

Case Report

Stargardt Disease with Late Revelation: Case Report

Yogolelo Asani Bienvenu^{1*}, Kintadi Luyingila Ginevra¹, Makumyaviri Mbuiro Julien¹, Babu Sapu Rebecca¹, Tambwe Ndumb Herve¹, Omewatu Mungomba Beaudry², Alfani Binti Lungwe Marie-Ange³, Kilangalanga Ngoy Janvier⁴

¹Eye Department, University Clinics Lubumbashi, DR Congo

²Pathology Department, University Clinics Lubumbashi, DR Congo

³English Department, Higher Pedagogical Institute, DR Congo

⁴Eye Department, Saint Joseph Hospital Kinshasa, DR Congo

*Corresponding author: Yogolelo Asani Bienvenu, Lecturer, Eye Department, University Clinics of Lubumbashi, DR Congo. Tel: +243814095671; Email: hassanyogo@yahoo.fr

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Abstract

The authors report a case of late-onset Stargardt's disease in a 56-year-old patient diagnosed for the first time at Lubumbashi University Clinics after bilateral, symmetrical visual acuity and fundus Flavoured tasks at the posterior pole of both eyes. This observation draws attention to the existence of Stargardt's disease at a late stage.

Keywords: Hereditary maculopathy; Late revelation; Stargardt's disease.

Introduction

Stargardt's disease is a bilateral and symmetrical maculopathy that progresses rapidly to macular atrophy and loss of central vision. It is an autosomal recessive disease that was isolated and 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; we can meet late forms of the fifties, and even forms associating neurological disorders. With the angiography (1970 to 1993), three essential diagnostic signs were identified, namely Bonnin's choroidal silence, the alteration of the pigmentary epithelium and the presence of the flavimaculate spots.

The frequency of this disease was 1/10000 in 1988 and today it is much more common in the North of France. The disease is diagnosed by a decrease in visual acuity at the beginning of childhood between 7 and 15 years; 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. Decrease of vision is the first functional sign; classically visual acuity falls in a few years and stabilizes after 4 or 5 years between 1 and 3/10 allowing reading closely. Vision continues to decline

more slowly up to 1/20 in more severe cases after twenty years of evolution. Visual field test shows a central scotoma more or less deep of 5 to 10° which can even go up to 15° in the later forms; the peripheral visual field is normal. Colour vision is often altered early and is paradoxical of red-green axis and blue-yellow axis. If visual acuity is less than 1/10, the colour vision will always be altered.

Fundus is normal at the early stage and disorients the diagnosis; the maculopathy appears quite quickly and the first signs are the reflections in «snail slime» on its surface or a reddish succulent colour called «vermilion». At the state stage, the centre of the macula is the site of xanthophyll pigmentation. In the late stage, the macula is the site of chorioretinal atrophy and a significant remodelling that can be pigmented. Positive diagnosis is made through ophthalmoscopy and angiography; the possible differential diagnoses are: macular dominant dystrophy, cone dystrophy, autosomal dominant spotted dystrophy simulating the fundus flavimaculate, mixed dystrophies with altered electroretinogram and inverse pigmentary retinopathy.

Nowadays, no treatment is available [1]. However palliative measures have been suggested (wearing tinted glasses, magnifying glasses, restriction of vitamin A supplements, psychological support, genetic counselling) [2] (Figure 1).



Figure 1: Retinogram right eye: macular atrophy, foveola of \pm oval aspect (in snail slime) with slight pigmentary remodelling, flavimaculate spots at the posterior pole.

Patient and observation

A 56-year-old man who presented at the clinic for decreased far vision for about 8 months; he was a teacher. No history was reported. The best corrected visual acuity (BCVA), both eyes were 1/10 with fault and he read Parinaud 3 with difficulties for near vision. The slit lamp examination of the globe was normal. Intraocular pressure (IOP) measurements were 17 mm Hg in both eyes. The colour vision tested with Ishihara Test showed a red-green axis disorder. Fundus photography showed the appearance of the macula in snail slime with pigmentary remodelling and the presence of flavimaculate spots at the posterior pole in both eyes (Figure 2).



Figure 2: Retinogram left eye: macular atrophy, foveola of oval aspect (snail slime) with pigmentary remodelling and flavimaculate spots at the posterior pole.

Discussion

Stargardt's disease is inherited macular dystrophy transmitted by the autosomal recessive mode; the disease is rare in sub-Saharan Africa region according to a study conducted by Tunji, et al. in Nigeria [3]. Our patient was probably the first case to be diagnosed in adults in Central Sub-Saharan Africa area. Patients with Stargardt's disease usually have a poor visual prognosis [4]. However, Nakao, et al. in Japan [2], observed in some patients despite the dark red foveal pigmentation, good corrected visual acuity despite the presence of the disease at an advanced stage; this is not our case [5]. Shah, et al. showed in their study that patients with Stargardt's disease respond well to magnification (magnifying loupe) [3]. This, simple bifocal glasses can be used at the first stage of the disease; such is the management scheme we suggested to our patient [6].

In this report, we found that our patient had difficulty to recognise red colour as well as green. Mäntyjärvi et al found the same result, where defect of the red colour became stronger at the advanced stage of the disease. Stargardt's disease has always been considered to be transmitted by an autosomal recessive way according to several studies [7]. Bither, et al. presented an unusual example of the dominant character of Stargardt's disease from a family tree study of an extended family of ten members [8].

Conclusion

Stargardt's disease is bilateral and symmetrical hereditary maculopathy which varies in presentation. Stargardt disease with late onset is rare but do exist. The management of that condition remains until now palliative (wearing tinted glasses, magnifying glasses, restriction of vitamin A supplements, psychological support, and genetic counselling).

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Ethical aspects

A patient's parent offered a verbal informed consent to this publication and we preserved his anonymity. The Ethics Committee of the University of Lubumbashi endorsed this study.

Statement

The manuscript has been read and approved by all the authors, the requirements for authorship as stated earlier in this document have been met, and that each author believes that the manuscript represents honest work.

Declaration of Interests

The authors declare that they have no conflict of interest in relation to this article.

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