Congenital Anomalies in Children

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Objectives

1. To outline common pediatric congenital anomalies involving ocular alignment, motility, the anterior segment and the posterior segment

2. To review the important steps in diagnosing, treating and managing pediatric ocular congenital anomalies

3. To discuss red flags and vision or life-threatening features of various pediatric congenital anomalies
Anterior Segment Anomalies
Pediatric Lens Disorders

• Abnormalities:
  ◦ **Opacification** (Lamellar, nuclear, lenticous/ lentiglobus, polar, posterior subcapsular)
  ◦ Size (Microspherophakia)
  ◦ Shape (Spherophakia, coloboma)
  ◦ **Location** (Subluxed)
  ◦ Development (Persistent fetal vasculature/ PHPV)

• Congenital cataracts – 1:250 incidence
  • Usually >3mm = visually significant
Congenital Cataracts

(Pictures)
Unilateral Congenital Cataract - Etiology

- Ocular anomalies
  - Coloboma
  - Aniridia
  - Uveitis
  - Anterior segment dysgenesis
  - Posterior segment tumours

- Idiopathic

- Trauma – **R/O non-accidental injury**
Non-Accidental Injury

**ALWAYS BE ALERT TO POSSIBLE NON-ACCIDENTAL INJURY**

• A dilated exam is critical
  o White centered hemorrhages
  o Macular schesis
  o Vitrous hemorrhage
  o Disc edema

• Other findings include:
  o Altered level of consciousness
  o Bruises
  o Long bone / spiral fractures
  o Multiple injuries in various stages of healing
Bilateral Congenital Cataracts - Etiology

- Craniofacial
  - Hallerman-Streiff, Rubenstein-Taybi, Smith-Lemli-Opitz, Other
- Hereditary (autosomal dominant)
- Idiopathic
- Metabolic
  - Galactosemia, Fabry, Wilson, mannisidosis, diabetes
- Infection (TORCH)
  - Toxoplasmosis, Syphilis, Rubella, CMV, Varicella
- Poison (Toxic)
  - Corticosteroids, radiation
- Chromosomal abnormality
  - Trisomy 21 (Down), 18 (Edward), 13 (Patau), Other
- Renal
  - Lowe, Alport
- Musculoskeletal
  - Conradi, Albright, myotonic dystrophy
- Ocular abnormalities
  - Aniridia, anterior segment dysgenesis
Congenital Cataract Assessment - History

History:
- Pregnancy: infections?
- Medical history: growth and development
- Family history: cataracts
Congenital Cataract Assessment - Exam

- Examine family members

- Visual Function
  - < 2 mos – fixation reflex not developed (absence is not abnormal)
  - >3mos preverbal – look at fixation behaviour, preference, objection to occlusion
  - School age – aim for 20/50 or better for school, driving
  - Bilateral – look at behaviour
  - Unilateral – surgery if VA <20/50-70 with optical and amblyopia treatment

- Visually significant
  - Anterior capsular opacities occluding entire pupil (no RR)
  - Post/ Central opacities - >3mm diam
  - Late: (indicates optimal time for treatment past, but can still improve)
    - Strabismus – unilateral cat
    - Nystagmus – bilateral cat
  - Can observe patients who have significant RR around opacity OR have an opacity with clear areas within
Congenital Cataract - Workup

Unilateral – no lab workup

Bilateral
○ No investigations needed if +FHx OR parents have congenital lens opacities
○ Labs:
  ○ TORCH titer
  ○ VDRL
  ○ Urine for reducing substances, amino acids
  ○ Serum PO4-
  ○ Serum Ca2+
  ○ Red cell galactokinase level
  ○ Serum ferritin
○ Consults
  ○ Genetics
  ○ Metabolics
  ○ Developmental peds
** NOT ALL CATARACTS NEED SURGERY!! **

- Good cycloplegic and subjective refraction
- Amblyopia management with glasses +/- patching
- Phenylephrine prior to patching in unilateral cases
- Importance of follow up for vision monitoring
- Lensectomy surgery when appropriate
- Secondary intraocular lens (depends on age, history of uveitis, cause of cataract)
Pediatric Lens Disorders

• Long discussion with parents are critical
  • Prognosis
  • Surgery & postoperative course
  • Risks (aphakic glaucoma)
  • Visual rehabilitation (glasses, patching)
  • Realistic expectations
  • Systemic conditions
Ectopia Lentis

- Subluxation of the lens
- May be very subtle and only detectable on slit lamp examination when fully dilated
- Children may present with progressive myopia and increasing astigmatism

**History:**
- Family history of connective tissue diseases, Marfan Syndrome
- Medical history: developmental delay?
- Ocular history: trauma?

**Examination:**
- Visual acuity
- Slit lamp exam – Phacodonesis? Iridodonesis? Stretched or broken zonules?
- Cycloplegic refraction
Ectopia Lentis - Etiology

- trauma
- Marfan Syndrome
- Homocystinuria
- Sulfite Oxidase Deficiency

(pictures)
Systemic Associations with Ectopia Lentis

(pictures)
Management of Ectopia Lentis

• Amblyopia management
• Correct refractive correction

• Look at position of lens
  • Bisecting pupil or extremely high astigmatism causing amblyopia = surgical lensectomy
  • Completely subluxed lens in vitreous or out of visual axis = treat as an aphakic patient (no lensectomy)

• Secondary sutured or iris-claw lenses are possible after lensectomy

• Pediatrician or pediatric geneticist to manage systemic investigations (if required)
Posterior Segment Anomalies
Colobomas

- A coloboma is a malformation that occurs in utero when one part of the eye fails to form due to improper fusion of the optic fissure
- Colobomas can occur in isolation or with other genetic syndromes
- Colobomas can affect
  - Eyelid
  - Lens
  - Macula
  - Optic nerve
  - Uvea
Colobomas

(pictures)
- Eyelid
- Lens
- Macula
- Optic nerve
- Uvea
Systemic Conditions Associated with Colobomas

(pictures)

CHARGE, etc
Management of Choroidal Colobomas

- Referral to pediatrician or pediatric geneticist
- Long discussion with family regarding guarded visual prognosis
- Management of other associated ocular conditions
  - Cataracts
  - Microphthalmia
  - Increased risk of detached retina - retina is hypoplastic overlying chroidal colobomas
- Amblyopia management
- Refractive correction if needed
- Watch for development of sensory strabismus and nystagmus
Optic Disc Drusen

- Most common form of pseudopapilledema
  - Other causes: hyperopia, myelinated NFL, hyaloid traction on disc

- Etiology: Cause is axonal degeneration

- Elevated optic nerve on examination +/- blurred disc margins, isolated hemorrhage, VF defect
  - Possible grey or yellow-white disc discoloration

- Superficial drusen may create “lumpy” yellow disc elevation, “grape-like” clusters

- Bilateral in 2/3 of patients, buried drusen in 86%
  - Becomes more obvious with age (mean age at diagnosis 12.1 years)
Optic Disc Drusen

(pictures)
Papilledema

• Red flags on history:
  o Headaches, nausea, vomiting
  o “Whooshing” sound in ears
  o Worsening headache noted after change in position (bending down)
  o Neurologic symptoms
  o Lethargy, loss of consciousness

• Red Flags on Examination:
  o Decreased vision
  o Visual field defect
  o Blurred disc margins, disc hemorrhages
  o (Absent spontaneous venous pulsations – absent in 25% of normal)
Optic Disc Drusen

- Investigations:
  - B-Scan / CT scan
  - HVF 24-2 / Goldmann VF – may show arcuate defect
  - Possible MRI brain and neurology referral if increased ICP is suspected
Motility Disorders
Duane Retraction Syndrome

• Congenital absence of 6th cranial nerve nucleus, lateral rectus is supplied by 3rd nerve

• Type I: Abduction defect

• Type II: Adduction defect

• Type III: Abduction and Adduction defect

• Features:
  o Ocular alignment (orthophoric, esotropic or exotropic)
  o Face turn possible
  o Upshoot or downshoot of eye in adduction
  o Narrowing of palpebral fissure with adduction
Systemic Associations with Duane Syndrome

- Etiology: (pictures)
  - Isolated
  - Goldenhaar Syndrome
  - Klippel-Feil Syndrome
Duane Syndrome

• Has tendency to affect left eye > right eye
• Affects girls > boys
• Goal: Maintain binocular single vision in primary position
• Surgical intervention offered when:
  o Significant anomalous head position
  o Overshoot or undershoot
  o Esotropia or exotropia in primary gaze
Möbius Syndrome

• Congenital developmental disorder affecting brainstem
  o 6th and 7th nerve palsies (lack of facial expressions, unilateral or bilateral abduction defects)
    o Feeding difficulties
  o Oral: Micrognathia, high arched palate, cleft palate, missing/ misaligned teeth
  o Hypotonia, digit anomalies
  o Hearing loss
  o Possible developmental delay (gross motor)
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Mobius Syndrome
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Overview

• Anterior Segment Anomalies
  • Pediatric Cataracts
  • Ectopia Lentis

• Posterior Segment Anomalies
  o Choroidal coloboma
  o Optic disc drusen

• Motility Disorders
  o Duane Retraction Syndrome
  o Möbius Syndrome
References

Hoyt
AAPOS