Infantile and Childhood Glaucoma

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Infantile Glaucoma

- **Primary** – PCG isolated developmental abnormality of anterior chamber angle

- **Secondary** – aqueous outflow reduced due to other congenital or acquired disease
Infantile Glaucoma: Classification

**Primary Congenital Glaucoma (PCG)**
- Isolated trabeculodysgenesis
- Bilateral in approx 75%
- Worse prognosis if present as neonate
Primary Congenital Glaucoma: Pathogenesis

- Traditionally described as due to Barkan’s membrane
- Developmental arrest of neural crest derived tissues in third trimester
- Compacted poorly differentiated abnormal trabecular meshwork
Infantile Congenital Glaucoma: Epidemiology

- **BIG eye study** (Papadopoulos et al IOVS 2007)
  - 99 cases
  - 47 PCG
  - 52 Secondary
  - Annual incidence of PCG in GB was 5.41 per 100,000 live births (1/18500)
  - Incidence in South Asian origin infants = 9x Caucasians.
Infantile Congenital Glaucoma
previous epidemiology

- 1/10,000 - 20,000 Western Europe
- 1/8,200 in Palestinian Arabs
- 1/1,250 in Slovakian gypsies
- Europe/USA/Japan M:F ratio = 2.5:1
- Equal sex distribution elsewhere
Primary Congenital Glaucoma: Genetics

- Most cases in UK “sporadic”
- Most prob AR, some AD
- Biallelic mutation of CYP1B1 gene (assoc with GLC3A) – encodes for cytochrome P450 1B1
- GLC3A (on 2p21) approx 90% familial cases
- LTBP2 = most of rest
- GLC3B on 1p36 also linked
- GLC3C on 14q24.3 linked
Infantile Glaucoma: Clinical Features

- Damage to endothelium results in localised or diffuse corneal oedema - photophobia and lacrimation
Infantile Glaucoma: Clinical Features

- Generalised ocular enlargement
- Corneal and scleral immaturity/plasticity
- Ceases by age 2 to 3
Infantile Glaucoma: Clinical Features

- Splits in Descemet’s membrane - ‘Haab’s striae’
- Horizontal and linear centrally
- Concentric with limbus peripherally
- Rare if >18 months or HCD<12.5mm)
Infantile Glaucoma: Clinical Features

- As IOP reduces, oedema clears – periphery first

- Photophobia often persists despite low IOP due to Haab's striae
Infantile Glaucoma: Clinical Features

Optic Nerve Cupping

- Morphological changes similar to adults
- Important both diagnostically and in evaluating progression
- Occurs early and rapidly
- Often reversible if IOP reduction before optic atrophy – but RNFL thinning on OCT may persist
Initial management- EUA

• pre-intubation IOPs
  - results can be variable – must **never** be only method of assessing glaucoma control
Initial management- EUA

• Record
  • Horizontal corneal diameters (should be 11mm or less in neonate, 12 or less in 1 year old)
  • Descemet’s splits
  • Gonioscopy
  • Pachymetry (CCT)
  • Axial length
Initial management - EUA

• Posterior Segment
  • Disc appearances (CDR >0.3 in an infant or >0.5 in child suspicious)
  • Other fundal pathology

• Retinoscopy

• B scan U/S **essential** if poor view of fundus

• UBM may be useful
Treatment – PCG

- Medical therapy is a **temporising**
  measure/adjunctive
- Care re side-effects
- Carbonic anhydrase inhibitors (oral/topical)
- Topical B-Blockers
- Pilocarpine
- Prostaglandin analogues
- [Brimonidine](https://en.wikipedia.org/wiki/Brimonidine) contraindicated
Goniotomy

- **Treatment of choice** in PCG
- Minimal trauma, conjunctiva not violated
- Can be repeated
- Needs good view of angle (pilocarpine, glycerol, strip epithelium)
- Expertise required
- Long term success up to 90%
- 60% in BIG eye study
- ?? Future role for trabectome
**Trabeculotomy**

- When cornea opaque
- Scleral flap & trabeculotomes - localisation of Schlemm’s canal often difficult
- Avoid upper quadrants
- Can use nylon loop to deroof 360°
- Hyphaema, endothelial damage, bleb formation, iris and retinal damage (360°)
- Illuminated catheter an option
- Combined Trabeculotomy- Trabeculectomy – technically more difficult but advantageous in complex cases

Trabeculodysgenesis

• **Amblyopia management**
  - Refraction
  - Occlusion

• **Rare sequelae**
  - keratopathy
  - retinal detachment
  - ectopia lentis
  - cataract

• **Lifelong follow up**

Relapse following goniotomy for congenital glaucoma due to trabecular dysgenesis
Differential Diagnosis

• Obstetric (Forceps) Damage
Differential Diagnosis

Large Eyes

- Megalocornea – usually X-linked
- Congenital high myopia
Secondary Congenital Glaucoma: Anterior Segment Dysgenesis Spectrum (ASD)

- **Axenfeld anomaly** – posterior embryotoxon with attached iris strands
- **Rieger anomaly** – as above plus abnormal iris, corectopia, polycoria
- RIEG/PITX2(4q25)
- RIEG2(13q14)
- FOXC1 and PAX6
- Very high glaucoma risk
Secondary Congenital Glaucoma: Anterior Segment Dysgenesis Spectrum (ASD)

Peters anomaly

- Central corneal opacity, defects in stroma and DM, iris strands, keratolenticular attachment
- Usually bilateral, sporadic
- Very high glaucoma risk
- PAX6 (11p13)
- RIEG/PITX2
- FOXC1
Secondary Congenital Glaucoma: Aniridia

- Bilateral iris absence/hypoplasia
- Panocular disorder PAX6(11p13)
- Glaucoma in up to 75%
- May be congenital but usually presents later in childhood/early adulthood
- Surgical management contentious
Infant with aphakic glaucoma

- Microphthalmic (HCD 9mm)
  AL – 17.5mm
- Microcoria/abnormal iris
- Dense nuclear cataracts
- Normal IOPs pre-op
- Bilateral lensectomy at 6/52
- Hazy corneas by 17 weeks
- IOPs 22mmHg
- HCD 10.5mm, AL – 19.0mm

- Management?