I. Case History
- 17 year old Hispanic male
- Presents for initial comprehensive eye exam
  - Sent from urgent care due to diagnosis of Alport Syndrome
- Patient complaining of blurry vision at distance without correction
- Ocular history: None
- Medical history: Alport syndrome, chronic kidney disease, bilateral hearing loss
- Medications: Enalapril 10mg
- Family history of chronic kidney disease (mother, brother)

II. Pertinent Findings
- BCVA 20/40 OD, 20/30-2 OS
  - First time glasses wearer, no amblyogenic refractive error, no strabismic/deprivation amblyogenic findings
- Recurrent corneal erosion OD
- Macula: annulus of focal, whitish-yellow perifoveal deposits/flecks OU
- OCT OU: normal inner/outer retina extending nasal from fovea; several areas of hyper reflective ILM/RNFL corresponding to dot/fleck macular deposits; inner/outer retina extending temporal to fovea thinned (ILM/RNFL visibly thinned); no macular hole formation
- Fundus photos show perimacular flecks OU

III. Differential Diagnosis
- Alport Syndrome

IV. Diagnosis and Discussion
- Alport syndrome affects 1 in 50,000 newborns and accounts for approximately 3% and 0.2% of ESRD in children and adults respectively
- Alport syndrome is a genetic condition characterized by the absence of collagen IV network from basement membranes caused by mutations in the COL4A5 (X-linked) or COL4A3/COL4A4 (autosomal recessive)
- Collagen IV α3α4α5 networks are important structural components in basement membranes of the glomerulus, cochlea, cornea, lens and retina
- Subsequently, nephritis, hematuria, progressive kidney failure and hearing loss are characteristic systemic findings
- Characteristic Ocular Findings
  - Cornea:
    - Recurrent corneal erosion
    - Posterior polymorphous dystrophy
  - Lens:
    - Anterior lenticonus (oil-droplet cataract)
  - Retina:
    - Perimacular fleck retinopathy
    - Peripheral coalescing fleck retinopathy
    - Temporal retinal thinning
    - Loss of foveal reflex
    - Macular hole
V. Treatment, management
- Phacoemulsification and IOL implantation for anterior lenticonus patients and management of RCE/PPMD
- Management of kidney disease and resultant hypertension
  - Medications
    - ACE inhibitors, ARBs, aldosterone inhibitor
  - Kidney transplantation
    - 3-5% risk of post-transplantation anti-GBM nephritis in males
  - Dialysis

VI. Conclusion
- Ophthalmoscopic evaluation can help confirm the diagnosis of Alport syndrome by identifying characteristic ocular manifestations
  - Presentation in females can vary widely therefore ocular signs can be helpful diagnostic indicators
- Intervention and treatment of ocular findings are not indicated other than treatment of decreased vision caused by cataracts or recurrent corneal erosions
- Management of kidney disease is paramount

References


Kashtan C. Alport syndrome; http://rarediseases.org/rare-diseases/alport-syndrome/


