Case Study

- 43 yo Caucasian Male
- Initial presentation for Presbyopia
  - -4.00D Myope
- Ochx: Unremarkable
- FOchx: unremarkable
- MHx: Unremarkable
- External / Slitlamp examination unremarkable
- IOPs: 15 Right and left
- Referred for investigation of 2 pigmented and 1 hypopigmented lesion

Right Pigmented lesion

Left Nasal

Left Temporal

DDx – Hypopigmented Lesion

- Window Defect
- Amelanotic Choroidal Naevus
- Myelinated NFL
- Choroidal Metastasis
- Retinoblastoma
- Choroidal osteoma
- Idiopathic sclero-choroidal calcification
- Combined Hamartoma of the Retina and RPE
- Astrocytic Hamartoma
Investigation:

- Raised or Flat
- What layers are involved (OCT)
  - Optical Properties
- Acoustic properties

Astrocytic Hamartoma

- Astrocyte (astroglia)
  - Characteristic shaped glial cells
- Retinal Astrocytes
  - Located almost entirely in the RNFL
    - Most numerous on the optic nerve head
  - Thought to be ‘sheaths’ for axons and vasculature
    - Blood brain/retinal barrier
- Hamartoma
  - A benign growth consisting of a disorganized mixture of cells and tissues normally found in the area of the body where the growth occurs.

Acoustic properties

Appearance / Progression

- Subtle initial appearance
  - Thickening of RNFL
- Become more opaque
- Yellow calcification
  - Mulberry appearance
- Autofluorescence
- Fluoroscein Angiography
- Endo or Exophytic
- Spontaneous regression / Stable / Progression
Acquired Retinal Astrocytoma

- More aggressive
  - Lacks Calcium
- Not associated with TS or NF1/NF2
- Shows progressive growth
  - Exudate
  - Vitreous Seeding and Haem
  - Retinal exudates and detachment
  - Neovascular Glaucoma

Astrocytic Hamartoma Associations

- Idiopathic
  - Solo lesions
- Phacomatoses
  - Tuberous Sclerosis (aka Bourneville’s disease)
    - Multiple hamartomas of all primary germ layers
    - Neurofibromatosis type 1 and 2
      - Primarily affects growth of neural tissues
- Retinitis Pigmentosa
  - DDx: Optic disc drusen

Tuberous Sclerosis - Diagnosis

- Clinical and Genetic diagnostic criteria 2012
- Genetic diagnostic criteria 2012
  - Autosomal Dominant
- Other ocular involvement:
  - Focal Iris Stromal Depigmentation (ash leaf)
  - Atypical Iris Coloboma
  - Hamartomas of Iris Pigment and ciliary epithelium
    - Iris Neovasc

NF1

- Incidence 1:4000
- Autosomal dominant
- Diagnosis based on 2 or more of
  - >6 Café-au-lait
  - 2+ neurofibromas
  - Axial or inguinal freckling
  - 2+ Lisch Nodules
  - Distinctive osseous lesions
  - First degree relative

NF2

- Incidence 1:35000
- Autosomal Dominant
- Diagnosis based on:
  - Bilateral acoustic neuroma
  - 1st degree relative with
    - Unilateral acoustic neuroma
    - 2+ of
      - Neurofibroma
      - Meningioma
      - Glioma
      - Schwannoma
      - Early Cataract

NF1 Ocular Manifestations

- Optic nerve glioma (15%)
- Meningioma
- Sphenoorbital Encephalocele
- Neurofibromas
  - Plexiform neurofibroma of eyelid
- Lisch nodules
- Ectropion Uvea (glaucoma)
- Astrocytic hamartoma (rare)
- Choroidal Naevi

Conclusions

- Astrocytic Hamartoms have wide range of appearances
- Imaging Armoury can help DDx Lesions seen later in life
- Paediatric Astrocytic Hamartomas need to be differentiated from Retinoblastoma
- Lesions should be assumed to be progressive
- Warrant systemic work up
- Multiple ocular complications of TSC, NF1, NF2
Acute corneal hydrops in post-LASIK ectasia
Sara N. Gaib, OD, FAAO

Disclosure Statement:
• Consultant – Bausch+Lomb

Please silence all mobile devices. Unauthorized recording of this session is prohibited.

Chief Complaint
• 48 year old male presents to Midwestern University Eye Institute
• Chief complaint: Constant hazy OS x 1 week with associated blur. Piggyback contact lens system doesn’t seem to be working as well. Reports mild serous discharge. No pain, no photophobia

Case History
• OHx:
  o S/P myopic LASIK OU (2005)
  o post-LASIK ectasia OS (2008)
• MHx:
  o Sleep apnea
  o Esophageal stricture

Objective testing
• VA (sc): OD: 20/20-2
  OS: LP PHNI
• Pupils: PERLL (-) APD
• EOM: FROM OU
• CF: FTFC OD, UTT OS

Anterior segment examination
<table>
<thead>
<tr>
<th>OD</th>
<th>OS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lids/lashes:</td>
<td>Clear</td>
</tr>
<tr>
<td>Conjunctiva:</td>
<td>Clear</td>
</tr>
<tr>
<td>Cornea:</td>
<td>LASIK scar</td>
</tr>
<tr>
<td>AC:</td>
<td>Deep/quiet</td>
</tr>
</tbody>
</table>

Specialized testing
• Spectralis OCT of the cornea
Diagnosis

• Acute corneal hydrops
  o rupturing of Descemet's and its underlying endothelium allowing fluid entry into the cornea

Corneal hydrops

• occurs in 3% of patients with keratoconus
• risk factors:
  o male gender
  o advanced ectasia
  o atopy
  o eye rubbing
  o eccentric cone location

Post-LASIK ectasia

• keratectasia was first described as a complication of LASIK in 1998
• develops > 1 yr after surgery in > 50% cases
• 1 in 2,000 for femtosecond laser flaps
• 1 in 1,000 for microkeratome flaps
• rate can be significantly higher (1 in 17) in unrecognized form fruste keratoconus

Management options

• patching, bandage contact lens, cycloplegia, hypertonic sodium chloride, steroidal and non-steroidal anti-inflammatory ophthalmic agents, as well as reassurance
• 2-4 months for corneal edema to subside
• use of intracameral air injections has been investigated, and although recovery time is faster, the final visual outcome is similar to conventional therapies

Management

• Rx:
  o Muro 128 2% solution qid OS
  o Muro 128 2% ointment qhs OS
  o Acuvail 0.45% bid OS

• RTC 1 month for follow-up

1 month follow-up: Vccl 20/40+

2 month follow-up: Vccl 20/20-
Long-term management

- GP popping out
- pt requests more stable CL option
- CL fit OS only:
  - Maxim Scleral 6.49/16.6/-10.25DS/Boston XO2

Discussion

- acute corneal hydrops is a rare complication of keratoconus, or even more rarely of post-LASIK ectasia
- caused by disruption in Descemet's leading to corneal edema
- largely managed with hyperosmotics
- consideration for scleral lenses in irregular cornea management

References

Posterior lenticonus in a patient with Marfan syndrome characteristics
Lorne Yudcovitch, OD, MS, FAAO
Pacific University College of Optometry
Disclosure Statement: Nothing to disclose

Case History
• 18-year-old undergraduate student
• Avid rower
• Presents for routine vision exam
• Chronic blurred vision, left eye – since early childhood
• No known medical problems
• No medications or medication allergies
• No known family ocular or systemic disease

Entrance tests
• Best-corrected visual acuities
  – OD 20/15, OS 20/200 (PH no improvement OS)
• Ocular motilities
  – Full and equal in all gazes OU
• Pupils
  – Equal & reactive to light and accommodation OU
• Visual fields
  – No defects seen on screening C-20-5 FDT OD, OS
• Binocularity
  – Unable to obtain stereopsis; small angle OS esotropia

Refraction
<p>| | | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>OD</td>
<td>+0.25-0.25 x 097</td>
<td>20/15</td>
</tr>
<tr>
<td>OS</td>
<td>-1.00-1.75 x 018</td>
<td>20/50</td>
</tr>
</tbody>
</table>

Anterior segment
• Ocular pressures
  – 14 OD, 12 OS @ 10:35 AM (Non-contact tonometer)
• Corneas
  – Clear all layers; no thinning or edema OU
• Corneal topography
  – Normal corneal curvature with no irregularities OU
• Anterior chambers
  – Deep and quiet OU
• Lenses
  – Capsule, cortex, and nucleus appears clear OU
LBY – Posterior Lenticonus in a Patient with Marfan Syndrome Characteristics

Posterior segment

- Dilated fundus exam
- No abnormalities seen on any structure
- Healthy retinal sheen OU
Direct ophthalmoscopy
- “Oil droplet” pupil reflex OS seen; OD normal

Repeat Biomicroscopy
- Subtle posterior lens ectasia OS seen on repeat optic section

Physical External Evaluation
- Tall, slender appearance
- Elongated fingers and toes
- Highly-arched upper palate
**Differential diagnosis**

- Primary leading diagnoses:
  - Posterior lenticonus OS
  - Marfan syndrome characteristics
- Others:
  - Anterior lenticonus
  - Lentiglobus
  - Spherophakia
  - Microspherophakia
  - Lens subluxation
  - Cataract

**Diagnosis and discussion**

Posterior lenticonus:
- 1 to 4 per 100,000 incidence
- Pathophysiology unknown
- Autosomal recessive (TDRD7 gene mutation)
- Usually unilateral; sporadic bilateral familial cases known
- Posterior opacity often associated
- Strabismus commonly associated
- Rare systemic associations (i.e. Alport syndrome)

**Marfan syndrome (MFS)**

- 1 per 3000 to 5000 prevalence
- Multisystem connective tissue disease (heart, eye, skeletal)
- Autosomal dom. (FBN1 gene mutation); variable expression
- Increased height and arm span
- Anterior chest wall abnormalities
  - pectus excavatum or carinatum
- Long fingers and toes (arachnodactyly)
- Narrow, highly arched palate
- Vertebral column deformities (scoliosis, thoracic lordosis)
- Dural ectasia in approximately 60% of patients
- Thoracic aortic aneurysms → aortic dissection/rupture

**Marfan syndrome and the Eye**

- Ectopia lentis in about half of patients with MFS
  - Dilation may be best to discover
- Myopia common
- Rare lens-related glaucoma
  - Usually from subluxed lens
- Retinal detachment rare complication
  - More risk post-cataract surgery

**Treatment and Management**

- Treatment and response to treatment
  - Patient education regarding findings
  - Physical exertion warning
  - Polycarbonate spectacle protection
  - Referral for physical, cardiac evaluation
  - Pt declined lens surgery, VT as treatment options
  - Genetic testing/counseling
- Diagnostic tests for lenticonus
  - Slit-lamp biomicroscopy
  - Pupillary reflex test
  - Anterior segment optical coherence tomography (OCT)
  - Scheimpflug photography
  - Ultrasound biomicroscope (UBM)

**Echocardiogram**

- Heart murmur
- Mitral valve irregularity
- Possible aortic root dilation
LBY – Posterior Lenticous in a Patient with Marfan Syndrome Characteristics

**Bibliography/literature review**


**Conclusion**

Clinical pearls/takeaway points:

- Pupillary reflex test is valuable in determining lenticonus diagnosis.
- Possible posterior lenticonus-Marfan syndrome connection.
- Cardiac evaluation is critical in patients with Marfan characteristics.
- Optometric examination may be first time Marfan syndrome identified.

Please complete your session evaluation using EyeMAP™ online at http://eyemap.cistems.net
Tweet about this session using the official meeting hashtag #aaoptom14.