A Case of Bietti's Crystalline Dystrophy

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Introduction

Bietti's crystalline dystrophy (BCD) is rare autosomal recessive ocular disease, with about 100 reported cases. Patient's typically present with decreased vision or nyctalopia. BCD is more commonly seen in Asian populations. The onset of disease occurs from teenage years to third decade with poor visual prognosis[1].

Findings include yellow-white crystalline lipid deposits in the retina or cornea, retinal pigment epithelium degeneration and sclerosis of the choroidal vessels[2,3].

Case Presentation

A 74 year-old Caucasian female presents for progressively worsening vision OU. She presented thirty years prior, with complaints of nyctalopia. At that time, she was diagnosed with "atypical Stargardt disease."

She denies any family history of ocular disease.

On presentation, BCVA 20/20- OD, 20/25+ OS. HVF 10-2 revealed advanced loss with only a remaining central island OU.

On dilated fundus exam, retinal crystalline deposits were noted.

The patient's genetic testing was positive for a pathologic variant in CYP4V2 gene, confirming the diagnosis of BCD.

Figures

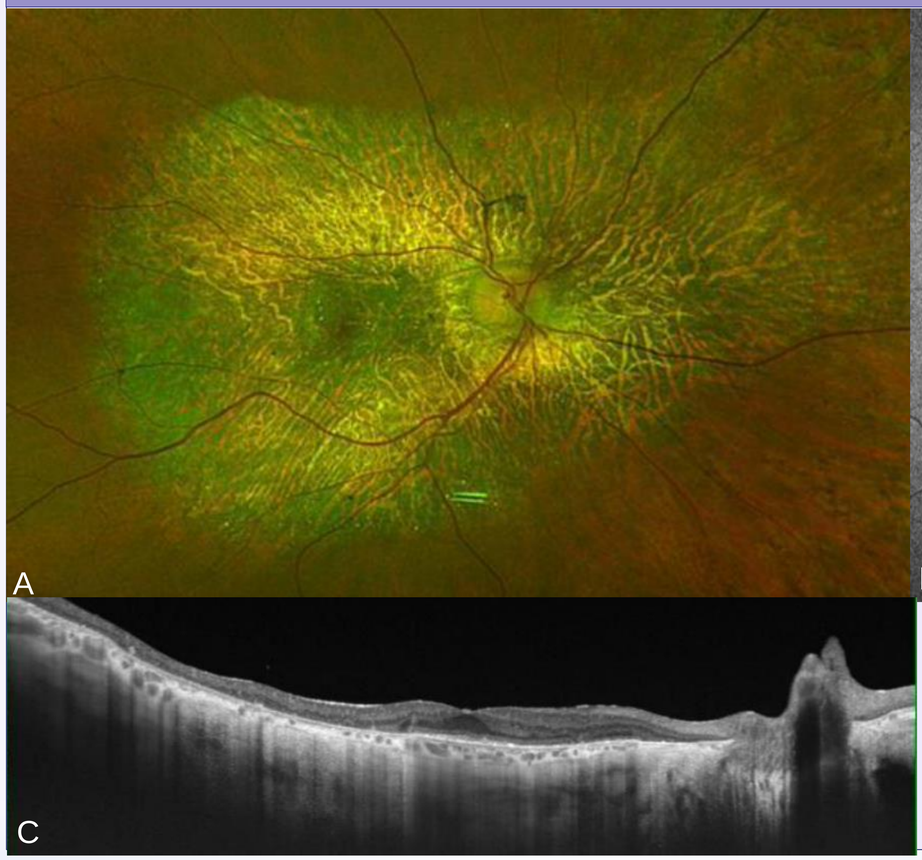

Figure A: Ultrawide field imaging of the right eye demonstrating the presence of fine yellow crystalline deposits.
Figure B: Fundus Autofluorescence of the right eye with hypoautofluorescence correlating to the retinal pigment epithelium atrophy in Figure A.
Figure C: Optical Coherence Tomography of the right eye with disruption of the outer retinal layers and diffuse retinal and choroidal thinning.

CONCLUSION

Though a rare entity, BCD may be misdiagnosed as a more common retinal condition, Stargardt disease. BCD can be distinguished from other retinal dystrophies by the presence of refractile retinal or corneal deposits.

Patients with BCD should be monitored annually, and examined for the development of choroidal neovascularization. Physicians should refer patient's to a low vision specialists upon diagnosis, especially given the progressive nature of this ocular disease.

This case raises awareness of BCD and encourages physicians to use genetic testing to guide the diagnosis in cases where there is clinical uncertainty.

References

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