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Case Report

Stargardt's Disease (Fundus Flavimaculatus)

Healthcare

Keywords: Stargardt's disease; bilateral visual loss; progressive; children..

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Abstract

Background: Stargardt's Disease is included in the group of degenerative macular diseases, which consists in progressive lost of cones in fovea of both eyes, leading to variable levels of central vision loss. It is symptomatically similar to age-related macular degeneration and it affects approximately one in 10,000 children, in ages between 7 and 12 years old. Case Report: We report a case of a 13 years old boy who presented with severely reduced bilateral visual acuity. He was admitted to handicapped children school, since he was suspected of malingering. He had no family history or previous ocular symptoms. His visual acuity was 20/200 s.c à 20/50 in midriasis. Fluorescein Angiography, with the characteristic aspect of "bull's eye" due to a "window effect" at the level of the perifoveolar despigmentation limit, was very helpful to decide the right diagnosis.Conclusion: Currently, there is no effective treatment for Stargardt's disease, but having the right diagnosis may assist the patient, family members and the society to adapt helpfull behaviours. Individuals benefit from the use of low vision aids and orientation and mobility training.

Introduction

Stargardt's disease (also known as fundus flavimaculatus and Stargardt's macular dystrophy) is the most common form of inherited juvenile macular degeneration. For a long time, it was believed that Stargardt's disease and fundus flavimaculatus were separate entities. However, persons with and without macular involvement have been found in the same pedigree. Usually it is inherited as an autosomal recessive, but it can be autosomal dominant. Stargardt's Disease is included in the group of degenerative macular diseases, which consists in progressive lost of cones in fovea of both eyes, leading to variable levels of central vision loss. The recessive pattern, which includes more than 90% of cases, is due to a defect at the chromosome 1q21-p13. The dominant pattern seems to be related to a change at chromosome 6, but some studies also reported the location on chromosome 12. The altered gene seems to be responsible for the transcription of protein that works like an outer molecular pump.

At fundus oculi, there's often the presence of yellowish flecks around the macula, a condition called *fundus flavimaculatus.* The foveal damage is possibly due to a defect on an outer molecular pump that would eliminate a specific substance from cones. This molecule builds up inside the rods, accumulating as a form of lipofuscine, which penetrates in the RPE, acting as a poisoning substance for this layer, making it susceptible to lysis. The photoreceptor layer in the fovea demands an enormous fagocytic and metabolic activity from the RPE (much greater than in periphery), what makes the atrophic changes in Stargardt's Disease to be found mainly at foveal area. Fundus Flavimaculatus happens as a consequence of the atrophic epithelium of the central area, that becomes hyperfluorescent, and the still intact epithelial cells immersed in lipofuscin, which absorbs the blue excitatory wave-lengths, making this perifoveal region hypofluorescent in pisciform shapes (corresponding to the fresh flecks at fundoscopy).

The main symptoms

Bilateral, decreased central vision in childhood or young adulthood. Visual acuity may start at the 20/40 level and later decline to 20/200 or slightly worse. Central scotoma (blind spot in central vision), abnormal color vision and photophobia are usually present. We should suspect of Stargardt's Disease in every young person

presenting with a history of progressive visual acuity loss, mainly if there was a family member with the same complaints.

After a visual acuity evaluation test, the following exams should be performed:

1. Ophtalmoscopy - It will show loss of foveal reflex and granulous aspect of RPE cells. The most characteristic finding are the yellowish flecks around the macular area, the fundus flavimaculatus .
2. Fluoresceinic Angiography – It is of great diagnostic value and it will show:

* The characteristic aspect of "bull's eye" due to a "window effect" at the level of the perifoveolar despigmentation limit,
* The coroidian silence, due to a mask effect in the periphery of retina, appearing as a shadow hypofluorescent caused by lipofuscine deposits in the peripheral retina .
* The Diffuse form of fundus flavimaculatus where the peripheral retina will appear hyperfluorescent. (Fig 5)

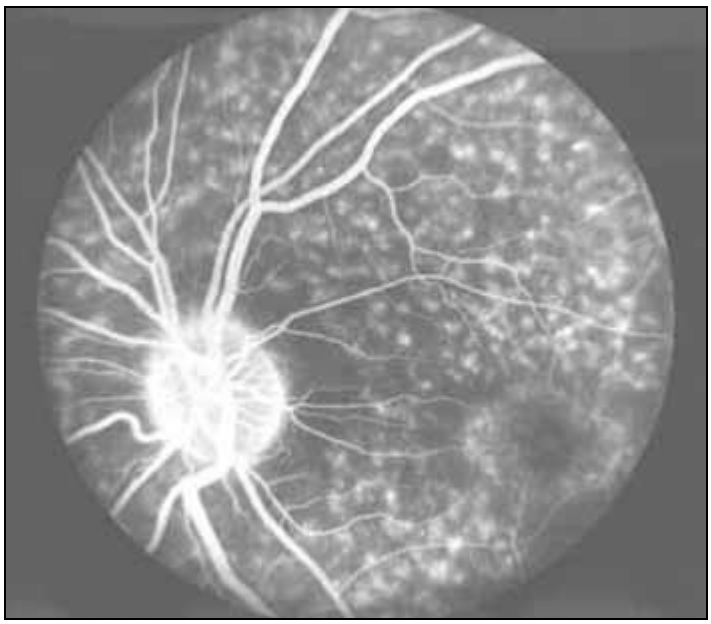


Figure nr. 5

Electric Diagnostic Tests - The Electro-oculography (EOG) is normal in early stages. As the RPE degeneration becomes more widespread, the EOG tends to show abnormality. The ERG * is normal or manifests only a slight delay in obtaining an otherwise normal maximal B-wave amplitude. Prolonged periods of dark adaptation can also be found.

Case Report

A.R is a 13 years old boy , who presented at the Eye Clinic, UHC "Mother Theresa",Tirana, Albania. He complains about severely reduced bilateral visual acuity.

His parents refer that they have noticed a different behaviour of their son four years ago. A.R refers gradual progression of reduced visual acuity. He is consulted from an ophthalmologist, but it was found no correlation with any ocular disease.

He had no family history of eye diseases, no previous ocular symptoms, a normal intrauterine development and a normal childhood till the age of 9 years.

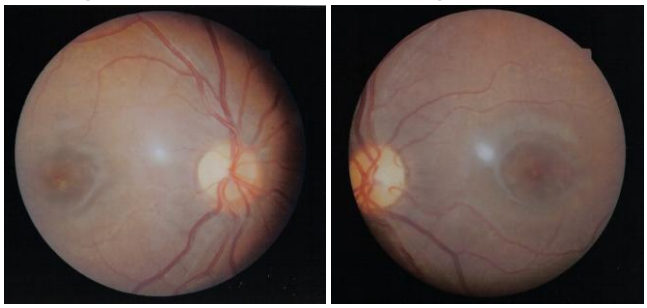
Suspected for malingering, he was admitted to handicapped children school at the age of 10. He was referred to our clinic for further investigations.

Examination of the eye: Visual acuity was 20/200 and 20/50 in midriasis with extracentric fixation. Slit lamp exam showed a normal conjunctiva, quiet anterior segment, transparent media. Pupils were round and reacting well to the light stimuli.

The color plate Ishihara test detected a mild red-green discromatopsia.

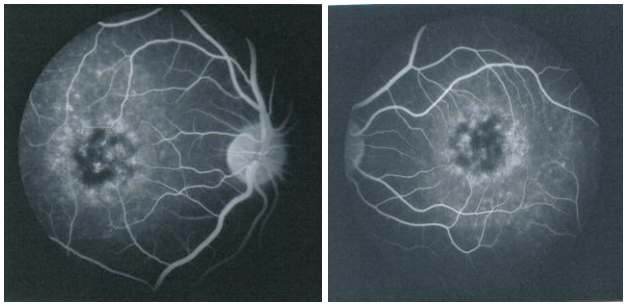
The fundus exam showed a normal optic nerve head had normal configuration, normal appearance of blood vessels, "beaten-bronze" foveal appearance and RPE degeneration. (Fig. 1-2)

Figure No. 1 **Figure No. 2**



The fluoresceine angiography was performed and showed macular hiperfluorescence due to a window defect and a "dark choroid", "bull's eye". (Fig. 3-4).

Figure No.3 **Figure No.4**



The visual fields tests were not reliable due to low performace. EOG and ERG couldn't be performed in our clinic, but based on what he concerns, history and our examinations, the diagnosis was Stargardt's Disease.

Management of the patient

The parents were advise to treat their child as a normal child with a visual function defect and support him to mentally adapt to the new condition. The child was advised to use magnifiers in order to enhance his life performance, to use sun glasse and to present to follow-up visits as advised.

Discussion

Stargardt's disease is usually diagnosed in individuals under the age of twenty, when decreased central vision is first noticed. It causes a progressive loss of central vision and, in the early stages, patients may have good visual acuity, but they may experience difficulty with reading and seeing in dim lighting. Other common symptoms of Stargardt's disease include blurriness and distortion. On examination, the ophthalmological findings vary

significantly with the progression of the disease. In fundus photos, patients with early Stargardt's disease appear to have simple macular degeneration. Children with the disease typically begin experiencing dark adaptation problems and central vision loss between six and twelve years of age, but symptoms may also first appear in adulthood. It is not caused by injury, infection or exposure to a toxic agent and it is not a malingering.

Conclusion

There is no treatment that proved to improve the visual loss or to retard the progressing of the disease. There are a few measures that should be adopted and that will help the patients solving some of the problems that they face. There are many kinds of binocular lens, either for improving near vision or distance vision, increase the visual field, protect against the light, watching, looking at the blackboard and improving the reading of the patients; the choose of the most useful lens for each patient should be performed by the physician and the patient together. The affected persons should learn to read in Braille, and have the other specific management for low vision conditions. A great number of the affected patients will be classified as owing legal blindness. Some doctors recommend the use of vitamins, in special A, E, C, Selenium and Betacarotene.

The level of visual acuity loss will depend on each case, starting to progress in tender ages and determining variable levels of visual defect, from discrete loss to complete lack of detail vision, with the presence of big central scotomas extending to peripheral areas. Complete blindness is a very rare event in the natural course of Stargardt's Disease. Because Stargardt's disease is an inherited condition, there is nothing that can be done to reduce the risk of developing the disease. However, recent findings in rodent models of Stargardt's disease find that unprotected, prolonged exposure to light can accelerate vision loss. Therefore, The Foundation Fighting Blindness strongly recommends that patients with Stargardt's disease wear brimmed hats or visors and sunglasses when outdoors.

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