Stargardt Disease: A Case Report.

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PURPOSE

Describe a case of stargardt disease

INTRODUCTION

Stargardt disease (STGD1) is an autosomal recessive retinal dystrophy due to mutations in ABCA4, characterized by subretinal deposition of lipofuscin-like substances and bilateral centrifugal vision loss. Low visual acuity often precedes funduscopic changes, varying according to age at onset of symptoms: the later the onset, the less likely visual loss.The diagnosis of Stargardt disease is based on clinical history and fundus changes keeping an eye out, complementary exams, such as retinography and optical evaluation tomography (OCT), play an essential role in the prognosis and evolution of the case

METHODS

Medical records review

RESULTS

Patient B. R. C., 16 years old, female, complaining of slow and progressive AV block since she was 10 years old. He mentions successive changes of corrective lenses in a short period of time, with no improvement in VA. With no other comorbidities, he refers to an aunt and brother with blindness, but without a defined diagnosis. On examination, the patient had a best-corrected VA of 20/400 AO. In the fundoscopic evaluation, the presence of fundus flavimaculatus and macular lesions in AO beaten bronze were observed (Figure 1 and 2). OCT was performed (Figure 3) which showed foveal deposits and changes in the ellipsoid layer of photoreceptors,

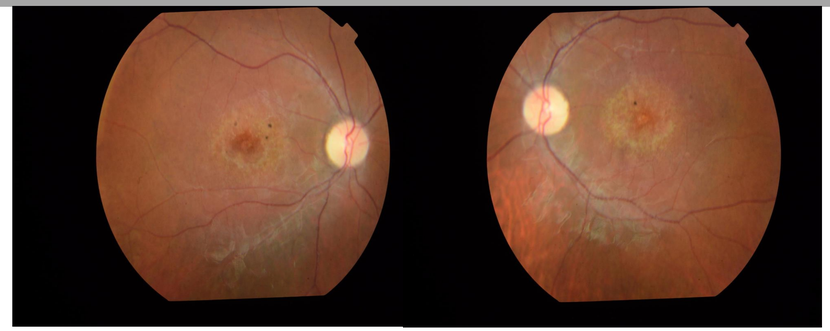

Fig. 1 and 2: retinography of the both eyes of the patient.

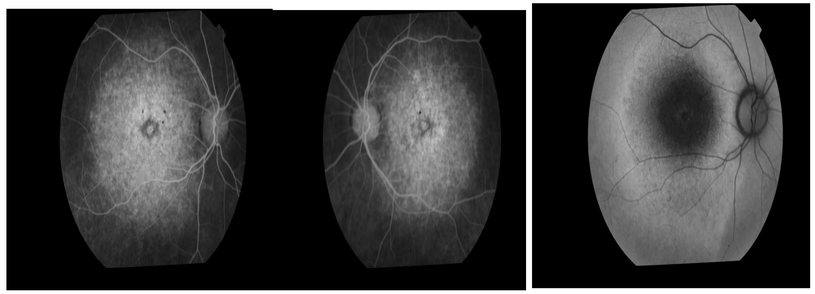

Fig. 3 and 4.Fluorescent angiograthy and right eye fundus autofluorescence

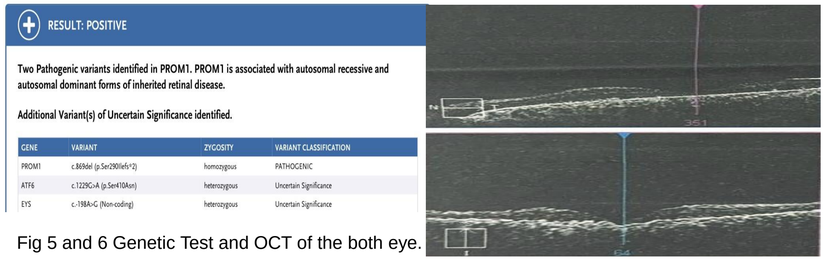

Fig 5 and 6 Genetic Test and OCT of the both eye.

in addition to marked foveal atrophy. Genetic test confirmed the diagnosis throught the pathogenic mutation of the PROMO 1 gene in homozygous. The pacient is still undergoing treatment and follow up at this service.

DISCUSSION

ABCA4 is a large, highly polymorphic gene, consisting of 50 exons, with over 900 disease-associated variants reported to date.This highly polymorphic nature and large number of variants make ascribing definite disease-causation problematic; moreover, the vast allelic heterogeneity makes genotype–phenotype correlations very challenging indeed. In general, missense variants are associated with milder, later onset disease. while null alleles are associated with more severe, earlier onset diseaseThere are many drugs that are already available or have been specifically developed that target different aspects of the visual cycle (vitamin A recycling pathway) and may thereby be poten- tially beneficial in slowing or stopping progression in STGD1

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