Keratoconus in a patient with Fabry disease: a multimodal imaging study

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PURPOSE

To report a case of bilateral keratoconus in a patient with Fabry disease. Reporting this unusual association can provide valuable insights into these two rare diseases.

CASE REPORT

A 43-year-old male was referred to the cornea Service of our eye Hospital to perform corneal topography due to corneal abnormalities detected after a routine ophthalmic evaluation. The visual acuity was poor in both eyes. The best corrected visual acuity was 20/100 and 20/200, with a refractive error of +0,50-5,50(60°) and +0,25-6,00(95°) in the right and left eye, respectively. At slit lamp examination, the patient presented corneal vortex opacities, increased vessel tortuosity of the conjunctiva and wedge-shaped lens opacites in both eyes (Fig.2). Intraocular pressure were within normal limits in bot eyes. Corneal topography showed a Stage II keratoconus according to the Amsler-Krumeich classification. Color fundus photography and autofluorescence imaging showed retinal vascular tortuosity in both eyes. Structural OCT examination revealed numerous hyperreflective foci (HRFs) within the inner retinal layers while OCTA showed FAZ enlargement in both superficial (SCP) and deep capillary plexus (DCP) (Fig.3). A clinical diagnosis of Fabry disease was suspected and confirmed by genetic test examination (**Galactosidase Alpha Gene: c.334C > T; p.Arg112Cys**) and by the detection of low a-Galactosidase A (GAL) enzyme serum activity. Three-year follow-up showed no progression of keratoconus in both eyes (Fig.1) reason why corneal crosslinking was not performed, and he was prescribed with rigid gas-permeable contact lenses.

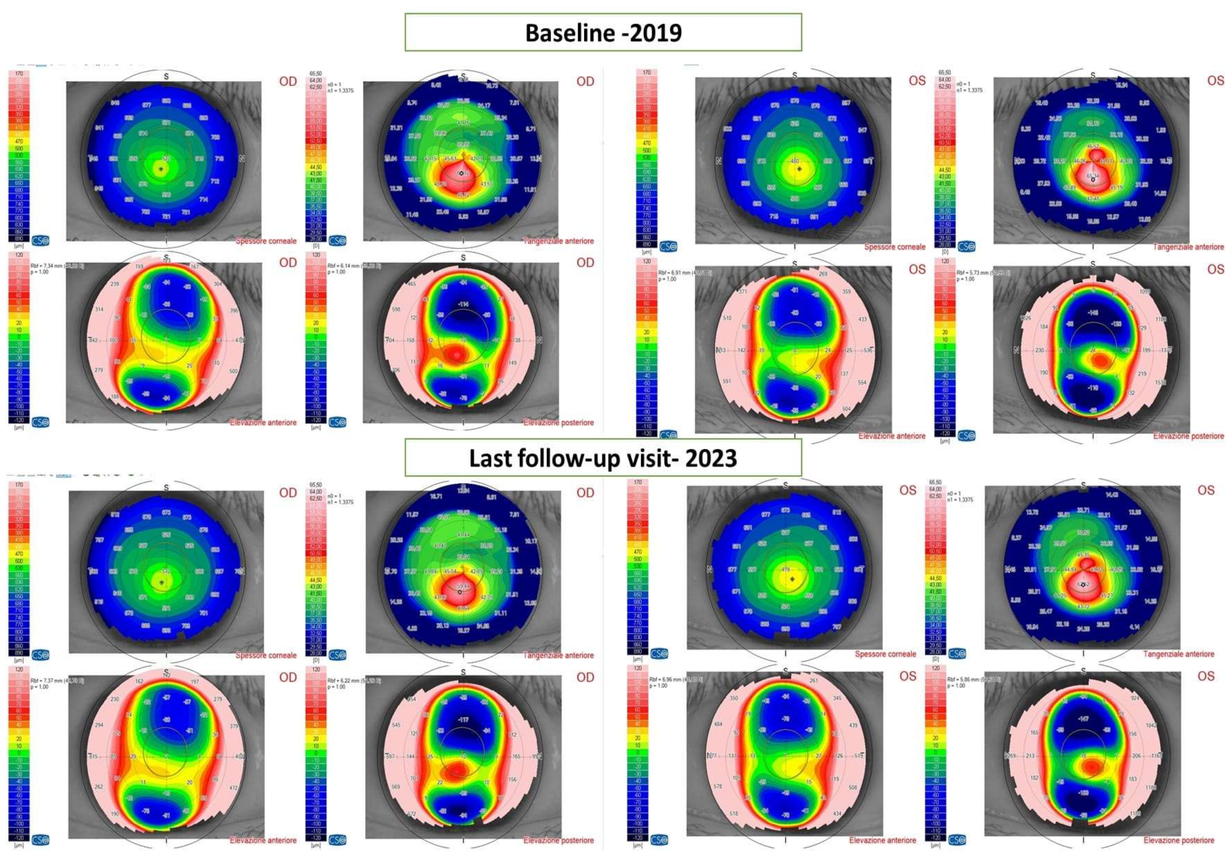

Figure 1: The corneal topography obtained using a combined rotating Scheimpflug camera and Placido-disk corneal topographer (Sirius, CSO, Italy), reveals an apex keratometric Reading of 60.12 diopters in the right eye and 65.34 in the left eye with an eccentric corneal steepening. The corneal thickness was 511 micron and 488 micron in the right and left eye, respectively. After 3-years follow-up the corneal topography showed no progression of keratoconus in both eyes

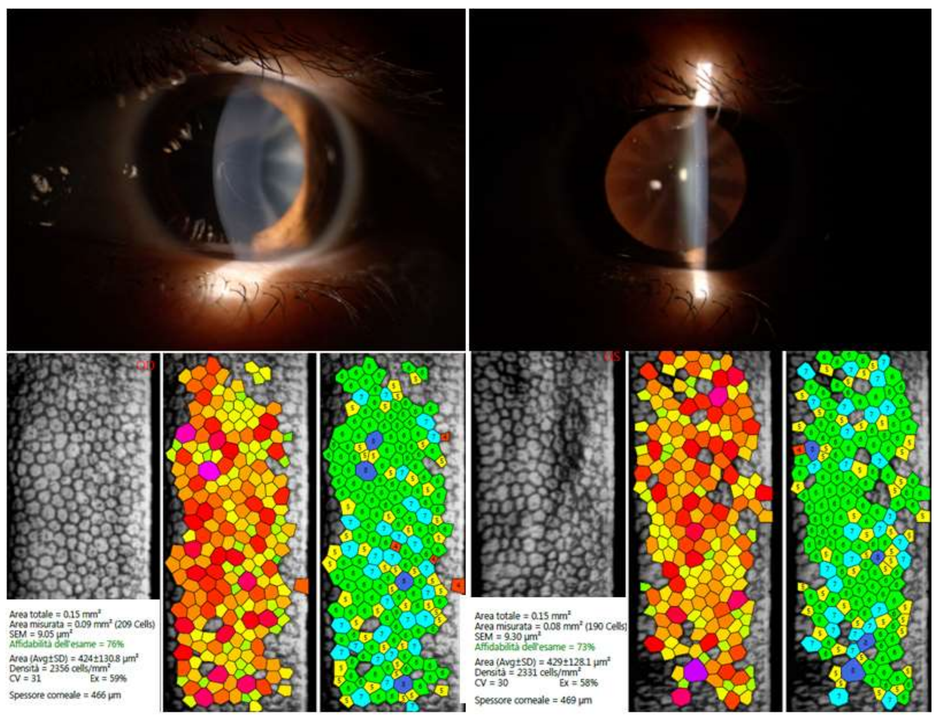

Figure 2: Slit lamp examination showing cornea verticillata and wedge-shaped lens opacites in both eyes. Corneal endothelial cells were within normal limits

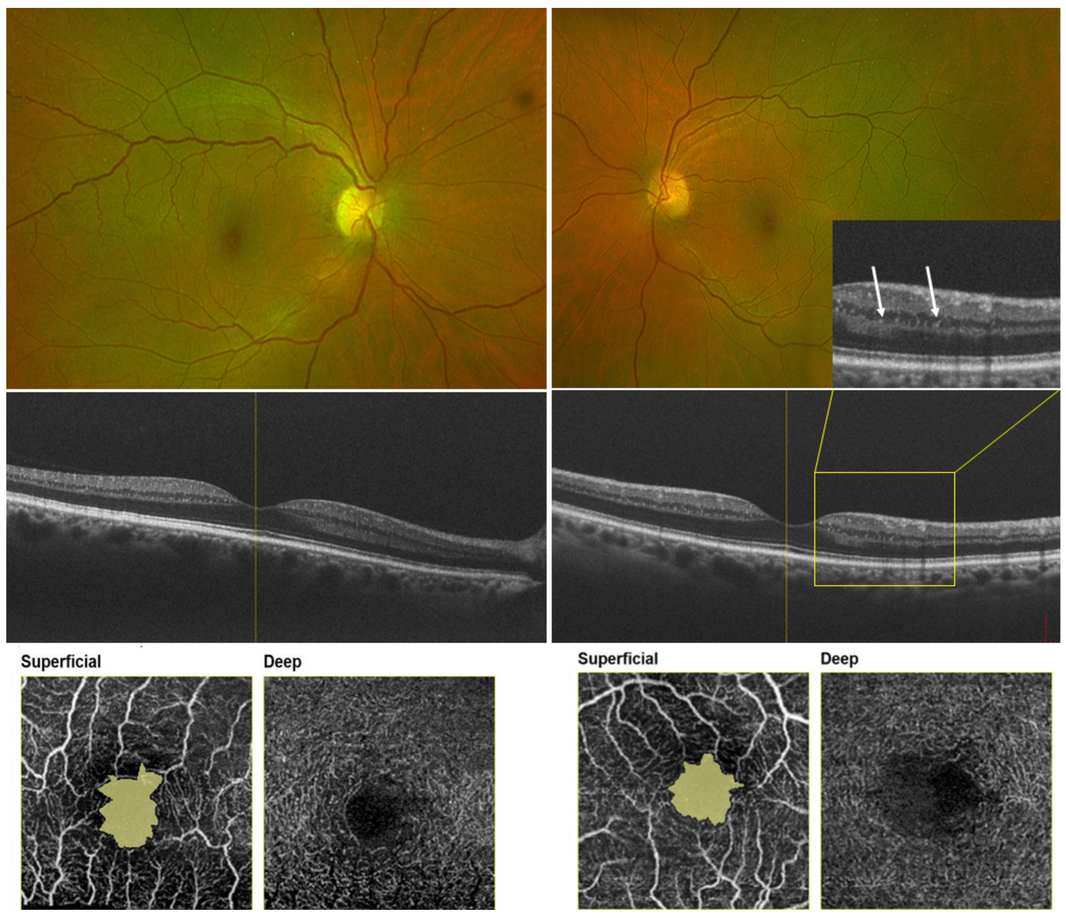

Figure 3: Ultra-wide field (UWF) fundus imaging (Optos, California) shows an increase in vascular tortuosity more evident in the right eye. Structural OCT examination (RS-3000 Advance SS-OCT, Nidek, Japan) showing Inner retinal hyperreflective foci. A magnification of the parafoveal region, highlights numerous HRFs mainly located in the inner nuclear layer, INL (white arrows). OCT-Angiography *(*RS-3000 Advance SS-OCT, Nidek, Japan) shows fovea avascular zone (FAZ) enlargement and vascular tortuosity in the superficial (SCP) and deep capillary plexus (DCP).

CONCLUSIONS

To the best of our knowledge, this is the first report to document keratoconus in a patient affected by Fabry disease. This report emphasizes the importance of screening, early diagnosis of this rare cause of decreased vision in patients affected by Fabry disease.

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