 20/60
IATS Vision Outcome in Unilateral CE

• VA 20/160

• More Intraoperative complications with IOL (11% vs 28%)

• More adverse events by 4.5 yo f/u with IOL

• Additional Surgeries with IOL

• No benefits of IOL
Glaucoma

- IATS: 5 yo post op: 17% (10 to 20%)
- Risk Factors
  - SX before 4 weeks of age
  - PHPV
  - Microcornea <9mm
Suspect Glaucoma

- Photophobia with Cloudy cornea
- Agitation
- IOP
- Serial refractions
- Axial length (EUA)
- Evaluation of Optic Nerve
- Pachymetry
- EUA
Strabismus

- Most unilateral cataracts
- 70% of bilateral cataracts
- Amblyopia 90%
- Often small angle deviations
- Nystagmus often lead to late diagnosis
Points to Remember

- Pediatric Cataract Sx Not Equal to Adult Cataract Sx
- Family needs psychological support
- Good Aphakia better than bad IOL
- Visual Rehabilitation after Sx More Important
Points to Remember

• Keep the Procedure as Simple as Possible and use familiar instruments

• Have someone watch Your Posture!
Enjoy the Journey!

If you can’t fly, then run.
If you can’t run, then walk.
If you can’t walk, then crawl,
but by all means, keep moving.

- Martin Luther King Jr.