Pediatric Retina Interesting Cases

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Case presentation
- 3 mo old child - high myopia
- -18 OD, -16 OS
- Cleft palate, micrognathia
- Pierre-Robin sequence
- Refractive correction given
- At age 2, retinal hole right eye – lasered
- 1 year later – detached OD, multiple vitreous condensations OS
- How should we manage her?

Stickler syndrome
Progressive, autosomal dominant connective tissue (collagen) disorder

Spondyloepiphyseal dysplasia
Midline palatal clefting
Facial dysmorphism, and hearing loss

Megaglobus and congenital myopia
Pathognomonic vitreous strands / condensations
Oral giant retinal tear and detachment

73% develop RD, >50% have bilateral RD

Retinal Detachment in Children
- Incidence ≤ 1 per 100,000 annual
- Trauma 40% (vs 10% in adults) (adults PVD)
- 70% male
- 35 -56% congenital anomalies
- 30% with hx of prior cataract surgery

Presentation
- Slower onset than adults
- Cognitive function – may not report changes
- Frequently present when macula of second eye is involved (30% with bilateral RD)
- Macula off RD and Proliferative vitreoretinopathy more common (20 – 60%)
- If bilateral, worse eye inoperable 60% of the time
- Giant retinal tears more common, 15–20%

Retinal Detachment
### Etiology

- Trauma
- Prior ocular surgery
- High myopia
- Sticklers syndrome
- ROP
- X-linked retinoschisis
- Persistent fetal vasculature
- Norrie’s disease

### Examination

- Visual acuity
- Red reflex abnormal
- Nystagmus, Alignment
- Intraocular pressure – hypotony with chronic RD
- Exam under anesthesia
- Ultrasound – RD, calcification (RB)

### Surgical Repair

- Relief of traction on the causative retinal defects
- Reapproximation of neurosensory retina to the retinal pigment epithelium (RPE)
- Creation of a retina to RPE adhesion

### Anatomy

- Smaller size
- Relatively large lens in comparison
- Adherent vitreous

### Scleral buckle

- Helps relieve peripheral vitreous traction
- Can be used alone if no PVR
- Can be used in combination with vitrectomy
- Spares the lens

### Vitrectomy in children

- Posterior hyaloid is resistant to peeling
- Lensectomy at the time of the repair is common
- Challenges of postoperative positioning - silicone oil is preferred to long-acting gas for tamponade
- Membrane peeling often required – use staining agents
Small Gauge Vitrectomy Technique

- 23 and 25 gauge instruments
- Trans-conjunctival pars plana trocars
- Decreased need to suture sclerotomies
- Eye is pressurized throughout surgery

Pars Plana Lensectomy

- Port is closer to tip of vitrector
- Easier access to surgical planes
- Particular advantage in resecting / segmenting preretinal fibrovascular bands

Subretinal bands of PVR

Staining of Membranes/ILM

Subretinal fluid drain, fluid-gas exchange, laser
Outcome

- Final anatomic success rate of 70 – 80%, 60% for age 11 and younger
- Single surgery success of 50 – 80%
- Only 30–40% of patients reach a final acuity of 20/200–20/400
- Due in part to the increased proportion of chronic detachments as well as amblyopia

Scleral buckle + vitrectomy + laser

Our patient - Prevention of RD

- In the largest cohort of Stickler syndrome patients, Prophylactic Cryotherapy is safe and markedly reduces the rate of retinal detachment
- High potential for myopia
- Can reduce this with use of laser, similar to findings of ROP studies

X-linked Retinoschisis

- Peripheral and Foveal Schisis

X-linked Retinoschisis

- Progressive, bilateral disease
- Prevalence 1 in 10,000 males
- Mutation in a protein involved in retinal cellular adhesion
- Radial streaks in macula due to foveal schisis (nearly 100%)
- Peripheral splitting of retina – temporal most common
- Giant retinal tears common, 15–20%

Prognosis

- Early, mild visual impairment
  - 20/30 to 20/60
- Later, more marked impairment
  - 20/30 to 20/400
- Average is 20/70 by mid adulthood
- Vision stable after age 20
- Scotomas, metamorphopsias
Management of Schisis RD

- include performing an inner wall retinectomy or dissecting the posterior hyaloid off the inner retinal wall
- pre-operative intravitreal autologous plasmin or microplasmin may assist
- Staining of hyaloid to assist in removal

Case 2

20/60

20/100

Fishman et al, 2006
Dorzolamide 2% tid
8 patients, improved cysts in 1 mo in 5/8, improved va

Persistent Fetal Vasculature

19 day old boy referred by pediatrician for absent red reflex
Does not follow objects or look at faces
Born at 38 weeks. Birth weight: 7 lbs 8 ounces.
Exam

VA: No response to transilluminator

Slit lamp exam: opaque posterior lenses with no view of fundus

Fundus exam: no view

Norrie Disease

- X-linked recessive: Deletion of NDP gene
- Ocular: Bilateral retinal dysplasia and proliferative changes in retina and vitreous resulting in retinal detachment and progressive atrophy of eyes
- Systemic: normal cognitive development until 2 years of age, then lose skills
- 1/3 develop hearing loss
- seizures, microcephaly, hypertensive crises, hypogonadism, and limb anomalies

Surgery

- Lensctomy, OD
- Vitrectomy with removal of posterior capsule fibrous plaque
- Posterior fibrous stalk extending from optic disc to posterior lens and iris causing traction. Funnel RD
- Dysplastic retina. Abnormal choroid with pigmented deposits.
- Extensive membrane peel and viscoelastic were unable to flatten retina. Eye closed.

Vascular disease
Case presentation

- 18 yo caucasian male
- Reduced vision x 2 months
- VA 20/50
Von Hippel Lindau

- Short arm of chromosome 3 (3p25)
- Autosomal dominant inheritance
- Tumor suppressor gene
- VHL protein suppresses VEGF production

Von Hippel Lindau

- Hemangioblastomas
  - Retina (most frequent and earliest manifestation of VHL disease)
  - CNS (most often cerebellum)
  - Pancreas, liver, epididymis
- Malignant cancers
  - Kidneys (clear cell renal cell carcinoma)
  - Adrenal glands (pheochromocytoma)

Management: laser and avastin injection
Case - 5 yo boy failed school screening

8 wks post Rx
VA 20/30

Etiology
- Retinal vascular anomaly
  - Telangiectatic and aneurysmal vessels
  - Venous dilation
  - Capillary dropout
  - Incompetence of retinal vessels
- Not hereditary
- Not infectious

Clinical Presentation
- Painless
- Males 3:1 Females
- No ethnic predeliction
- Unilateral in 80%
- Bimodal distribution
  - Age 4 to 10, age 30 to 40

Coats disease - 8 yo boy with 20/20 OU
Treatment

- Indicated if exudate threatens vision or may result in retinal detachment
- Photocoagulation
  - Treat vessels directly with large spots, moderate intensity
  - Sectoral PRP

Fibrosis may limit visual outcome

12 yo female, 20/25 OD, 20/20 OS

Age 14, progressive decrease in VA over 2 mo - 20/70 OD
Optic Nerve Pit with serous retinal detachment

Optic nerve pit

- 1 in 10,000 eyes
- Male = Female
- 10 – 15% bilateral
- 70% on temporal side of disc
- Fluid enters through the pit and between retinal layers, creating a schisis
- May develop a secondary retinal detachment

Natural History

- 15 patients with ON pit and serous detachment followed for 9 years
- Most present with 20/40 – 20/60
- Each lost 3 or more lines within 6 months
- 80% lost vision to 20/200 or worse
- May develop lamellar macular hole
- Other reports – 20% may resolve spontaneously

Anatomy of an Optic Pit

Treatment Options

- Laser barricade
- Pneumatic retinopexy +/- laser
- Vitrectomy (creation of PVD) +/- fluid/gas exchange +/- laser

21 yo female with ON pit
VA 20/40
Laser barricade
Pediatric Retina – some of our most challenging, interesting, and rewarding patients

Questions?