

Severe Stargardt disease with peripapillary sparing

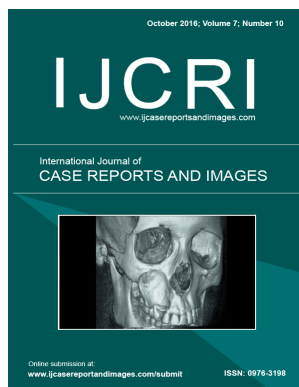
Heather Leisy, Meleha Ahmad, Nathaniel Tracer, R. Theodore Smith

ABSTRACT

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CASE REPORT

A 59-year-old white male with a long-standing history of Stargardt disease (STGD) presented for routine follow-up and yearly ocular fundus imaging. The patient reported progressive visual loss starting at approximately age fourteen and was subsequently diagnosed with STGD by an outside physician a few years later. Visual acuity at diagnosis was 20/200 bilaterally and progressively deteriorated to count fingers at 1 foot OD and 20/250 OS at the present visit. Past medical history was significant for chronic venous hypertension and deep vein thrombosis. There was no other significant past ocular history. There was no significant family ocular history.

Recent genetic testing revealed homozygous mutations in the ABCA4 gene for p.A1598D. As part of monitoring disease progression, infrared imaging, optical coherence tomography (OCT), and color fundus photography were performed. Infrared (Figure 1) and autofluorescence (Figure 2) imaging of this patient were significant for perimacular and peripheral geographic atrophy and pigment changes, consistent with severe STGD. On imaging, the peripapillary region was, however, spared.

DISCUSSION

Stargardt disease is the most common form of juvenile macular dystrophy caused by mutations in the ABCA4 gene that encodes the transporter protein for vitamin A derivatives. Prognosis varies and in this case the images demonstrate widespread degeneration of the retina. There is, however, peripapillary sparing, which is quite unusual in such cases of later stage STGD [1].

The missense mutation 4793 C→A, first described by Maugeri et al. in their evaluation of cone-rod dystrophies,

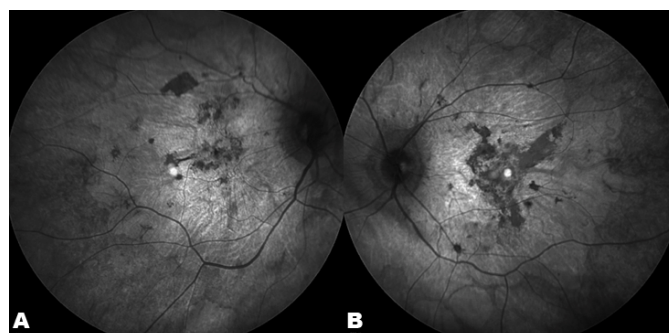


Figure 1: Infrared image showing severe Stargardt disease. (A) Right eye, and (B) Left eye infrared wide-angle image illustrating widespread geographic atrophy, pigment deposition and peripapillary sparing.

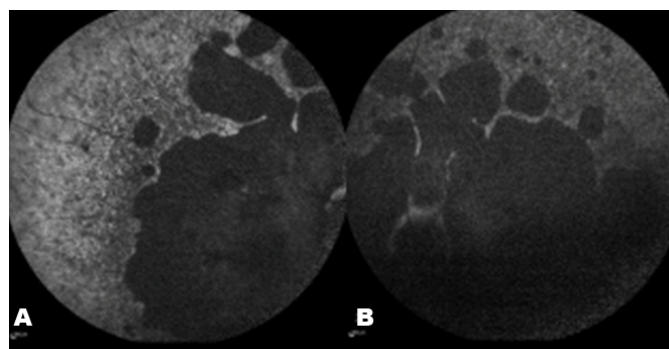


Figure 2: Autofluorescence imaging showing severe atrophy of the retinal pigment epithelium (RPE) in and around macula bilaterally, but with peripapillary sparing in (A) Right eye, and (B) Left eye.

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causes the formation of the aberrant protein (p.A1598D) [2]. It is unclear whether patients with homozygous mutations in the ABCA4 gene tend to present with more severe phenotypes [3, 4], or whether it is the severity of the mutation involved that determines the phenotype in homozygotes. With over 600 mutations identified in the ABCA4 gene, [5] genotype-phenotype correlation will become increasingly important in understanding STGD.

CONCLUSION

Severe stages of Stargardt disease with extensive atrophy and pigment deposition may have peripapillary sparing. Particular phenotypic presentations of Stargardt disease could have underlying mechanisms explained by genotype. Noting the phenotype-genotype correlation may help elucidate different presentations of the disease.

Keywords: Stargardt disease, Lipofuscin, Retina

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Author Contributions

Heather Leisy – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Meleha Ahmad – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Nathaniel Tracer – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Theodore Smith – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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