10.6.1. Childhood Glaucoma (I)

Primary Congenital Glaucoma (PCG)

epidemiology
- PCG accounts for majority of primary pediatric glaucomas
- 70% bilateral
- 75% diagnosed within the first year of life
- 65% M
- ethnicity
  - 1 in 1250 — Slovakian Roms
  - 1 in 18,500 — Great Britain
- consanguinity greatly increases the risk
- without a family history of PCG, an affected parent has a 2% chance of having a child with PCG
- newborn PCG — 25%

pathogenesis
- increased outflow resistance through the trabecular meshwork
- developmental anomaly of the neural crest–derived tissue of the anterior chamber angle
  - dysgenesis and compression of the trabecular meshwork
  - anterior insertion of the iris root

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clinical presentation
- classic triad
  - epiphora
  - photophobia
  - blepharospasm
- increased corneal diameter and enlargement of the globe (buphthalmos)
  - until age 3, elevated IOP causes the cornea to stretch
- Haab striae
  - breaks in Descemet membrane
  - corneal edema
  - corneal opacification
- >age 3 years
  - cornea does not continue to enlarge
  - scleral stretching causes progressive myopia

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worse prognosis
- newborn PCG
  - 1/2 patients with newborn PCG progress to legal blindness
  - glaucoma diagnosed >1 year of age
  - corneal diameter >14 mm
- prognosis best for patients whose glaucoma is diagnosed between 3 and 12 months
  - vast majority respond to angle surgery

differential diagnosis
- PCG should be considered in the differential diagnosis of any child who presents with tearing
  - See Table 6-3
  - milder cases have fewer signs and symptoms and may be misdiagnosed as nasolacrimal duct obstruction

treatment
- surgical intervention
- medical therapy
  - limited long-term value
  - may be used to temporize or reduce corneal edema to improve visualization during surgery

Juvenile Open-Angle Glaucoma

- 4-35 years
- autosomal dominant
  - most cases
  - GLC1A locus
- TIGR/MYOC gene
  - trabecular meshwork inducible glucocorticoid response/myocillin
- it does not usually cause corneal enlargement or Haab striae
- progressive myopia
  - may continue to develop
- angles appear normal
- treatment
  - medical therapy
    - usually unsuccessful
  - angle procedures
    - may be helpful in select cases
  - trabeculectomy or implantation of a glaucoma drainage device

Classification

Primary
- developmental glaucomas with congenital anomalies of the filtration angle — See Table 6-1
- classic features
  - enlarged and/or cloudy corneas
  - Haab striae
  - elevated IOP
- types
  - congenital glaucoma (congenital open-angle glaucoma)
    - newborn PCG — at birth or <1 month of age — 25%
    - infantile PCG — <2 years of life
    - late-diagnosed PCG — >2 years of age
    - 75% diagnosed within the first year of life
  - juvenile open-angle glaucoma — later in childhood or in early adulthood — 4-35 years
  - glaucoma associated with systemic diseases
    - chromosomal disorders
    - connective tissue disorders (ie, Marfan and Stickler syndromes)
    - phakomatoses
  - glaucoma associated with other ocular anomalies
    - aniridia
    - Peters anomaly
    - Axenfeld-Rieger syndrome

Secondary
- trauma
- intraocular neoplasms
- inflammation
- lens-induced disorders
- surgical interventions
- angle closure
- infection
- neovascularization
- corticosteroid use
- elevated episcleral venous pressure
- See Table 6-2

Genetics

- genetic testing should be considered for
  - parents of pediatric glaucoma patients
  - adults whose glaucoma had its onset in childhood or early adulthood
- PCG
  - sporadic — most cases
  - familial — 10%–40%
    - autosomal recessive
      - incomplete or variable penetrance
      - 3 genetic loci
        - GLC3A — CYP1B1 gene — drug-metabolizing enzyme cytochrome P450, family 1, subfamily B, polypeptide 1
        - GLC3B
        - GLC3C — LTBP2 gene — latent transforming growth factor beta-binding protein 2
- juvenile open-angle glaucoma
  - autosomal dominant — GLC1A locus — TIGR/MYOC gene — trabecular meshwork inducible glucocorticoid response/myocillin
  - sporadic — occasional
- other ocular anomalies
  - Axenfeld-Rieger syndrome — autosomal dominant
    - in contrast to PCG
      - PITX2 — at 4q25
      - FOXC1 — at 6p25
      - variable interaction between these 2 genes may underlie the diverse phenotypic expression
  - Peters anomaly
    - PITX2
    - FOXC1 — AD
    - PAX6
    - CYP1B1
    - most cases are sporadic
  - aniridia
    - autosomal dominant — 2/3 — PAX6 gene
    - sporadic — 1/3

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