AMERICAN ACADEMY of OPTOMETRY

Visual consequences of common (and not so common) pediatric conditions: things you don't want to miss!

Marcela Frazier OD, MPH, FAAO
mfrazier@uab.edu

Disclosure Statement

- Nothing to disclose

ACADEMY 2012 PHOENIX

Objectives

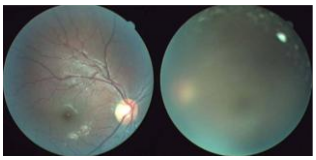
- To review the visual consequences and manifestations of pediatric systemic diseases
- To improve early detection and prevention of visual sequelae from pediatric systemic diseases
- To review visual manifestations that may lead to diagnoses of systemic disease

12 YO Caucasian female

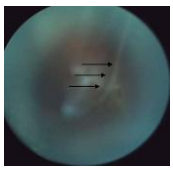
- Here for routine yearly eye exam
- Normal birth and health history, no meds
- VA cc OD -2.00 20/20 OS -1.50 20/60
- Ref OD -2.00 20/20 OS -3.50 20/20
- PERRL -APD
- We had been seeing her yearly for the last 5 years

7 YO Hispanic male

- Mom reports child complains of: 'Reduced vision in left eye after car accident'
- Normal birth and health history. No meds
- 20/20 OD 20/400 OS
- A/C 4+ Cells and flare OS. Fundus:



- Toxoplasmosis IgG+ IgM-
- Toxocariasis=ELISA?



2 YO Caucasian Male

- Mother reports: 'his eyes turn in when looking up close since last month'.
- Pt was born at 30 wks gestation, 4lbs 3 oz
- Child is not walking yet, has hypotonia. Saying only 'mama' and 'dada'
- No dysmorphic features
- Mother's history unremarkable other than took *escitalopram* (SSRI) during pregnancy
- Family history of fragile X syndrome

2 YO Caucasian Male

- Fixes and follows small objects OD/OS
- Pendular, small amplitude nystagmus that stays horizontal on upgaze
- TAC 20/200 OD/OS
- Dynamic Ret +3.50 lag OD/OS
- ~20 ^CRET @ near, appears ortho at dist
- Wet Ret +7.50 DS OD; +6.50 DS OS

2 YO Caucasian Male

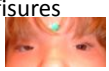
- Referred for MRI, neurology consult and genetic eval

2 YO Male

- Bilateral CONH confirmed
- No Septo Optic Dysplasia
- Choromosome analysis showed 5 q (long) deletion (not cri du chat=short arm of 5)
  - Strabismus
  - Anemia that may lead to leukemia
  - delayed growth and development
  - Hypotonia
  - Epicanthal folds

Eye manifestations of chromosomal anomalies

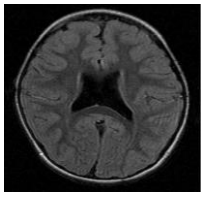
- Strabismus
- High refractive errors
- Hypo/hypertelorism
- Blepharophimosis
- Up or down slanting of palpebral fisures
- Ptosis
- Epicanthal folds



- Microphthalmia
- Enophthalmus
- Colobomas
- Corneal Opacities
- Cataracts
- Reigers
- PHPV
- Glaucoma
- Optic nerve hypoplasia

Septo Optic Dysplasia

- CNS anomalies, absence of septum pellucidum, agenesis of corpus callosum.
- May have hypopituitarism (refer to endocrin.)
- Growth delays
- Seizures
- MR



8 YO Hispanic male

- Mother reports child eyes have 'recently started dancing'
- Child has neurofibromatosis 1 (and family history of NF1)
- VA 20/40 OD/OS uncorrected
- PERRL –APD
- Rapid, conjugate 'nystagmus' in all directions

Acquired Nystagmus

- Signals a Neurological problem
  - Downbeat nystagmus (Arnold-Chiari's syndrome, spinocerebellar degeneration, stroke, MS, nutritional)
  - Upbeat nystagmus (brainstem lesion, MS)
  - Periodic alternating nystagmus (stroke or MS)
  - Rotary Nystagmus (vestibular issues)
  - See-Saw nystagmus (suprasellar lesion like pituitary tumors; bitemporal hemianopia is common)
  - Abducting nystagmus (MLF lesion, along w INO)

Neuroblastoma

- Referred for full body scan and neurology consult:

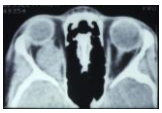
Neuroblastoma:

Malignant tumor which manifests as a lump or mass in the abdomen or around the spinal cord in the chest, neck, or pelvis.

Often present at birth, but is most often diagnosed much later when the child begins to show symptoms of the disease, such as **Opsoclonus**

Neurofibromatosis

- Lisch nodules (in NF1)
- Retinal hamartomas
- Neurofibromas of the eyelid
- **Optic nerve gliomas** (15-40% of children w NF1)
- Prominent corneal nerves
- Glaucoma



Other Phakomatoses

- **Tuberous Sclerosis:** Nodular hamartomas in cerebral hemispheres.

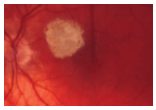
  May have *epilepsy, MR, and abnormal behavior.*

Ash-Leaf Spots

Glial hamartomas of the retina

Papilledema

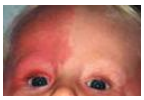
Optic atrophy



Other Phakomatoses

- **Sturge-Weber Syndrome:** **(**Encephalotrigeminal angiomatosis)
- Vascular malformations of the meninges, brain, skin, and eyes.

Port Wine Stain



Ocular signs:

Uveal hemangiomas

Glaucoma

Other Phakomatoses

- Angiomatosis retinae (Von Hippel-Lindau disease)
  - Retinal and optic nerve hemangiomas
- Ataxia telangiectasia (Louis-Bar syndrome):
  - Telangectasia of conjunctiva
  - Oculomotor abnormalities (similar to OMA)
- Wyburn-Mason syndrome
  - Racemose (worm-like) Hemangiomatosis

16 YO Caucasian male

- Patient complains of 'Red, painful eyes for two days'
- Normal birth and health history
- Has had pain and stiffness in the middle part of his spine for two months
- BCVA OD 20/100 OS 20/80 PHNI
- PER mid dilated –APD

16 YO male

- A/C 4+ Cells and flare OU
- Patient reports his eyes have gotten red and painful like this a couple more times previously

Recurrent Uveitis

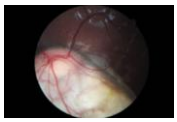
- Ankylosing spondylitis
- Periarteritis nodosa
- Reiter's syndrome
- Rheumatoid arthritis
- Sarcoidosis
- Scleroderma
- Systemic lupus erythematosus

Reiger's and Axenfeld's Systemic Associations

- Facial, dental, and umbilical anomalies
- Growth hormone deficiency
- Myotonic dystrophy
- Atrial Septal Defect
- Cholestasis fro Arteriohepatic dysplasia (also associated with optic nerve head drusen)
- Hydrocepahlus
- Skeletal abnormalities
- Eye muscle problems
- Hearing loss

CHARGE

- Colobomas (iris, ON, retina, choroid)
- Heart Disease
- Atresia of the nasal choanae
- Retarded growth and Development
- Genital hypoplasia
- Ear anomalies



Gillespie Syndrome

- Cerebellar ataxia, mental delays, cardiopulmonary and vertebral problems
- Ocular findings:
  - Aniridia
  - Glaucoma
  - Telecanthus
  - Strabismus
  - Hyperopia

Marfan Syndrome

- Problems with: heart, blood vessels, lungs, eyes, bones and ligaments
- Ocular findings:
  - Myopia
  - Astigmatism
  - Superior temporal lens subluxation
  - Glaucoma

Ehrler-Danlos syndrome

- Connective tissue disorders
- Ocular findings:
  - Epicanthal Folds
  - Blue sclera
  - Myopia
  - Microcornea
  - Keratoconus
  - Glaucoma
  - Ectopia lentis

Goldenhar Syndrome

- Hemifacial microsomia, preauricular apendages, facial palsy, deafness, vertebral abnormalities
- Ocular findings:
  - Lid coloboma
  - Limbal dermoids
  - Strabismus
  - Duane syndrome

Waardenburg Syndrome

- Sensorineural hearing loss
- Hair hypopigmentation (white forelock or white hairs at other sites on the body)

Ocular manifestations:

- Iris pigmentary abnormality
  - Iris pigmentary abnormality
  - Dystopia canthorum (lateral displacement of inner canthi)