Anterior Segment Disease in Pediatric Patients
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Disclosure

Faculty, Planners, reviewers and others who control educational content have disclosed they do not have any relevant financial relationships with commercial interests

Objective

- Identify and treat various pediatric anterior segment diseases with an effective treatment plan
Anterior Segment Dysgenesis

- Developmental arrest or incomplete migration of mesodermal or neural crest tissues
- External examination in the newborn period may reveal abnormalities
- For milder asymptomatic abnormalities, diagnosis may be made at the time of a routine examination.
- Visual deficits are variable.

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Posterior Embryotoxon

Axenfeld's Anomaly
Rieger's Anomaly

- combination of Rieger's anomaly and systemic abnormalities, which include
  - microdontia and a flat nasal bridge with maxillary hypoplasia

Rieger's Syndrome

- combination of Rieger's anomaly and systemic abnormalities, which include
  - microdontia and a flat nasal bridge with maxillary hypoplasia

Posterior Keratoconus

- This developmental defect is usually unilateral.
- There is a focal indentation of the posterior cornea with overlying stromal scarring
- The vision is usually not greatly affected.
Peters' Anomaly
- Central corneal opacity, usually with strands of adherent iris.
- Lens may have an anterior polar cataract or keratolenticular adhesion.
- Glaucoma is common.
- Associated ocular abnormalities include microphthalmos, persistent hyperplastic primary vitreous, and retinal dysplasia.
- Systemic abnormalities may be found: mental retardation, urogenital, craniofacial and skeletal abnormalities.

Pathology
- Focal absence of Descemet's membrane and endothelium in the area of central opacity.

Treatment and Management
- Secondary complications such as glaucoma require ongoing care and follow-up.
- May require penetrating keratoplasty.
**Scleracornea**

- Partial or complete replacement of clear cornea with white vascularized scleral tissue. Anterior chamber angle abnormalities and glaucoma common, as are cornea plana and high-level refractive errors.
- Associated with deafness, cerebellar and skeletal abnormalities, trisomy 13 and 18.
- Penetrating keratoplasty may be required if central cornea is opaque.

**Other Causes of Corneal Opacity**

- Tears in Descemet's membrane: birth trauma, congenital glaucoma.
- Ulcers: HSV, congenital rubella, neurotrophic corneas. Metabolic conditions: bilateral diffuse opacity, usually systemic abnormalities.
- Endothelial dystrophies: bilateral corneal edema.
- Dermoids: unilateral or bilateral, white vascularized mass that may have hair and lipid.
Microcornea

- Cornea horizontal diameter less than 11 mm
- Undergrowth of the optic cup during development.
- Autosomal dominant
- Associated with Weill-Marchesani, Ehlers-Danlos, and Rieger syndromes and Norrie’s disease

Congenital Glaucoma

- Developmental abnormalities of the AC angle lead to obstruction of aqueous outflow and subsequent glaucoma early in life
- Isolated congenital glaucoma is seen in approximately 1 per 30,000 live births.
- Isolated trabeculodysgenesis is seen in 50% of patients with congenital glaucoma.
- Cases are 90% sporadic, 10% autosomal recessive. Bilateral in 75%.
- Male patients, 65%; female patients, 35%.

Congenital Glaucoma

- Excessive tearing, ocular pain, photophobia, irritability, crying (especially when exposed to light).
- Blepharospasm.
- Corneal edema
- Corneal stromal opacity and scarring attributable to Haab’s striae (horizontal breaks in Descemet's membrane)
- Glaucomatous optic nerve atrophy and visual field loss.
- Enlarged eye (buphthalmos).
Congenital Glaucoma

- Flat iris insertion: The iris inserts into the TM anterior to the scleral spur.
- Wraparound iris: The iris is concave peripherally but sweeps up and over the TM with a "Barkan's membrane."
- Glaucoma progresses and the eye enlarges, causing buphthalmos and blindness.
- Difficult disease to manage; despite surgical treatment, many eyes continue to have glaucomatous damage.
- Some patients have continued maturation of angle structures leading to a halt in the progression of the disease.
- Prognosis improves the later the disease presents in life.

Hoskins' anatomic classification of developmental glaucomas

1. Isolated trabeculodysgenesis (malformation of trabecular meshwork in the absence of iris or cornea abnormalities)
   A. Flat iris insertion
      1. Anterior insertion
      2. Posterior insertion
      3. Mixed insertion
   B. Iridotrabeclodysgenesis (trabeclodysgenesis with iris abnormalities)
      A. Anterior stromal defects
         1. Hypoplasia
         2. Hypertrophy
      B. Anomalous iris vessels
         1. Persistence of tunica vasculosa lentis
         2. Anomalous superficial vessels
      C. Structural anomalies
         1. Holes
         2. Colobomas
         3. Aniridia
   C. Corneotrabeclodysgenesis (usually has associated iris anomalies)
      A. Peripheral
      B. Midperipheral
      C. Central
Congenital Glaucoma

- horizontal diameter greater than 13 mm corneas
- overgrowth of the optic cup during development
- usually X-linked recessive; 90% of affected patients are males
- associated with Marfan syndrome, craniosynostosis, and ichthyosis
- corneas in this disorder are often steep, and the patients are usually myopic.

Megalocornea

- horizontal diameter greater than 13 mm corneas
- overgrowth of the optic cup during development
- usually X-linked recessive; 90% of affected patients are males
- associated with Marfan syndrome, craniosynostosis, and ichthyosis
- corneas in this disorder are often steep, and the patients are usually myopic.

Other Ocular Size Conditions

- Nanophthalmos: small, anatomically normal globe.
- Microphthalmos: small globe with multiple abnormalities.
- Anterior megalophthalmos: large anterior segment with multiple abnormalities.
- Buphthalmos: large globe attributable to infantile glaucoma.
Cornea Plana

- Congenitally flat cornea, with a radius of curvature of less than 40 diopters (D), but commonly 30 to 33 D (similar to the scleral radius of curvature).
- Autosomal dominant or recessive. Occurs because of developmental arrest of the growth in utero that leads to the usual increase of corneal curvature relative to that of the sclera.
- High-level hyperopia.
- May also have glaucoma, indistinct limbus, microcornea, sclerocornea, and other ocular abnormalities.

Microphthalmos with cyst

- The eye is small and malformed, and a cyst is contiguous with the globe.
- The cyst is formed from proliferating retina.
- This abnormality occurs when the embryonic (choroidal) fissure fails to close.
Corneal dermoid

- collection of ectodermal elements such as sweat glands, hair follicles, and sebaceous glands on the corneal surface.
- These lesions are well circumscribed and elevated.
- In this case, abnormal lashes from the lower lid are rubbing on the cornea; however, in some cases, lashes are found exiting from the substance of the dermoid.

Limbal dermoid

Lipodermoid

- These tumors are composed primarily of fatty tissue and are usually located beneath the conjunctiva on the lateral aspect of the globe.
- The posterior aspect of the lesion often extends far posteriorly and cannot be identified in this patient.
- These tumors can be removed for cosmetic reasons, but it is important to excise only the anterior aspect of the tumor and not to attempt excision of the entire lesion.
Important facts

- 33% - idiopathic - may be unilateral or bilateral
- 33% - inherited - usually bilateral
- 33% - associated with systemic disease - usually bilateral
- Other ocular anomalies present in 50%
Classification of congenital cataract

Anterior polar  Posterior polar  Coronary  Cortical spoke-like

Lamellar  Central pulverulent  Sutural  Focal dots

Etiology

Unilateral vs Bilateral
Anterior polar cataract
May be dominant inheritance

- Unilateral, sporadic
- Small, less than 2 mm
- Dense, discrete
- Usually nonprogressive
- Not associated with a systemic disease

Anterior polar cataract
Capsule
Pyramid

With persistent pupillary membrane
With Peters anomaly

Posterior polar cataract

- Persistent hyaloid remnants
- Posterior lenticulitis
- Persistent hyperplastic primary vitreous
**Persistant Hyperplastic Primary Vitreous**

- Sporadic inheritance
- Associated with microcornea
- Fibrovascular stalk from posterior capsule to optic nerve
- Ciliary processes are pulled towards the center of the pupil

**Posterior Lenticous**

- Sporadic inheritance
- "Oil droplet" appearance in central red reflex
- Axial in location
Coronary (supranuclear) cataract

- Round opacities in deep cortex
- Surrounded nucleus like a crown

Workup of Unilateral Cataract

- History
- Slit lamp exam (diagnose specific morphological features i.e. PHPV and posterior lenticous)
- TORCHES titers
Cortical spoke-like cataract

Systemic associations
- Fabry disease
- Mannosidosis

Fabry's Disease
- Characterized by small dark purple skin lesions
- Symptoms include diffuse pain and malaise
- Other ocular findings include whorl-like corneal opacities, conjunctival and retinal vessel tortuosity, periorbital and retinal edema
- Urine sediments shows "Maltese Cross" figures
Lamellar cataract

- Central disc shaped opacity and radial spokes
- Associated with Wilson's disease
- Causes minimal visual impairment

Systemic associations:
- Galactosaemia
- Hypoglycaemia
- Hypocalcaemia

Lamellar cataract

- Round central shell-like opacity surrounding clear nucleus
- May have components

Usually dominant inheritance
Oil - Droplet Cataract

- Faint opacity in posterior cortex
- Associated with galactosemia
- May be reversed with treatment

Galactosemia

- Autosomal recessive
- Cataracts can be the sole manifestation of the disease
- Reversible with a galactose free diet
- Urine should be tested for reducing substances
Central pulverulent cataract
Dominant inheritance

- Spheroidal opacity within nucleus
- Relatively clear centre
- Non-progressive
Sutural cataract

- Usually X-linked inheritance
- Opacity follows shape of Y suture

Focal dot opacities

- Blue dot cortical opacities
- Common and innocuous
- May co-exist with other opacities

Causes of cataract in healthy neonate

- Hereditary (usually dominant)
- Idiopathic
- With ocular anomalies
  - PHPV
  - Aniridia
  - Coloboma
  - Microphthalmos
  - Buphthalmos
Causes of cataract in unwell neonate

**Intrauterine infections**
- Rubella
- Toxoplasmosis
- Cytomegalovirus
- Varicella

**Metabolic disorders**
- Galactosaemia
- Hypoglycaemia
- Hypocalcaemia
- Lowe syndrome

Lowe’s Syndrome

- X-linked recessive
- Characteristic facies with frontal bossing and chubby cheeks
- Cataracts present at birth
- Associated glaucoma and a miotic pupil
- Renal tubular dysfunction is seen early
- Urine should be tested for amino acids
Workup of Bilateral Cataracts

- Family history
- TORCHES
- Urine for reducing substance
- Focused examination in coordination with a pediatrician