Unilateral Visual Loss as the Initial Presentation of Fabry Disease: A Diagnostic Challenge

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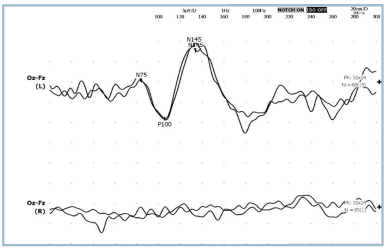
Introduction

Fabry disease is a rare X-linked lysosomal storage disorder caused by mutations in the GLA gene. It typically manifests with multi-organ involvement. Ocular signs such as cornea verticillata or retinal vessel tortuosity may occur early, but isolated acute visual loss is uncommon. We report a case of Fabry disease who presented solely with rapid-onset unilateral vision loss, initially misdiagnosed as optic neuritis.

Materials and Methods

A 32-year-old male presented with progressive visual decline in the right eye, accompanied by periocular and temporal pain. Within two days, the condition worsened to near-complete visual loss. Initial ophthalmologic evaluation showed increased retinal nerve fiber layer (RNFL) thickness on Optical Coherence Tomography, and orbital CT revealed diffuse thickening of the right optic nerve. A presumptive diagnosis of optic neuritis was made. He underwent diagnostic workup including brain and spinal MRI, that showed no abnormalities, visual evoked potentials (absent responses in the right eye), lumbar puncture, anti-AQP4 and anti-MOG antibody testing, autoimmune screening, and thrombophilia panel—all unremarkable.



VEP Destra / Sinistra 32x24 - Stim. 30 primi						
	N75	P100	N145	Amp N75	Amp P100	N75-N145
1.1 Oz-Fz Sinistra	83,5	105,6	133,9	9,90	17,07	50,40
1.2 Oz-Fz R. Sinistra	84,0	105,6	132,5	9,63	18,42	48,53
2.1 Oz-Fz Destra	--	--	--	--	--	--
2.2 Oz-Fz L. Destra	--	--	--	--	--	--

Confronto - Sinistra / Destra	
1.Sinistra / Destra	2.Sinistra / Destra
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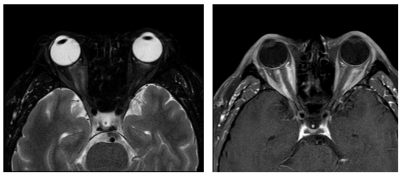
Results

At follow-up, a more detailed ophthalmologic examination revealed corneal and retinal abnormalities suggestive of Fabry disease, confirmed by genetic testing (amino acid substitution in exon 7 GLA c. 1081 G>A gene). No other systemic involvement was found upon cardiologic, neurologic, and nephrologic evaluation. The decrease in visual acuity was interpreted as a consequence of ischemic optic neuropathy and the patient was started on enzyme replacement therapy with agalsidase beta.



Discussion

Fabry disease has a highly variable clinical presentation, often leading to delayed diagnosis. In this case, the isolated and acute visual symptoms led to an initial misdiagnosis of optic neuritis. However, the lack of confirmatory findings from MRI and cerebral spinal fluid studies, combined with absent visual evoked potentials, prompted reconsideration of the differential diagnosis. Ocular manifestations of Fabry disease—though frequently underrecognized—can provide key diagnostic clues. This case underscores the importance of considering Fabry disease even in the absence of systemic signs. Early diagnosis allows timely initiation of specific therapy and may delay or prevent progression to irreversible organ damage.

Bibliography

1) Michaud M et al. When and How to Diagnose Fabry Disease in Clinical Practice. Am J Med Sci. 2020.
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