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Seroreactivity against retinal proteins in a case of POC1B gene associated cone dystrophy with normal funduscopic appearance: a systematic approach to diagnosis

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ABSTRACT

Purpose

To report a case of cone dystrophy, associated with autosomal recessive homozygote POC1B gene variant, mimicking autoimmune retinopathy.

Case

A 45-year-old female presented with a complaint of decreased vision in both eyes. Her best corrected visual acuity was 20/32 in the right eye and 20/50 in the left eye. Anterior segment and dilated fundus examinations were unremarkable. Spectral domain optical coherence tomography showed a subfoveal blurred dome-shaped ellipsoid zone and an extinguished interdigitation zone affecting the entire macula. Full field electroretinography revealed reduced cone responses. The differential diagnosis included inflammatory chorioretinopathies, autoimmune retinopathies (paraneoplastic or nonparaneoplastic), and hereditary retinal dystrophies. No remarkable finding was observed on combined fluorescein and indocyanine green angiographies. Paraneoplastic autoimmune antibody panel revealed nothing; however, aldolase, enolase, pyruvate kinase M2, and glyceraldehyde-3-phosphate dehydrogenase antibodies were positive on autoimmune retinopathy panel. To exclude hereditary retinal dystrophies, whole-exome sequencing (WES) was applied. WES identified an autosomal recessive homozygote POC1B gene variant (c.680A>G, p.His227Arg). Cone dystrophy diagnosis was given.

Conclusion

Cone dystrophy associated with POC1B gene variant may present without visible fundus abnormalities. It should be kept in mind that retinal autoantibodies may be positive in such a hereditary dystrophy case due to long-term exposure of the immune system to self-antigens. Therefore, autoimmune retinopathy is a diagnosis of exclusion and should not be diagnosed until all other causes, including hereditary dystrophies, have been ruled out.

KEYWORDS:

Autosomal recessive cone dystrophy
proteome of the centriole 1b gene

autoimmune retinopathy
electroretinography