Importance of Optical Coherence Tomography and Early Cone Dystrophies:
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Abstract: Cone dystrophies have life changing implications. In early symptomatic cases, young patient’s biomicroscopic macular findings may appear completely normal. Optical Coherence Tomography (OCT) can provide practitioners with proper visualization of early photoreceptor loss.

Case History:
- Patient Demographics: 14-year-old white female
- Chief Complaint: The patient complains of decreased visibility of softball while batting.
- Ocular History / Medical History: None (No family ocular history) / Asthma
- Medications: None
- Patient referred to retinal specialist by local OD for central visual field defect.
- Retinal specialist saw the patient, observed no macular changes, noted a central visual field defect, questioned whether it was mild amblyopia but referred for MRI.

Pertinent Findings:
- Clinical:
  - BCVA: OD: 20/30\(^2\), OS: 20/25\(^2\)
  - Refractive Error: -0.75 OD, -1.00 OS
  - Pupils/EOMS/CVF: Normal OU
  - Color Vision (Ishihara): 1/14 OU (not tested on initial examination)
- Physical:
  - Lids/Lashes/Conjunctiva/Cornea/Lens: Normal OU
  - Optic Nerves: 0.3 C/D OU-- Round, pink, distinct OU
  - Macula: Normal OU
  - Periphery: Normal OU
- Radiology Studies:
  - MRI (with and without contrast): Normal
- Other Tests:
  - HVF 30-2: Small central scotoma
  - OCT: Central foveal gap missing Ellipsoid and Interdigitation Zones OU (not tested on initial examination)
  - Fluorescein Angiography: Dark central choroid OU; Potential beginning of bullseye defect OU;
  - ERG: Normal Photopic, Normal Scotopic OU

Differential Diagnosis:
- Primary/Lead: Cone Dystrophy
- Others: Rod-Cone Dystrophy, Stargardt’s Disease, Solar Retinopathy, Color Blindness

Diagnosis and Discussion:
According to the Gass Atlas, a cone dystrophy is a “heritable dystrophy in which predominately the cone system is affected”\(^1\). The patient may notice small changes in visual acuity, color vision, light sensitivity, and central visual field defects. The changes in the outer retina (photoreceptors) are generally due to mechanisms such as “defective outer segment morphogenesis, protein transport along the cilium, phototransduction, or cellular interaction”\(^3\).
Visualization of the cone dystrophy depends on the progression of the disease. In some early cases, the macula may appear normal with mild pigmentary changes. Later stages of the disease present as more of a bull’s-eye pattern of retinal pigmented epithelium changes.

Fluorescein angiography (FA) normally shows depigmentation evidence in central macula\(^1\). The patient’s FA showed some retinal pigmentary changes, but she also had a dark central choroid. Early electroretinogram (ERG) findings are usually normal but progress over time. The 14-year-old’s ERG was normal for both rod and cone functionality. OCT can show outer retinal thinning and foveal changes such as “foveal cavitation”\(^2\). These findings can vary, but in early cases loss of the photoreceptor interdigitation zone (IZ) and lack of hyperreflectivity of the ellipsoid zone (EZ) are present on the scans\(^3\). The lack of the IZ and decreased hyperreflectivity of the EZ in this patient’s OCT helped solidify the diagnosis of a cone dystrophy.

Skilled retina specialists can have a difficult time spotting this condition in its early stages. Repeating the field as a 10-2 could have helped with a more accurate depiction of the visual field defect. If the field was analyzed further, one could have observed if it crossed the vertical midline or whether it was congruent. The results would have probably negated the order of an MRI and an OCT would have most likely been ordered earlier.

Further genetic testing is available since it is a heritable disease. There exist autosomal dominant, recessive, and X-linked forms of cone dystrophies\(^1\). Genetic testing should include ABCA4, CNGB3, ELOVL4, RDS, CRX, RET-GC1, and GCAP1\(^1,2\).

**Treatment and Management:**

- Recommended genetic testing. The mother and daughter are waiting to seek further testing with a geneticist.
- AREDS 2 was considered as a treatment, but upon investigation, research shows increased amounts of vitamin A with diseases associated with the ABCA4 gene can advance the degradation of the macula\(^4\).
- Bibliography:

**Conclusion:**

- Ultimately, OCT technology should be utilized as a primary standard of care in suspects with cone dystrophies. An early cone dystrophy macula may appear normal to the average eye, but the enhanced viewing of the interdigitation and ellipsoid zones will provide the clinician with an accurate diagnosis.