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Ocular Manifestations of Fabry Disease

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Abstract | **Case Report**

Fabry disease (FD) is an X-linked lysosomal storage disorder caused by deficient α-galactosidase A activity, leading to systemic glycosphingolipid accumulation [1]. Ocular findings (such as cornea verticillata and conjunctivalor retinal vascular tortuosity) are frequent and may precede or accompany renal, cardiac, and cerebrovascular disease. We report the case of a 35 years old male admitted for a renal failure which the ophthalmological exam oriented the diagnosis toward Fabry disease.

Keywords: Fabry disease – Cornea verticillate – renal failure.

INTRODUCTION

Fabry disease (FD) is an X-linked lysosomal storage disorder caused by pathogenic variants in the *GLA* gene, which encodes the enzyme α-galactosidase A. Deficiency or dysfunction of this enzyme leads to progressive intracellular accumulation of glycosphingolipids within various tissues and organs. This accumulation underlies the multisystemic manifestations of the disease, involving the renal, cardiac, neurological, and ocular systems. [2].

Renal involvement represents one of the major clinical features of FD and may range from isolated proteinuria to progressive chronic kidney disease and, ultimately, end-stage renal failure. [3]. Ophthalmic manifestations are among the earliest and most consistent clinical indicators of FD.

CASE REPORT

We report the case of 35 years old male admitted for a renal failure who reported bilateral blurry vision. The ophthalmological exam revealed better visual acuity corrected to 18/20 0SD. The slit-lamp exam: bilateral epithelial corneal deposits radiating from the central cornea consistent with cornea verticillate (Fig 1). The fundus examination showed normal optic discs and maculae with moderate tortuosity of retinal arterioles and venules. (Fig 2)

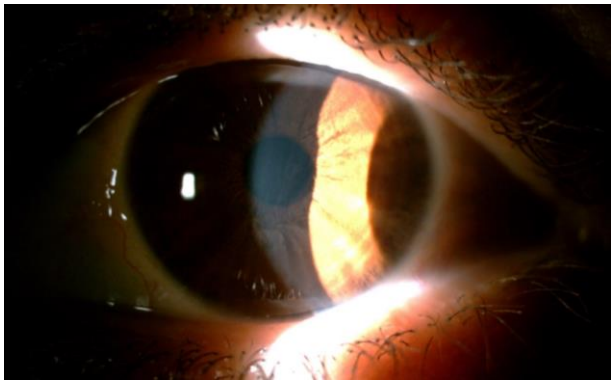

Fig 1: Cornea verticillata

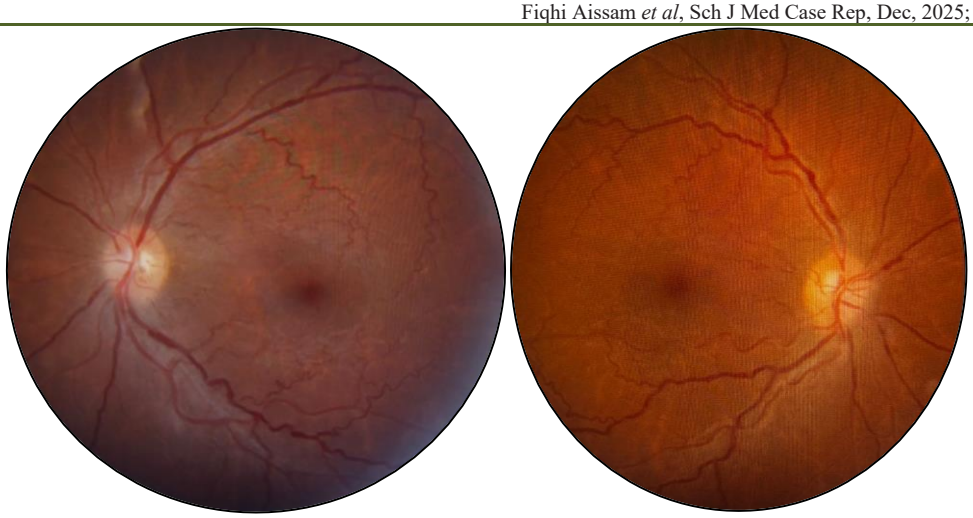

Fig 2: Fundus exam showing moderate vascular tortuosities

The patient underwent renal biopsy that showed enlarged, vacuolated podocytes and other glomerular cells containing lamellated inclusions on electron microscopy, which are considered pathognomonic for the Fabry disease. The genetic test was performed and confirmed the genetic disorder.

DISCUSSION

Ocular involvement is a well-documented and highly prevalent feature of Fabry disease, often providing early diagnosis. [4].

Cornea verticillata represents the most frequent ocular manifestation of the disease. Numerous studies have reported its presence in the vast majority of affected males and in some heterozygous females [8]. It appears early in the disease course and is asymptomatic, although some patients may report blurry vision. The detection of cornea verticillata during slit-lamp examination is highly suggestive of Fabry disease and further systemic evaluation is essential. [5].

Conjunctival and retinal vascular tortuosity are also common findings. The conjunctival vessels may appear dilated or telangiectatic, while the retinal vasculature often show increased tortuosity [6]. Several studies have suggested a possible correlation between the degree of vascular tortuosity and the severity of systemic disease or rate of progression. Advanced imaging techniques such as optical coherence tomography angiography (OCTA) have further enhanced our ability to detect subtle microvascular alterations, even in asymptomatic patients, highlighting the value of modern imaging in early disease monitoring. [7].

Lens changes (anterior capsular and posterior subcapsular opacities) are also described in affected individuals. These lens opacities, while not usually visually significant, contribute to the ophthalmic profile of Fabry disease and can aid in diagnosis.

Although less frequent, serious ocular complications such as retinal vascular occlusions (central retinal artery or vein occlusion), vasculopathic events, and optic neuropathies have been reported in isolated case studies. These manifestations are often associated with more advanced systemic involvement and may occur in conjunction with cerebrovascular events.

Large-scale prospective and registry studies have confirmed that ocular features are both common and characteristic in Fabry disease. They not only serve as important diagnostic clues but may also reflect the extent of systemic involvement. Consequently, comprehensive ophthalmologic evaluation plays a crucial role in the diagnosis, baseline assessment, and long-term follow-up of patients with Fabry disease.

CONCLUSION

Fabry disease is a rare hereditary disease. Ophthalmic manifestations are among the earliest and most consistent clinical indicators of FD. Early identification of these symptoms may alter the natural course of the disease and improve long-term outcomes.

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Informed consent was obtained from patients included in this study.

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