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STARGARDT'S DISEASE: A CASE REPORT

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ABSTRACT

Stargardt's disease is a bilateral symmetrical and progressive macular dystrophy that is transmitted in autosomal recessive or dominant pattern. It usually starts between the ages of 6 and 20 years and typically leads to a rapid loss of central vision.
The authors describe clinical features of a 28-year old male patient with fundus findings of *Stargardt's disease* whose symptoms were said to have started about 19 years prior to presentation. Previously there has been a single reported case in Nigeria.
We present this case that would be discussed along the lines of presentation, pathophysiology, management. We introduce an innovative and cost effective method of fundus examination using mobile phone technology.

Keywords: Stargardt's; Central vision; Mobile Phone.

INTRODUCTION

Karl Stargardt first described this condition in 1909 when he reported 7 patients from two families with a recessively inherited disease characterized by macular dystrophy surrounded by deep yellow-white lesions.[1]

Stargardt's disease is caused by mutations in the *ABCA4* gene, which encodes an ATP-binding cassette (ABC) transporter protein expressed by rod outer segments.[2] This genetic defect has a profound effect on the visual phototransduction cycle.[3] There is accumulation in the outer segment discs of of N-retinyl-ethanolamine(A2E) which is a component of lipofuscin that is toxic to the retinal pigment epithelium(RPE) and photoreceptors.4[5,6]

Subsequently, there is a degeneration of Photoreceptor and RPE cells by way of membrane permeability, lysosomal dysfunction and detachment of pro-apoptotic proteins leading to cell death and subsequent photoreceptor dysfunction/loss.3[4,7]

Stargardt's disease is rather a rare condition. However it is the most commonly inherited childhood and adulthood macular dystrophy[2,4] In the United States, it has an incidence of between 1 in 8000 and 1 in 10000.8 It has also been reported to account for 7% of all retinal dystrophies.[7] Till date there has been only one reported case in 2 Nigerian siblings.[3]

Stargardt's disease is caused by mutations in ABCA4 gene located on chromosome 1p21-p13.5 The most common inheritance pattern is autosomal recessive. However it can also be autosomal dominant.[3,9] Age at presentation varies even among siblings with onset being commonly in childhood and with another peak occurring in early adulthood and less frequently in later adulthood.[4] Evidence suggests that the severity of the disease is associated with age of onset of the disease with manifestation in childhood being more severe than with adult onset disease.[4]

Clinically, it is a heterogenous disorder.4 Initial symptoms include bilateral central visual loss characterised by blurred vision, central scotomas and/or

dyschromatopsia.4[5] On examination of the fundus, the macula may appear normal at the early stages or show non-specific mottling, progressing to an oval snail slime or beaten-bronze appearance. There may be yellowish pisciform deposits which may be absent especially if the illness has its onset from childhood.4 Subsequently there is geographic atrophy which has bull's eye configuration with or without Subretinal fibrosis.7[10]

CASE REPORT

A 28 year old male, AS genotype who is a sales representative and presented to our hospital on account of gradually progressive painless blurring of vision for distance and difficulty reading in both eyes for over 19 years. Ocular examination showed unaided distance visual acuity of 6/60 and a near visual acuity of N24 in both eyes. There was no significant improvement on refraction. Anterior segment examinations were normal in bilaterally.

Intraocular pressure was measured using Goldmann tonometer, being 12mmHg in the right eye and 13mmHg on the left side. Visual field was assessed using a Humphrey visual field analyzer and found a central scotoma in both eyes (figures 1 and figure 2). Dilated funduscopy and fundus photographs taken with a +20D lens and an Android phone with a camera FV-5 application showed a typical bronze beaten appearance. Systemic examination was unremarkable (figures 3 and 4). Systematic examination was unremarkable.Examination of one of his 5 other siblings did not show similar features. Patient was given telescopes and binoculars after low vision assessment and encouraged to wear sunglasses with ultra-violet ray protection and to avoid vitamin A supplementation.

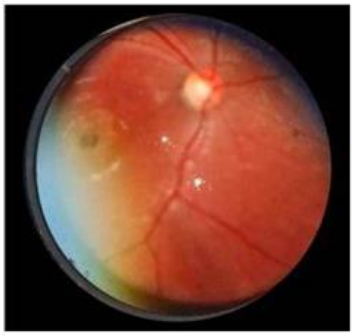

Figure 1: Right Fundal Picture taken with a mobile phone and +20D Lens

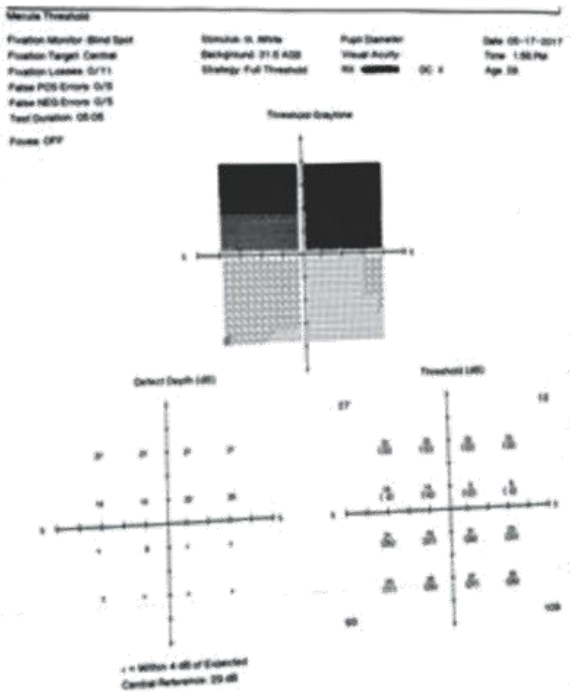

Figure 2: Right Macula Threshold Visual Field Analysis

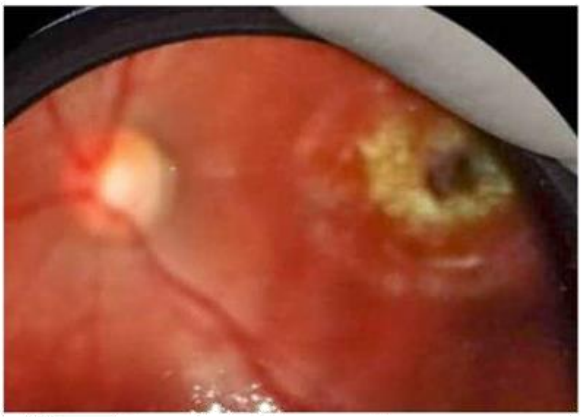

Figure 3: Left Fundal Picture taken with a mobile phone and +20D Lens

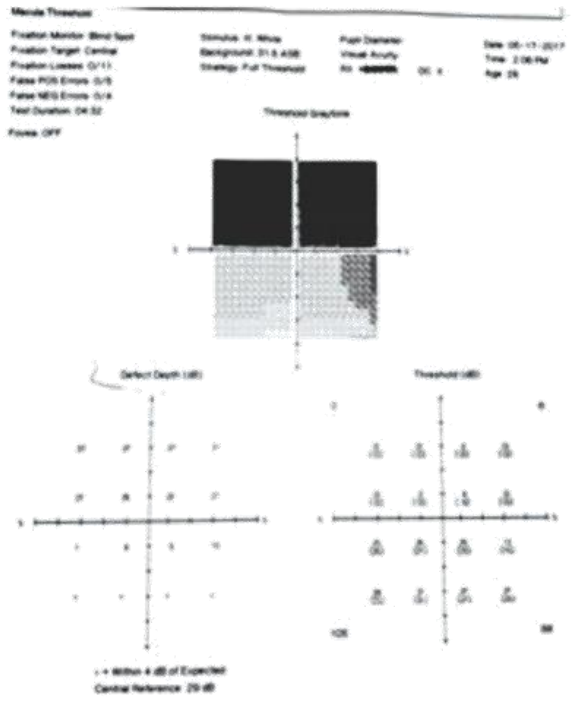

Figure 4: Left Macula Threshold Visual Field Analysis

DISCUSSION

Stargardt's disease is usually associated with marked dimunition of vision starting at an early age.[4] The patient presented had a remarkable decrease in vision for both distance and near objects since his teenage years which did not improve with regular refraction requiring the use of low vision aids to maximize his vision. [2,5] The index patient did not have any ocular or systemic associations. This was similar to the findings in the first case reported by Stargardt.[1] Though hereditary and can also affect other siblings, the only sibling (an elder sister of the patient) who volunteered to be examined showed no features suggestive of Stargardt's disease. Stargardt's disease is quite a rare condition and a high index of suspicion is essential in making the diagnosis.[2,4] The patient had a characteristic bronze beaten appearance of the macular surrounded by yellowish pigment deposits.

There could be atypical fundus findings which include pallor of the optic disc, attenuated retinal vessels, retinal pigmentation in the form of bone spicule, cicatricial chorioretinitis, retinal pigment hyperplasia, subretinal neovascularisation and fibrosis.[7] There were no systemic associations found in this patient.1 Once a diagnosis of Stargardt's disease is suspected, a more detailed history and examination are recommended. There are established traditional ways of examining the fundus and taking clinical photographs which might not be readily available especially in resource poor countries like Nigeria.

For this patient, fundus photographs were taken with a +20D lens and an Android phone with a camera FV-5 application. The rise in complexity and flexibility of application in mobile phone technology which is easy to use, has made possible detailed examination and documentation of the fundus affordable both in the clinic and outside the clinic settings.

For the patient, this also increases the ease with which such findings are transmitted to other specialists in different parts of the world thus allowing for detailed fundal examinations to be done in developing countries using a much less expensive but available mobile phone technology.

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