A Rare Case of Peripheral Cone Dystrophy

We present a rare variation of cone dystrophy presenting with normal vision, mild color defects, paracentral scotomas, paracentral retinal thinning, and normal-appearing fundus. No longitudinal reports exist in literature, but four-year follow-up indicates relative stability.

I. Case History

- Patient Demographics
  - 26 year old Korean/Caucasian male presents for annual eye exam.
- Chief Complaint
  - Blur at distance without glasses.
  - No other visual complaints.
  - No complaints of hemeralopia, nyctalopia, or photophobia.
- Ocular History
  - Myopia and astigmatism only.
- Family Ocular History
  - No history of eye disease or reduced vision in extended family.
  - Patient has no siblings.
- Medical History
  - Unremarkable with no systemic diseases.
- Medications
  - None.

II. Pertinent findings

- Best-corrected visual acuity 20/20 OD, OS.
- All confrontation testing is normal.
- Anterior segment findings are normal.
- Color vision with HRR plates and standard illuminant C:
  - OD: mild indeterminate red-green defect, moderate tetartan (blue-yellow) defect.
  - OS: mild indeterminate red-green and blue-yellow defect.
- Dilated fundus exam reveals normal-appearing fundus OU aside from mild temporal pallor of the optic disc OS.
- Visual field testing:
  - Initial FDT screening field: symmetric paracentral scotomas OD, OS.
  - Three subsequent HVF 30-2 visual fields: confirmed paracentral scotomas OD, OS.
  - HVF 30-2 four years later: no clear progression of visual field loss OD, OS.
Baseline HVF 10-2 four years later: symmetric paracentral scotomas up to 4 degrees from fixation OD, OS.

- Macular OCT:
  - Paracentral retinal thinning corresponding to location of visual field defects in both eyes, with slightly more thinning OD. Raster scan reveals no loss of any retinal layers and intact EZ/IZ lines in both eyes.
  - Macular OCTs four years later: no progression of retinal thinning in both eyes.
- Fundus autofluorescence:
  - Symmetric subtle hyperfluorescence in nasal perimacular area OD, OS
- Full-field ERG and mfERG have been ordered. Results will be included in the poster.

III. Differential diagnosis

- Peripheral cone dystrophy
- Occult Macular dystrophy
- Cone-rod dystrophy
- Toxic retinopathy
- Oligocone trichromacy
- Retinitis pigmentosa

IV. Diagnosis and discussion

- Peripheral cone dystrophy is a rare cone dystrophy characterized by:
  - Visual acuity normal to slightly reduced\(^1\)
    - 20/50 or better in all patients under 75 years of age without comorbidities
  - Normal-to-slightly reduced color vision\(^1\)
  - Paracentral scotomas\(^2\)
  - Paracentral retinal thinning with no loss of retinal layers as observed on OCT\(^3,4\)
  - Normal fundus appearance aside from mild temporal pallor in some cases\(^2,3,4,5\)
  - Full-field electroretinogram (ERG)
    - Scotopic ERG: normal rod function\(^2\)
    - Photopic ERG: severely reduced cone function\(^2\)
  - Multifocal ERG
    - Slightly detectable to near-normal response in macular area\(^2\)
    - Severely reduced peripheral amplitudes\(^2\)
  - Subtle parafoveal hyperfluorescence as observed in fundus autofluorescence\(^2\)
  - Normal fluorescein angiography\(^2\)
- Visual symptoms commonly include hemeralopia and photophobia.
- It is considered a subgroup of cone dystrophy, and only ten cases have been reported in the literature to date.
- Compared to other cone dystrophies, peripheral cones are predominantly affected resulting in normal-to-slightly-reduced color vision and better visual acuities.\(^5\)
- Autosomal recessive inheritance has been suggested, but there are no known causative genes.
• Disease progression is largely unknown due to the small number of reported cases, variance of disease presentation at older age, and no case reports following patients over the course of several years.6,7

V. Treatment, management

• Full-field ERG and multifocal ERG required for definitive diagnosis.
• Educate the patient:
  o The condition is inherited.
  o There are no known treatments for this specific condition at this time.
  o Final visual outcome is not fully known because this condition is so rare.
• Monitor visual function with HVF 10-2 fields and Macular OCT for any progression of visual field defects or further retinal thinning.

VI. Conclusion

• Clinical Pearls
  o Fundus autofluorescence is a helpful tool in highlighting subtle retinal disease, including various cone dystrophies, that may not be detectable on fundus exam.
  o Peripheral cone dystrophy is a rare subgroup of cone dystrophy that predominantly affects peripheral cones, and therefore has different symptoms and clinical signs than other cone dystrophies.
  o Macular OCT can be a helpful tool in diagnosing peripheral cone dystrophy and in monitoring disease progression.
  o Long-term prognosis for peripheral cone dystrophy is unclear due to limited number of reported cases.

Bibliography: