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Short Communication

Ocular Signs in Fabry Disease

Mehdi Khamaily, Joumany Brahim Salem, Imane Tarib, Dahi Sidi, Salma Bajjouk, Rachid Zerrouk, Yassine Mouzari, Fouad El Asri, Karim Reda and Abdelbarre Oubaaz*

Department of Ophthalmology, Mohammed V Military Teaching Hospital, Morocco

Abstract

Fabry disease is a rare, hereditary disease characterized by a deficiency of an enzyme, α galactosidase A (α gal A), responsible for progressive damage to many organs, leading to various symptoms, Ocular damage, particularly to the cornea, is sometimes a precious element helping the positive diagnosis of the disease. We report the case of a 40-year-old patient diagnosed with Fabry disease, with bilateral conjunctival vascular toruosities, a "cornea verticillata and a peripheral cortical cataract.

Better knowledge of ophthalmological signs, allows better screening and can participate in the evaluation of the effectiveness of substitute therapy.

Keywords: α galactosidase A; Cornea verticillata; Hereditary; Farby disease

Introduction

Fabry disease is a rare, hereditary disease, the transmission of which is linked to the X chromosome. It was first described in 1898 by Johannes Fabry and William Anderson [1].

It is characterized by a deficiency of an enzyme, α galactosidase A (α gal A), contained in lysosomes. This deficit causes the accumulation within the lysosomes of a glycosphingolipid, Globotriaosylceramide (GB-3), responsible for progressive damage to many organs, leading to various symptoms: ocular, digestive, cutaneous, cardiac, renal, and pulmonary but also involvement of the central and peripheral nervous system [2]. The disease develops gradually and the diagnosis is often made late.

Ocular damage, particularly in the cornea, is sometimes a precious element helping the positive diagnosis of the disease.

Materials and Methods

We report a case of a 40-year-old patient, diagnosed with hypertrophic heart disease, and Fabry disease since 2012, and who consults in ophthalmology as part of his general check.

Results

The patient underwent a complete ophthalmological examination which revealed better visual acuity corrected to 16/20 OSD, a normal shirmer test, in the slit lamp: bilateral conjunctival vascular toruosities (Figure 1), with a "cornea verticillata and a peripheral cortical cataract (Figure 2), normal Intra Ocular Pressure and fundus with no vascular dilatation.

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***Corresponding author:** Mehdi Khamaily, Department of Ophthalmology, Mohammed V Military Teaching Hospital, Faculty of Medicine and Pharmacy, Mohammed V University Rabat, Morocco, E-mail: mehdi.khamaily@gmail.com

Discussion

The most distinctive and common clinical feature of FD (90%) is whorl-shaped bilateral keratopathy [3], known as cornea verticillata, secondary to GB-3 deposits at the level of the epithelial basement membrane, and visualized as yellowish-brown inclusions emanating radially; the corneal stroma and the endothelium are spared [4]. Confocal microscopy has a great interest in the detection of subclinical lesions, and can represent a reliable tool for the early diagnosis and the follow-up of the disease [5]. In children, the presence of eye signs, especially the cornea verticillata, is correlated with a severe form of the Fabry disease [6].

Conjunctival involvement is a tortuosity of the conjunctival vessels secondary to a disturbance of the endothelial architecture by accumulation of GB-3. The lens involvement is represented by two types of clouding, the "classic" Fabry cataracts which result from deposits of GB-3 in the lens epithelium and which are better visible in back-illumination, which appear as cloudiness under-capsular or dendritic away from the visual axis, and cataracts in "Helices" which are in rays oriented radially in the equatorial region or in sub capsular [7].

These cataracts appear in the second decade of life and are seen in up to 70% of men, less often in women [8].

Dry eyes are present in approximately 50% of patients with Fardy's disease, following deposits of GB-3 in the lymph nodes and directly in the lacrimal gland [9]. Other eye damage described but much less rare, corneal edema, conjunctival chemosis, and chronic uveitis [10].

Conclusion

Fabry's disease remains potentially serious due to its systemic damage, and despite the frequency of ocular damage, which is a precious tool for positive diagnosis, the visual prognosis is very rarely involved.

Better knowledge of ophthalmological signs, allows better screening and can participate in the evaluation of the effectiveness of substitute therapy.

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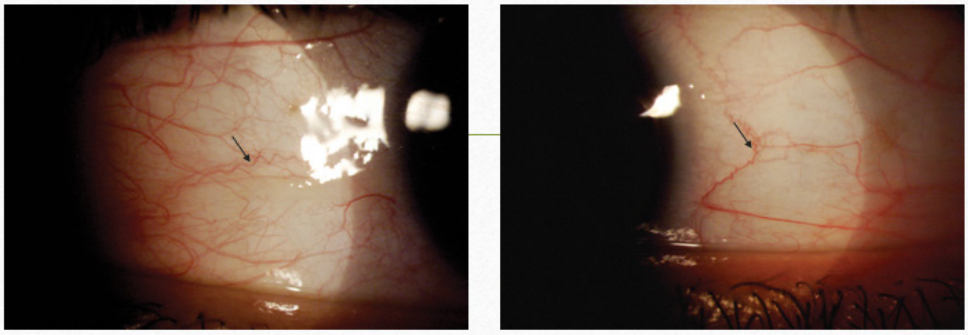

Figure 1: Tortuosities of the conjunctival vessels in the two eyes (arrows).

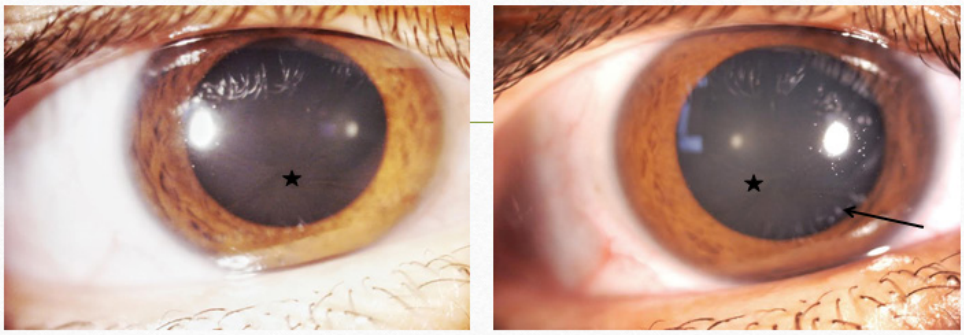

Figure 2: Cornea verticillata (stars), Peripheral cortical cataract (arrow).

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