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Stargardt disease multimodal analysis. Case Report

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PURPOSE

To report a case of a 31-year-old female patient with Stargardt's disease describing her clinical characteristics using multimodal analysis.

INTRODUCTION

Stargardt's disease (STGD) is the most common childhood recessively inherited macular dystrophy. This condition is characterized by mutations most commonly in the *ABCA4* gene, on chromosome 1, which encodes a retinal transported protein. It results in the accumulation of visual cycle kinetics-derived byproducts at the retinal pigmented epithelium (RPE) with secondary photoreceptor dysfunction.[1]

Males and females are equally affected as it is an autosomal disorder. Patients affected may be asymptomatic, and when symptomatic, bilateral central visual loss, photophobia, mild red-green dyschromatopsia, slow dark adaptation and central scotomas are the most common complaints.[1] Although there is no curative treatment for this disease, there are many lines of research on this topic. Clinical and diagnostic tests can confirm the disease and provide patients with an accurate prognosis.

METHODS

Medical records assessment.

CASE REPORT

R.C.B 31-year-old female patient is being monitored at Fundação Dr Joao Penido Burnier due to visual loss on both eyes that began 10 years ago. She reports nyctalopia that progressively worsened and slow dark adaptation.

Ophthalmological history: denies previous surgeries.

Family Background: Cousins with progressively visual loss

Visual Acuity

Right eye: 0.4

Left eye: 0.3

Biomicroscopy: No changes

Fundoscopy: Images

OD

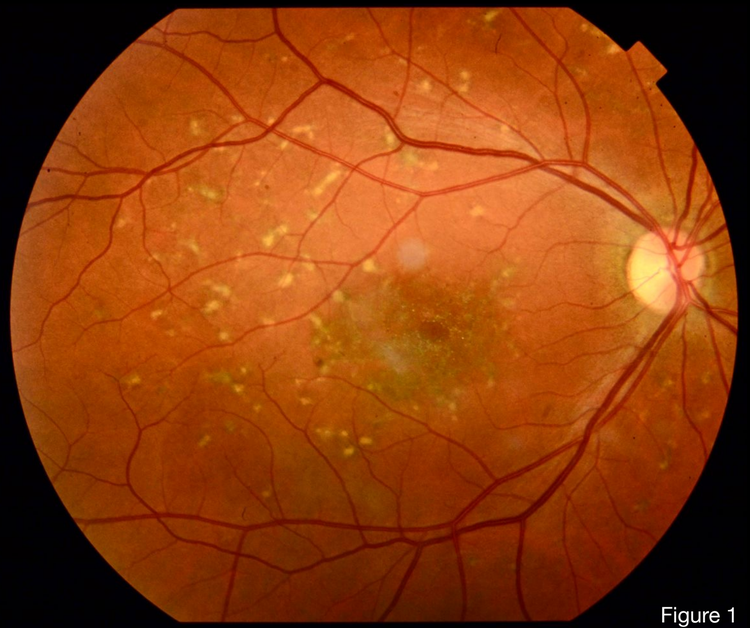
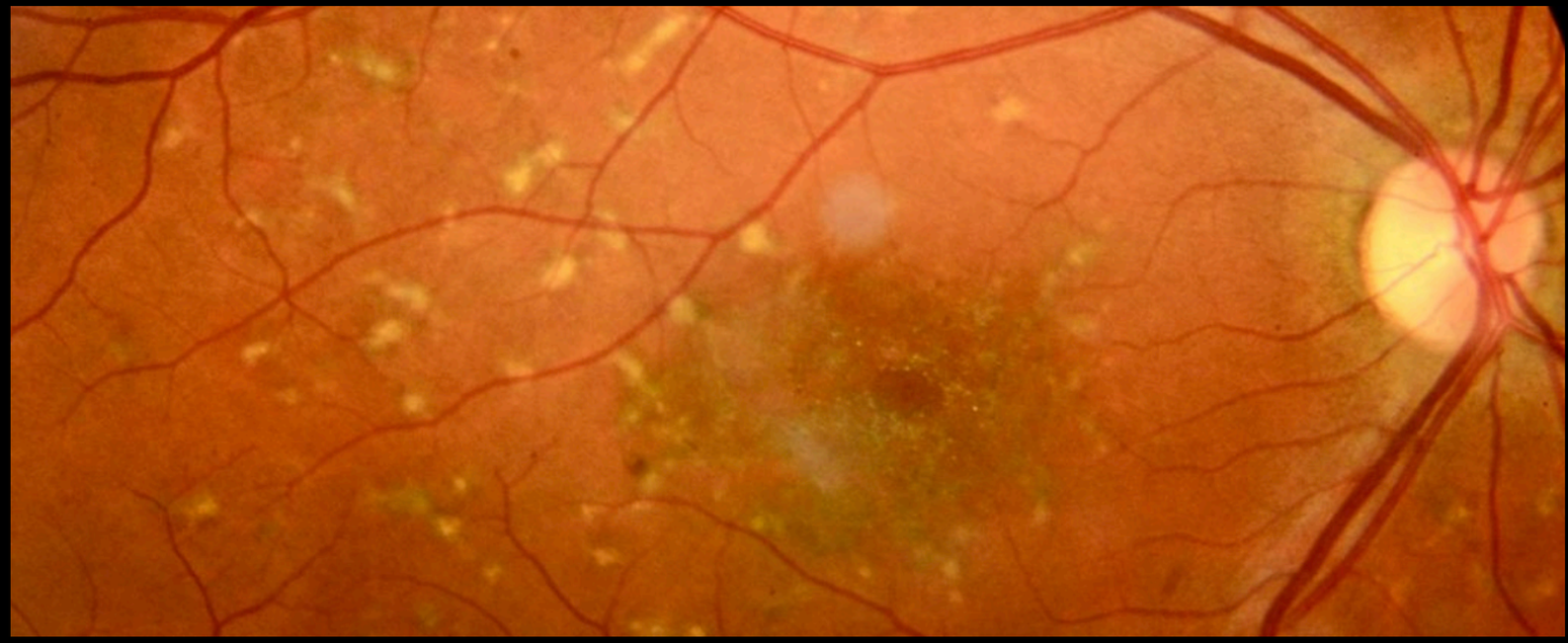


Figure 1



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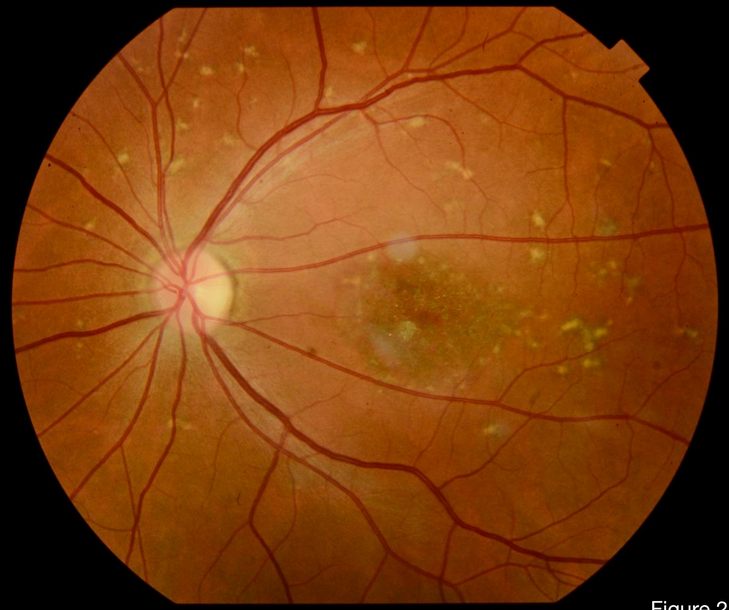

Figure 2

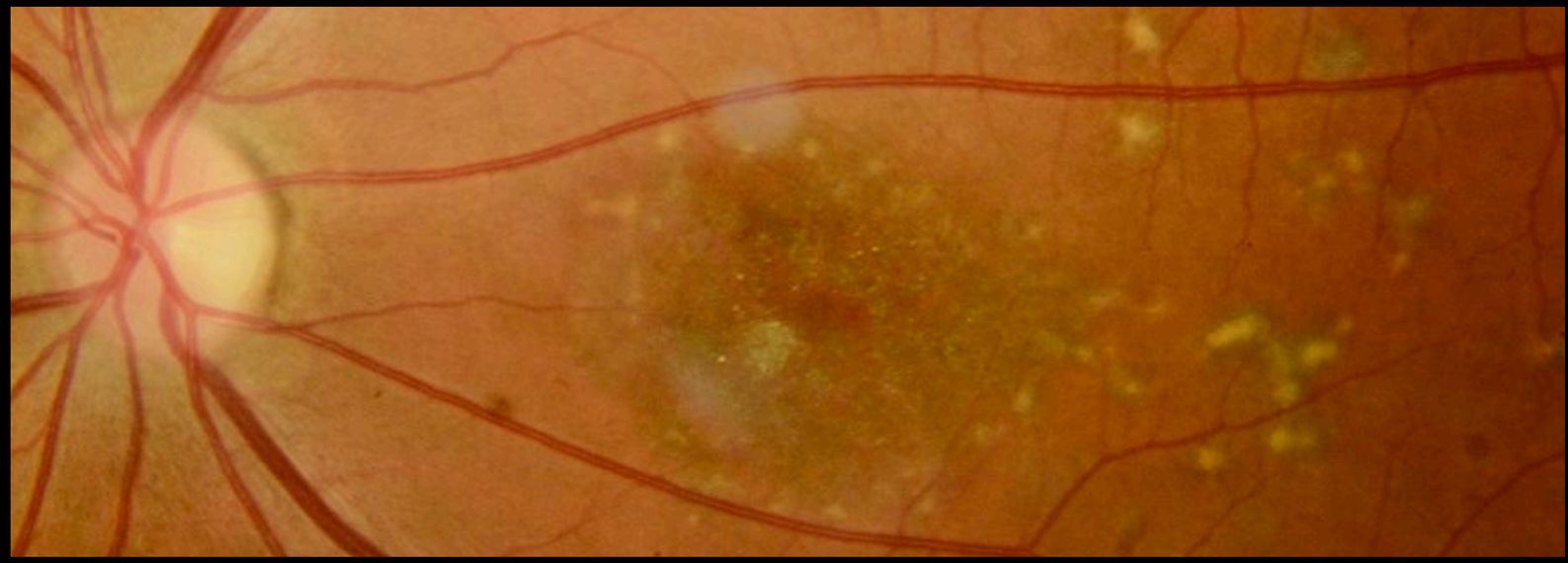

Figure1,2: Fundus photograph of both eyes shows the presence of multiple yellow flecks in the posterior pole

OD

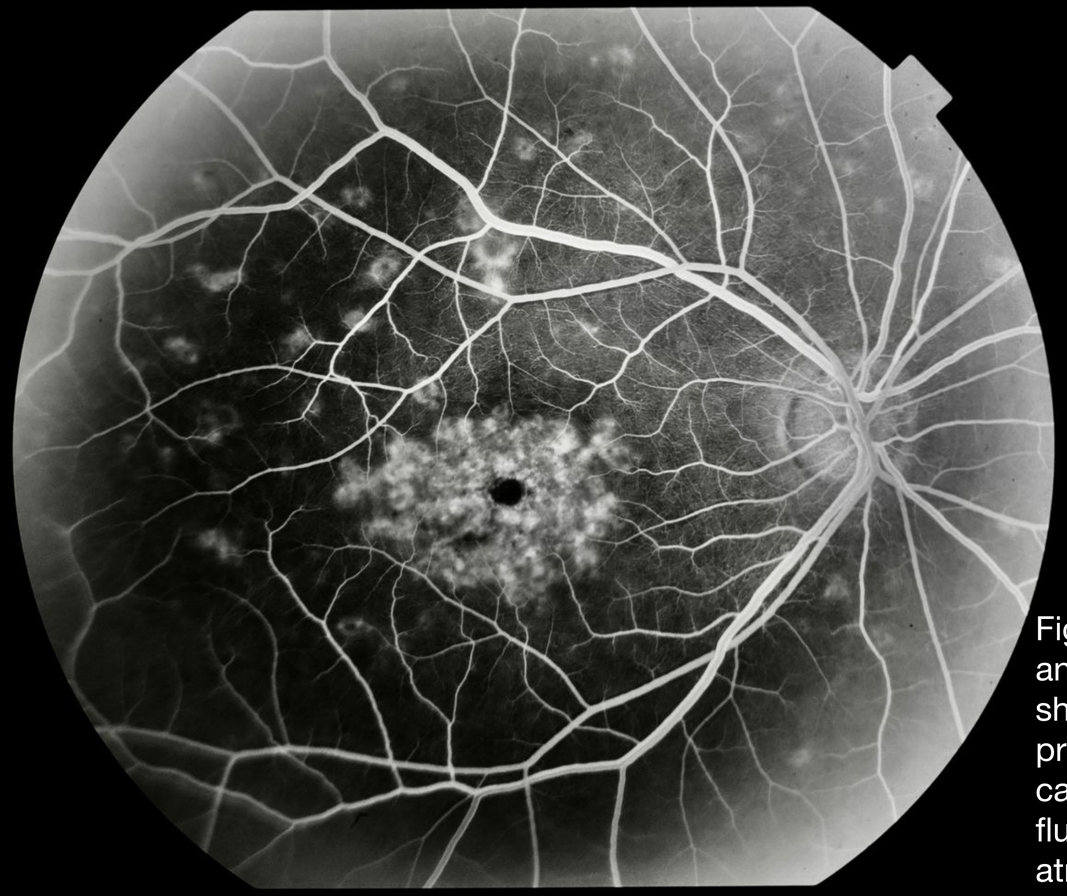

Figure 3

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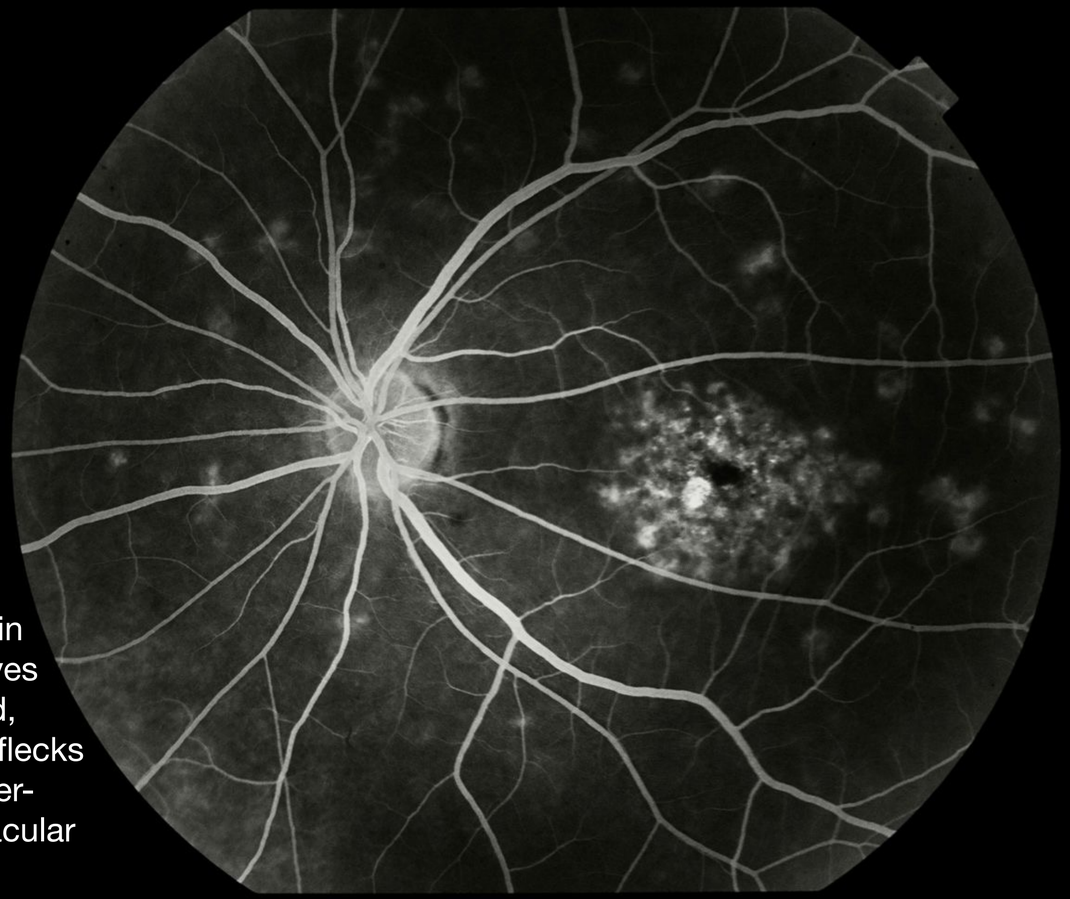

Figure 4

Figure 3,4: Fluorescein angiogram of both eyes showing dark choroid, presence of multiple flecks causing areas of hyper-fluorescence with macular atrophy

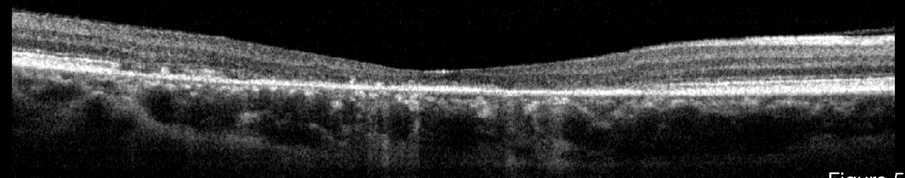

Figure 5

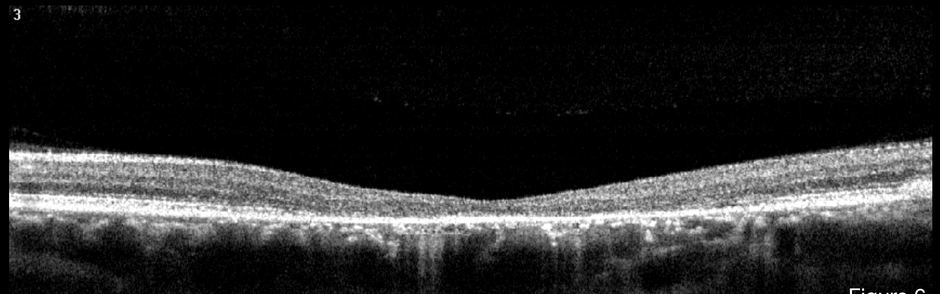

Figure 6

Figure 5,6: The OCT scan centered on the fovea shows thinning of the neurosensory retina, atrophy of RPE and ellipsoid zone

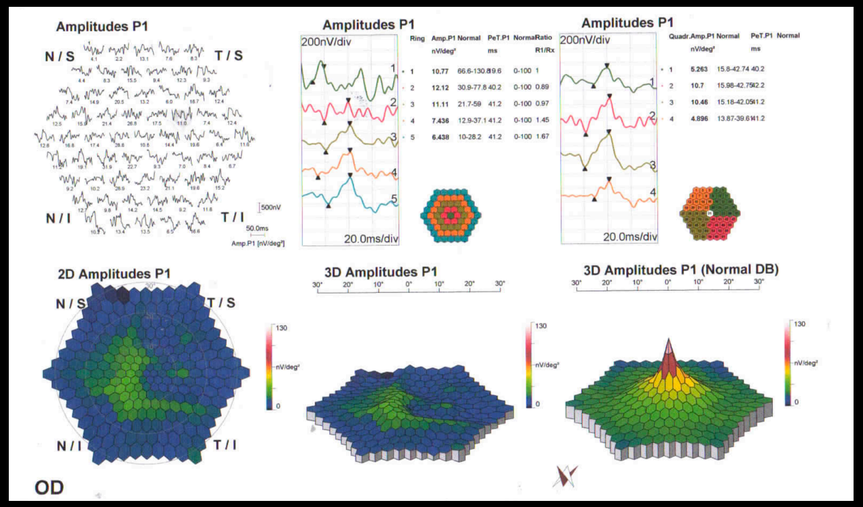

Figure 7

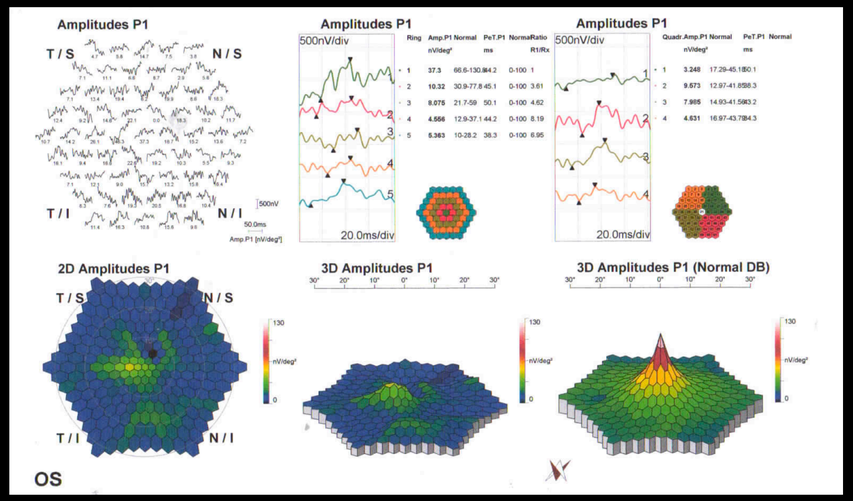

Figure 8

Figure 7,8: Multifocal ERG abnormality with generalized paramacular, centromacular and perimacular dysfunction

DISCUSSION

This pathology affects the posterior pole, with variable extension its diagnosis includes funduscopy, investigating family history, assessing visual acuity, visual field testing, fluorescein angiography, fundus autofluorescence, electroretinography and optical coherence tomography (OCT). [1]

Patients with Stargardt's complain of low vision from the initial stage, and as it is a relatively rare, its clinical diagnosis is subject.

The disease is chronic and incurable, so the treatment includes photoprotection with sunglasses to protect the eye and wear a hat are recommended as well as reducing vitamin A in your diet and advise patients to avoid smoking.

The treatment of the disease is not yet known and carrying out genetic testing is still difficult to do.
In addition to the concern with making the correct diagnosis, monitoring the progression through serial exams is important to offer guidance on the prognosis. According to the Fishman's classification[1], changes in the electroretinogram shows a more aggressive course and can be an ally when estimating the prognosis, just like an earlier onset is associated with a more aggressive form of the disease.[2]

Therefore, while there is no established treatment and there is difficulty in carrying out genetic testing, as in the case of the patient in the case report, it should be considered to routinely request an electroretinogram to provide guidance on visual prognosis, as patients with this abnormal test have a worse prognosis.[2 3]

Because of that patient education and you seeing are critical for the management of this disease. Talk about the prognosis it's important to counseling patients about their mental state and explain that the disease is progressive with a declining vision. It's necessary to inform about the low-vision and suports then.

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