10 Pediatric Retinal Diseases you don’t want to miss!

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Retinal Update
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Top 10
- ROP
- Retinoblastoma
- Coats disease
- Persistent fetal vasculature
- Familial Exudative Vitreoretinopathy
- Sticklers
- Retinal dystrophies
- Pigmented lesions
- Pars Planitis
- Trauma related
- Zebras

Scope of the Problem: USA
- #1 cause of blindness in children in the US
- ≈500,000 infants/year born premature
- Occurs in smaller and younger infants
  - GA of 23 weeks or less – 75% with ROP
  - GA of 24 weeks – 70%
  - GA of 25 weeks – 50%
- Severe ROP is more common (AP-ROP)
  - 500-700 babies a year with severe visual loss

Retinopathy of Prematurity
(ROP)

- Aberrant retinal development
  - Arrest of retinal vascularization
  - Co-development of retina and vasculature
  - Severity:
    - Gestational age
    - Birth weight
    - Systemic health
    - Genetic predisposition

Aggressive, Posterior ROP (AP-ROP)
- Low BW, early GA
- Zone 1 disease
- Vascular activity with high VEGF levels
- Increases risk of RD
Late Reactivation

Children s/p anti-VEGF can have extensive areas of abnormal retina

Develop late RDs – years after previous treatment

Active NV

Laser vs Avastin – Zone 1

PMA 56 weeks
Anti VEGF and laser may not be enough

Stage 5

ROP
- Late complication- pediatric RD
- Take a good history
  - Born premature, myopia, h/o treatment
- Can present with Total RD-Leukocoria

Retinoblastoma
- Leukocoria
- Elevated retinal mass, calcifications
- RB1 - tumor suppressor gene
- Unilateral or bilateral
- Age of presentation – less than 5
- Intraarterial therapy-melphalan
- Survival rate, secondary cancers
Elevated retinal mass s/p intraarterial therapy

Retinoblastoma
- Treatment: Intraarterial therapy
  - Enucleation, Chemotherapy, External beam radiation, local therapy

Survival rates
- 5 year survival rate – 95%
- Eye preservation – 70-80%

Intraarterial therapy

Coats’ Disease
Coats’ Disease
- Exudative Retinopathy
- 2/3 present before 10 years of age
- 90% males, 90% unilateral

Coats’ disease
- Treatment
  - laser ablation and anti-VEGF injection
  - Visual prognosis dependent on VA at time of diagnosis
  - 20% with vision better than 20/200
  - Vision limited by macular exudate

6 year old with 20/30 VA

Residual macular exudate limits vision

Current Therapeutic Approach
- Scleral Buckle, external drainage, cryo
- Surgical intervention can prevent phthisis, but poor visual prognosis
Persistent Fetal Vasculature Syndrome

- Failed regression of hyaloid system
  - Unilateral
  - Previously known as PHPV
- Broad spectrum of presentation
  - Mittendorf dot
  - Bergmeister papilla
  - Persistent TVL
  - Persistent hyaloid artery

Requires surgical intervention

FEVR

- Familial Exudative Vitreoretinopathy
- Mutations in the NDP, FZD4, LRP5, and TSPAN12 genes
- Variability of disease severity in family members with same mutation
- Typically AD

Unexplained visual loss

- 14 year old male presents with visual loss for 6 months – thought he needed an update of contact lenses
- No health issues
- VA 20/30 OD, 20/100 OS
- “Possible ERM OS”
Familial Exudative Vitreoretinopathy

- Vitreoretinal dystrophy
  - Premature arrest of peripheral vascularization
- Macular dragging and knife-like folds
  - Full-term infants
  - 85% bilateral 15% unilateral
  - Variable tempo & progression
  - Chronic, lifelong disease
- Hereditary
  - Autosomal dominant, X-linked, AR
FEVR

Treatment

- Performed indirect laser to areas of avascular retina OU
- Despite laser – progressive macular distortion OS
- PPV performed OS

POM #2 s/p ppv/MP

Progression of OD

POW #2 s/p SB/PPV/MP/Sf6

**Early detection can result in good visual prognosis**
FEVR
- Macular distortion
- Bilateral but asymmetric
- Present at any age
- Check family members (50% transmission)

Stickler and Stickler-like Vitreoretinopaties

Sticklers
- Most common cause of inherited RD
  - Myopic
  - Craniofacial Abnl
  - Peripheral lattice
  - Posterior tears
  - Giant retinal tears
  - Early cataract

Sticklers
- Type I – Type 2 Collagen (Collagen 2A1)
  - Worse visual prognosis
- Type II – Type 11 Collagen (Collagen 11A1)
- Type III – Collagen 11A2 – no ocular sx
- High risk of RD!!! What do we do with these children??
Natural History Data

- Known COL2A mutations
  - Group 1 – observation, no laser
  - Group 2 – prophylactic treatment
  - Group 3 – RD one eye, treatment other eye

- 211 patients
- Mean F/U 11.5 years


Natural history

- Group 1 (control group)
  - 73% RD
  - 48% Bilateral

- Group 2 (peripheral treatment)
  - 8% RD
  - 0% Bilateral

- Group 3 (RD one eye, treatment other)
  - 10% RD

Retinal Dystrophies

- Retinitis Pigmentosa
- Stargardts

Observation is not a good option!!

- Genetic defect, high myopia, peripheral pathology – refer for evaluation

Unexplained visual loss

8 year old male with 20/40 VA OU

“Can’t see at night”
Maternal cousin “blind”
“Normal eye exam”: 1 year ago
OCT – small island of central photoreceptors
10 year old brother  20/30 OU

“Can’t see at night”  
Maternal cousin  “blind”  
“Normal eye exam” - 1 yr ago

OCT – small island of central photoreceptors

Retinitis Pigmentosa
- Loss of primarily rod photoreceptors
- Night blindness and peripheral vision loss
- ERG, VF, FA, OCT
- Prevalence 1:5000
- Vit A, DHA, Lutein – Ophthalmic Nutrition
- No Vitamin E
- Clinical trials

Stargardts disease
- Mutations involving the *ABCA4* gene
- Defect in Vitamin A metabolism
- Autosomal recessive
- FDA approved clinical trials

Stargardt disease
- Decreased vision, scotoma, decreased dark adaption
- 20/20-20/400
- Variable presentation - prognosis based on age of onset
Pigmented Lesions

- Combined hamartoma of RPE and retina
- CHRPE- congenital hypertrophy of RPE
- Bear tracking
- Choroidal Nevus - Choroidal Melanoma

Background

- Rare, benign lesions
- Histologically - disorganized, elevated pigmented mass involving the RPE, retina and vitreous
- Glial, vascular, pigment epithelial
- Can be thickened gray-white retinal and preretinal tissue - contraction of the inner surface
- Preretinal, intraretinal and subretinal components

POY #1
Pigmented Lesions

CHRPE
- Flat with lacunae
- Low risk of malignant conversion
- Monitor with color photography

CHRPE bear track configuration
- Concern for Familial Adenomatous Polyposis
- Gardner’s syndrome = intestinal polyposis, hamartoma of the skeleton, and multiple soft tissue tumors

CHRPE with bear tracks
- Familial Adenomatous Polyposis
  - 80% have bear tracking
  - Bilateral, extensive
  - Increased risk of colon cancer
    - 7% risk by age 21, 87% by age 45 and 93% by age 50
    - Genetic testing for APC tumor suppressor mutation

Pars planitis
- Many causes of inflammation in children
  - Pars planitis - idiopathic
  - Infectious - Toxocara
  - Inflammatory - JRA

Pars planitis
- Dense vitreous debris, snowballs and snowbanks
- Extensive w/u – then treat with steroids/ablate
Trauma related

- Choroidal rupture
- Commotio
- Sclopetaria
- Dialysis
- Vitreous base separation
- GRT

Choroidal rupture

Commotio

Sclopetaria
Retinal Dialysis

Avulsion of vitreous base

Tear with RD

Zebras

Laser pointer injuries

10 year old male -
650nm red laser pointer
13 year old male—
green laser pointer 532nm

Summary
- Many diseases can cause visual loss in children
- Early identification = better VA
- If in doubt - refer