Case Report

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A Case of Late Onset Stargardts Disease

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Abstract

Purpose: We report a case of late-onset Stargardt's disease in a 51-year-old patient who came with bilateral, symmetrical decrease in the visualacuity .Stargardt disease is the most common form of juvenile macular degeneration. Clinically, it is characterized by pisciform flecks at the level of the retinal pigment epithelium and a bull's-eye maculopathy. This observation draws attention to the existence of Stargardt's disease at a late stage.

Keywords: Stargardts disease , Late onset ,Vision loss.

INTRODUCTION

Stargardt disease is the most common type of hereditary autosomal recessive macular dystrophy.1 The estimated incidence of disease is 1 in 10,000 (live births), and it characteristically presents in juveniles and young adults.[2] It is a bilateral and symmetrical maculopathy that progresses rapidly to macular atrophy and loss of central vision.It was described perfectly in 1909 by the German physician Karl Stargardt. As a particularity, it is a disease that can be transmitted according to the autosomal dominant mode, in late forms of the fifties, and even forms associating neurological disorders are noted.[3] Patients begin to experience a bilateral, gradual decline in their vision between the ages of 6 and 20 years in classical form. This condition affects retinal pigment epithelium . In people with Stargardt disease the RPE collects lipofuscin, which can lead to vision problems.[4] A small number of families were found to have an autosomal dominant pattern of inheritance. This form is called a "stargardt like" disease, and looks similar to the autosomal recessive form. These two forms are actually different diseases with different mechanisms. Autosomal recessive Stargardt disease is caused by mutations in a gene called ABCA4. A second gene called ELOVDT has been found to be the cause of the autosomal dominant form of Stargardt-like disease. Severity of disease may be related to how severely a gene change affects gene function.[1] The late onset of the disease is not exceptional; the adult form (third and fourth decade); the atypical form (fifth and sixth decade) can also be encountered. Visual acuity is the first functional sign; classically, it falls in a few years and stabilizes after 4 or 5 years .[3]

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CASE REPORT

We report a case of 51-year-old patient who came for consultation for decreased far vision since 1 year. No significant systemic history was present. Visual acuity was counting fingers at 1 meter in both eyes. Red green color defect noted on color vision test on ishihara chart. The anterior segment examination on slit lamp was normal. The ocular tension taken with the applanation tonometer was 16 mm Hg. The color vision tested with the Ishihara Test showed a red-green disorder. Dilated examination onindirect ophthalmoscopy (fundus image– figure1) showed elongated light colored piscifom cluster of flecks at the RPE, RPE atrophy at the macula seen bilaterally, obscuration of choroidal vessels and hyperpigmentation was noted, Oct showed (figure 2) atrophic changes and thinning of retinal layers at the macula.

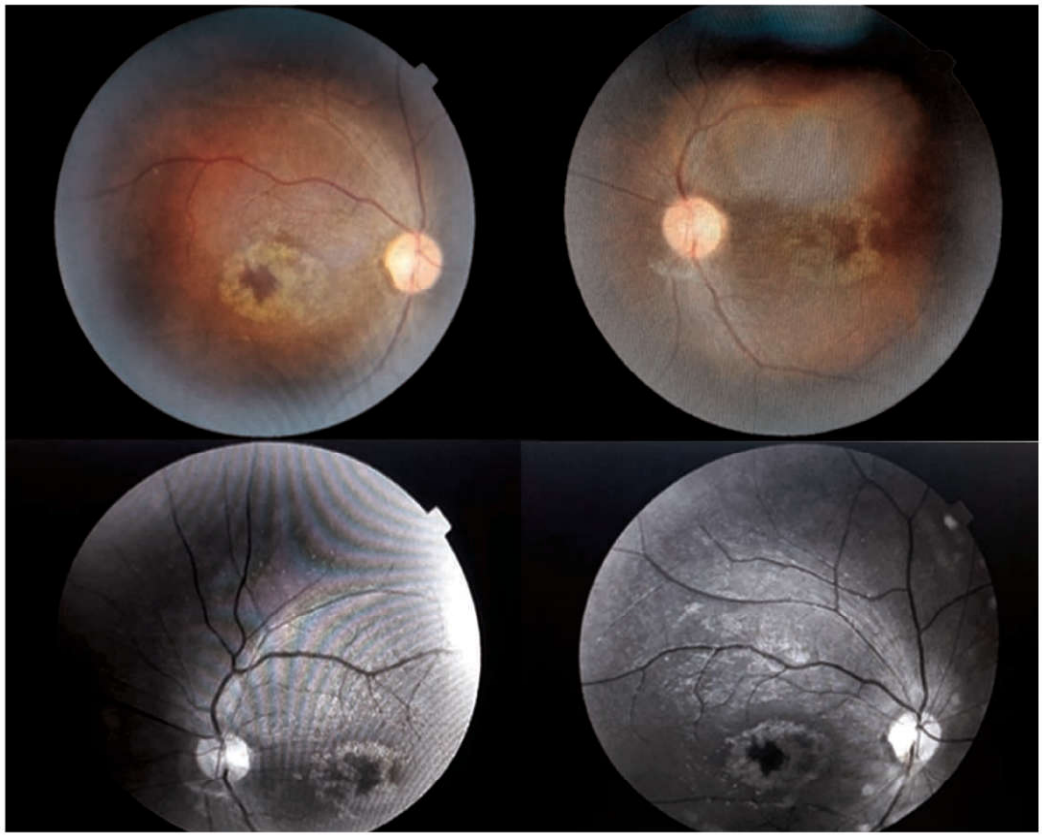

Figure 2: OCT images .

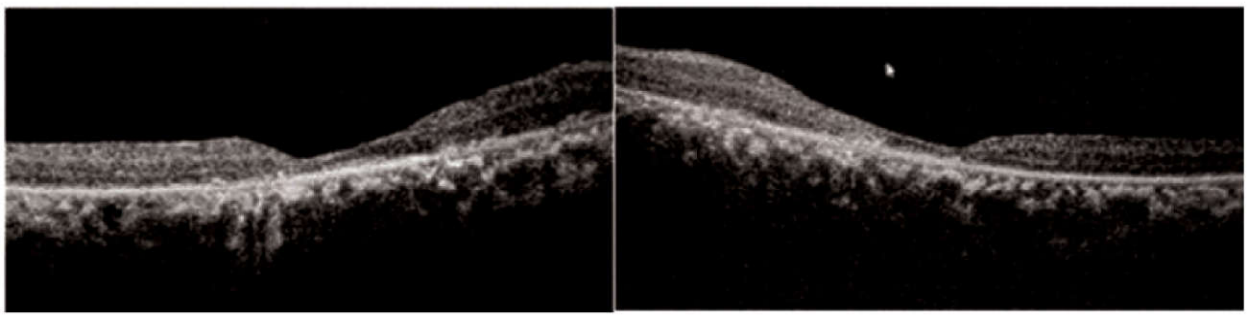

Figure 2: OCT images .

DISCUSSION

In our case,non specific mottling was seen at the fovea. Oval macular lesion about 1.5 disc diameter in size was seen giving the appearance of Bull's eye maculopathy. Macular lesion surrounded by yellow white flecks was noted. Atrophic

changes in the RPE and secondary changes in the photoreceptors.[1] Patients with Stargardt's disease usually have a poor visual prognosis. However, in a study conducted by Nakao and al, Japan in 2012, they observed in some patients despite the dark red foveal pigmentation, good visual acuity corrected despite the presence of the disease at an advanced stage.[5] In a study done in Pakistan in 2008 by Shah and al, it shows that patients with Stargardt's disease respond well to magnification (magnifying glass). Thus, simple bifocal glasses can be used at the first stage of the disease.[6] In our study, we found that our patient had difficulty to recognize red color as well as green; this is the finding made by Mäntyjärvi et al. where the defect of the red color became stronger at the advanced stage of the disease. Stargardt's disease has always been considered to be transmitted autosomal recessive according to several literatures.[7] In a 1988 study conducted by Weber et al., they present an unusual example of the dominant character of Stargardt's disease from a family tree study of an extended family of ten members.[8] At present there is no cure for Stargardt disease and there is very little that can be done to slow its progression. Wearing sunglasses to protect the eyes from UVa, UVb and bright light may be of some benefit. U-V blocking sunglasses are generally recommended for outdoors. For people who already have significant vision loss, low vision aides are available.[1,9,10]

CONCLUSION

Stargardt's disease is bilateral and symmetrical hereditary maculopathy, which varies in presentation. Stargardt disease with late onset is rare but exists. The management of the condition remains palliative (wearing tinted glasses, magnifying glasses, psychological support, and genetic counselling)

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