LATE ONSET FOVEAL SPARING STARGARDT DISEASE: A CLINICAL AND MOLECULAR DESCRIPTION

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PURPOSE

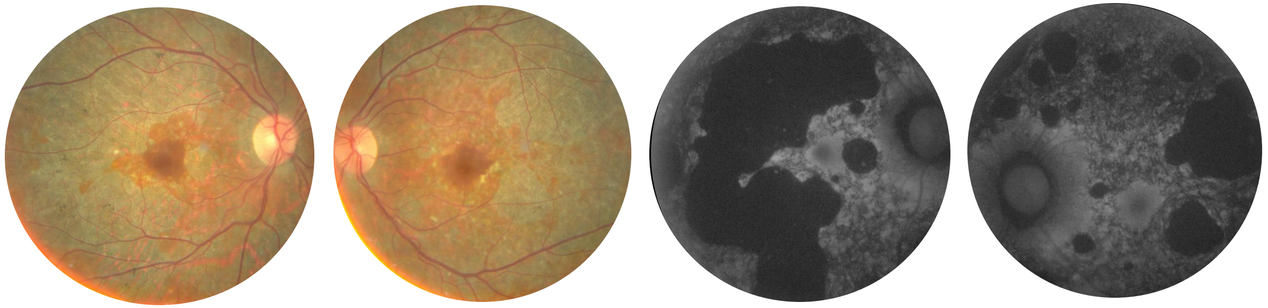
We aim to report a case of late-onset Stargardt disease confirmed by genetic testing and discuss the typical clinical features and, also, differential diagnosis.

METHODS

The case report was elaborated from the analysis of medical records of one reference hospital in Belo Horizonte / MG.

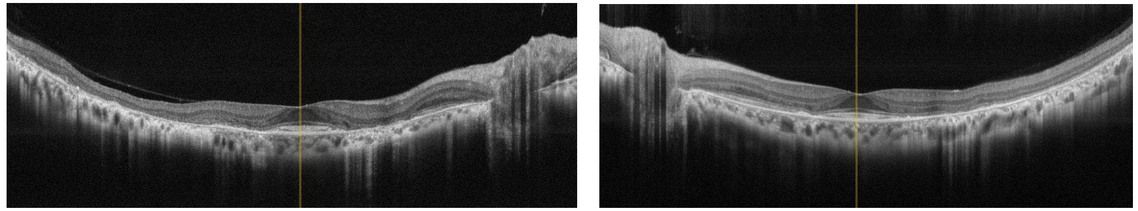
CASE REPORT

A 54-year-old woman presented with no complaints for medical appointment. Family ocular history included possible macular degeneration in her parents. Being treated for hypothyroidism. Vision was 20/20 in both eyes. Anterior segment examination was unremarkable. Fundus examination of both eyes revealed irregular flavimaculatus flecks and sharply delineated areas of chorioretinal atrophy at the posterior pole until the midperiphery as well as a parafoveal atrophy of the RPE sparing central fovea .



Autofluorescence shows hyperautofluorescent flecks and hypoautofluorescent areas corresponding to the atrophy ones.

An OCT scan through the fovea clearly shows the preserved cone photoreceptors in the central fovea surrounded by atrophy of the outer retina and RPE.



Genetic testing was performed and revealed a pathogenic heterozygous ABCA4 c.5196+1056A>G (Intronic) variant and a Uncertain Significance heterozygous variant ABCA4 c.5672_5673delinsCT (p.Leu1891Pro).

DISCUSSION

Stargardt disease is typically described in young patients but may develop later in adulthood and masquerade as age-related macular degeneration and several other conditions. We report a case of an asymptomatic patient with extensive areas of chorioretinal atrophy sparing central fovea that prompted us to perform genetic test confirming the suspected clinical diagnosis of late onset foveal sparing Stargardt disease. Faced with a phenotype that mimics so many retinal diseases, genetic testing is essential for diagnostic confirmation.

REFERENCE

1- van Huet, Ramon AC, et al. "Foveal sparing in Stargardt disease." *Investigative ophthalmology & visual science* 55.11 (2014): 7467-7478.

2- Lambertus, Stanley, et al. "Progression of late-onset Stargardt disease." *Investigative ophthalmology & visual science* 57.13 (2016): 5186-5191.

3- Alsberge, Joseph B., and Anita Agarwal. "Late-onset Stargardt disease." *American Journal of Ophthalmology Case Reports* 26 (2022): 101429.