Use of Genetic Testing to Confirm Late-Onset Stargardt Disease

Michelle K. Man, OD, FAAO; Raman Bhakhri, OD, FAAO; Ashley M. Speilburg, OD, FAAO; Fred Collison, OD, FAAO
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INTRODUCTION

Stargardt disease is an autosomal recessive inherited retinal dystrophy caused by ABCA4 gene mutations. It is characterized by retinal yellow or white flecks and RPE atrophy. Symptoms traditionally occur during the first two decades of life, leading to gradual central vision loss. However, a less common and late-onset subtype of Stargardt can also occur. It typically presents in the fourth or fifth decades of life, is usually not as severe and less progressive; therefore, it may be confused for other maculopathies making genetic testing a vital test in confirming the diagnosis.

CASE REPORT

A 68-year-old Black female presented to the clinic with the chief complaint of floaters OU. Her systemic history was significant for osteoarthritis, hyperlipidemia, and hypertension, which were controlled with medications. Her best-corrected acuities were 20/20- in each eye. Dilated examination confirmed vitreous syneresis OD and a complete PVD OS, consistent with her chief complaint. Incidental findings included macular pigmentary changes in a parafoveal bullseye pattern OU (Image 1a) and additional testing was ordered.

IMAGE 1A

Optos fundus photography displays parafoveal macular pigmentary changes in the right and left eye.

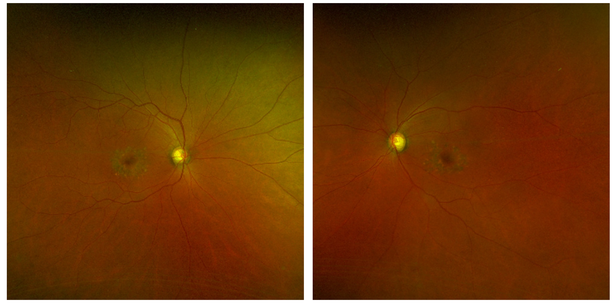


IMAGE 1B

Fundus autofluorescence of the right and left eye shows parafoveal hyper and hypo autofluorescence corresponding to areas of retinal involvement (flecks) and RPE atrophy respectively.

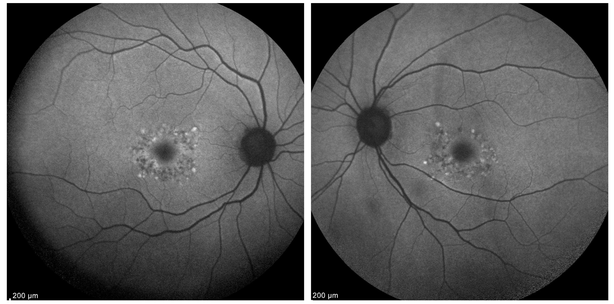


IMAGE 2

High definition 5-line raster macular OCT images of the right and left eye reveal disruption of the IS-OS junction of the photoreceptors, sparing the fovea. The external limiting membrane is subtly enhanced in both eyes.

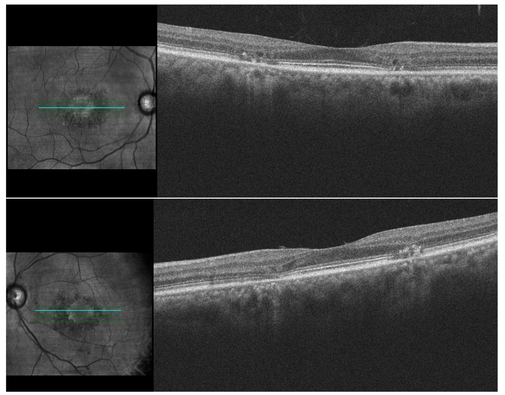
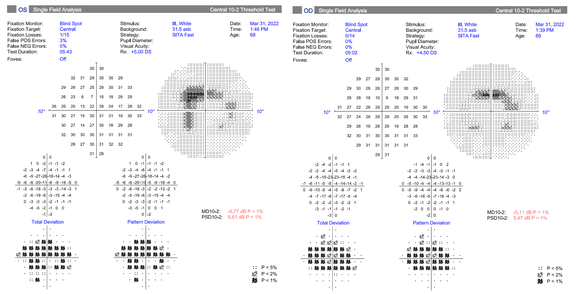


IMAGE 3

HVF 10-2 SITA Fast testing demonstrates bilateral paracentral relative scotomas, corresponding to the parafoveal lesions.



RESULTS

Optical coherence tomography (OCT) testing revealed parafoveal disruption of the inner segment-outer segment (IS-OS) junction of the photoreceptors, sparing the fovea OU (Image 2). Fundus autofluorescence (FAF) showed hyper and hypo autofluorescence corresponding to areas of retinal involvement and RPE atrophy respectively (Image 1b). Baseline Humphrey Visual Field (HVF) 10-2 testing resulted in a central ring scotoma in each eye (Image 3). She denied hydroxychloroquine use. Due to these findings, the patient was scheduled for genetic testing (Invitae, ID your IRD program) which confirmed an ABCA4 mutation (c.6079C>T). She was diagnosed with late-onset Stargardt disease and genetic counseling was provided.

CONCLUSION

Late-onset Stargardt disease usually presents with a milder decrease in visual acuity and more subtle retinal findings when compared to traditional Stargardt disease. This report highlights an example of incidental diagnosis during routine eye examination for an asymptomatic patient. Genetic testing (Invitae, ID your IRD program) supported the diagnosis. In this case, progressive vision loss is unlikely but regular follow up examinations with OCT and FAF will be useful to monitor the disease.

REFERENCES

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CONTACT INFORMATION

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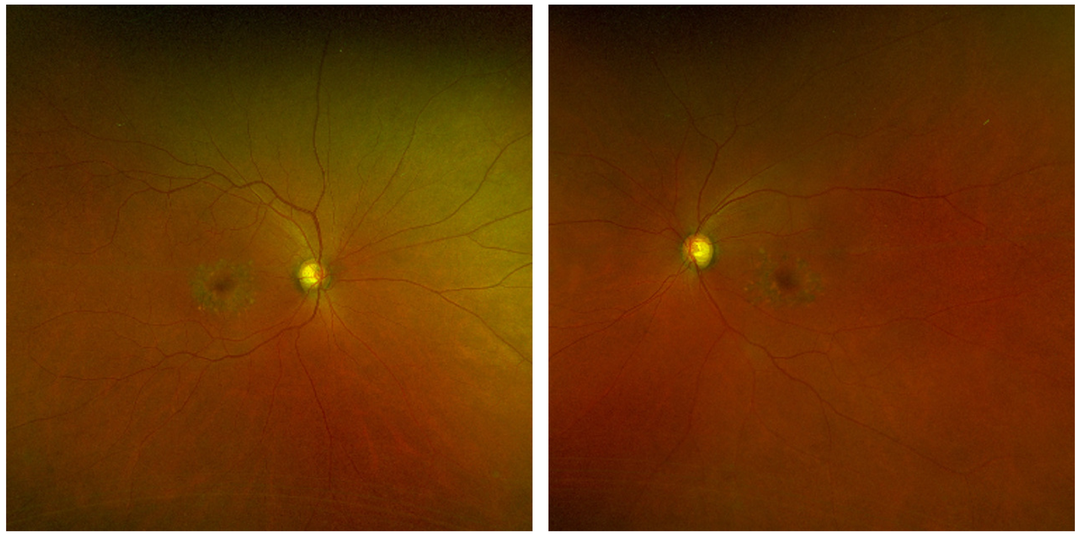

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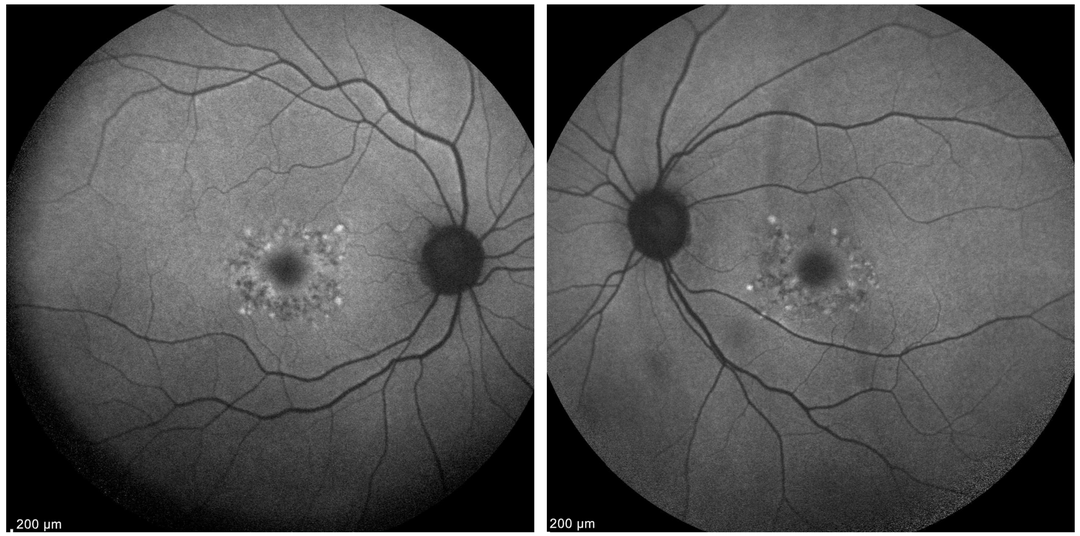

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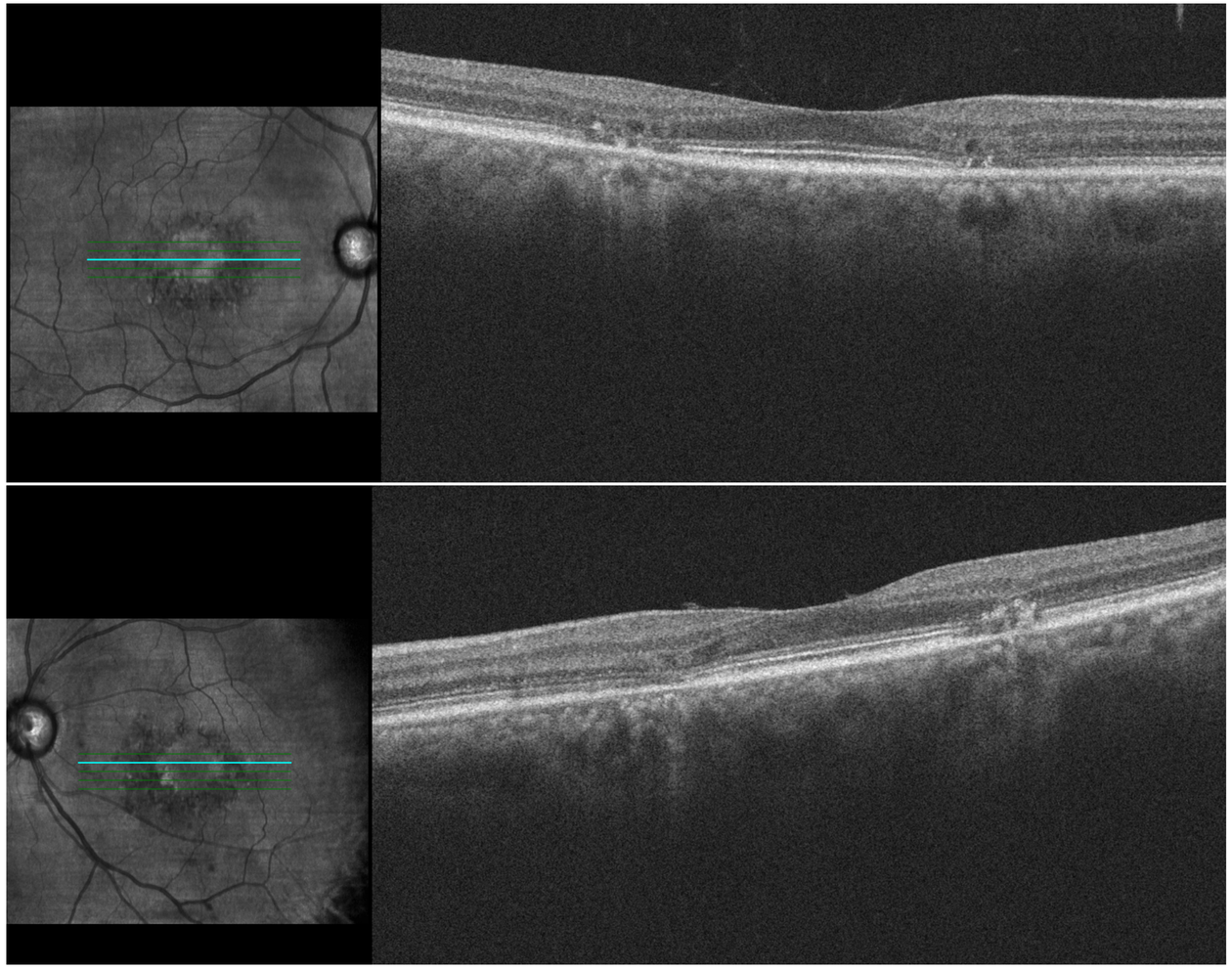

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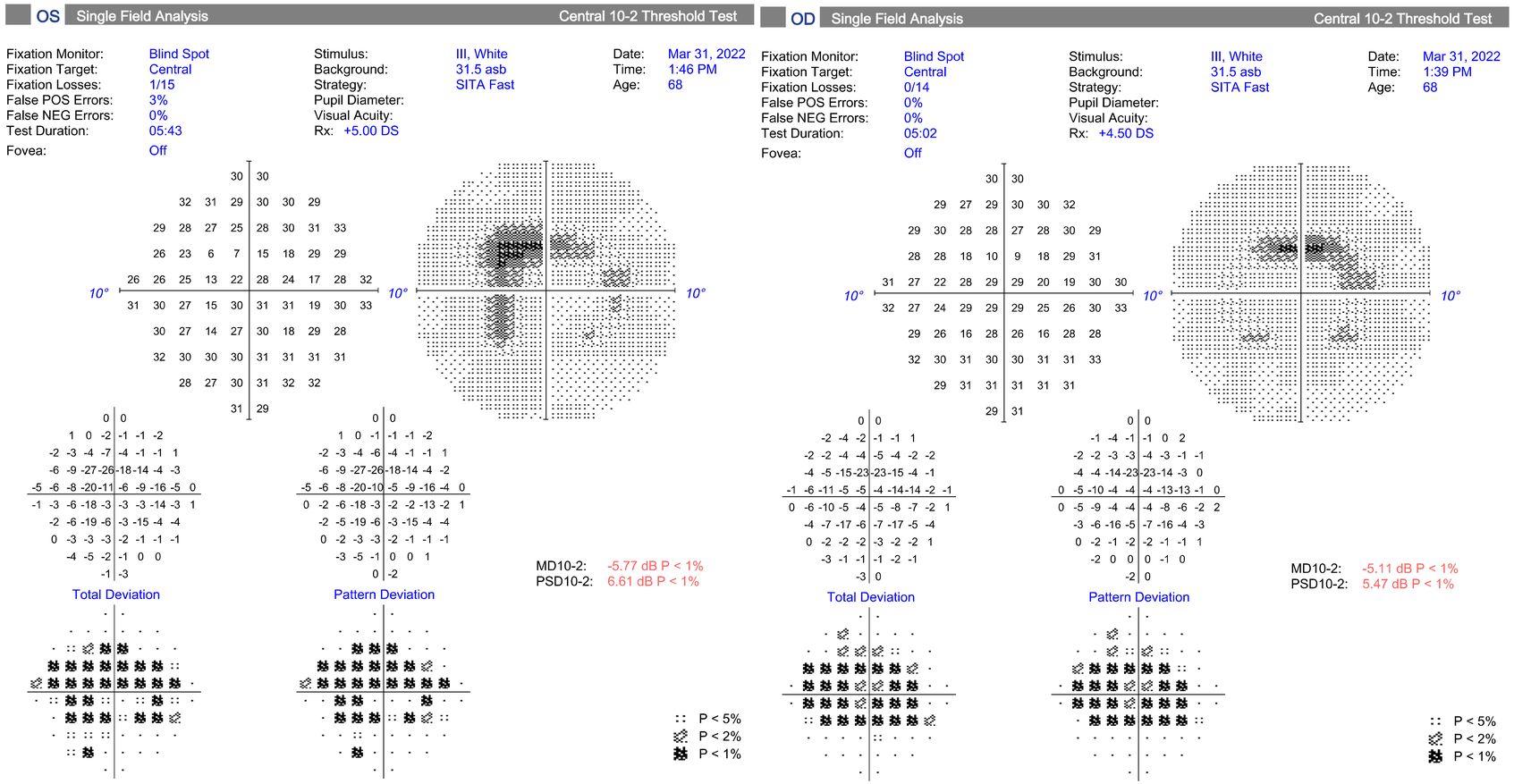

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