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X-Linked Retinitis Pigmentosa Revealed by Tapetal Like Reflex: A Case Report

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

The tapetal-like reflex (TLR) is a rare, golden, bright, scintillating, particulate reflection observed during indirect ophthalmoscopy, typically sparing the fovea. It resembles the reflection seen in the eyes of certain vertebrates. TLR has been noted in female carriers of X-linked retinitis pigmentosa (RP) and has also been observed in a healthy young male.

Keywords: X-linked Retinitis pigmentosa; Retinitis pigmentosa; retinal degeneration; retinal reflex.

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1. INTRODUCTION

"Retinitis pigmentosa is a group of inherited retinal diseases characterized by degeneration of rod and cone photoreceptors (Meindl et al., 1996). Human inherited retinal degenerations are genetically heterogeneous, with well over 100 genes implicated to date" (Roepman et al., 1996). "Measurements of retinal function, such as the electroretinogram, show that photoreceptor function is usually compromised many years before symptomatic night blindness, visual field scotomas, or decreased visual acuity occur. More than 45 genes have been identified for retinitis pigmentosa" (Murga-Zamalloa et al., 2010; Khanna, 2015).

"X-linked Retinitis Pigmentosa GT Pase Regulator is a GTPase-binding protein encoded in humans by the RPGR gene, which is located on the X chromosome and is commonly associated with X-linked retinitis pigmentosa (XLRP)" (Churchill et al., 2013). "In photoreceptor cells, RPGR is localized in the connecting cilium, which connects the protein-synthesizing inner segment to the photosensitive outer segment and is involved in modulating cargo trafficking between the two segments" (Glomset & Farnsworth, 1994).

2. CASE PRESENTATION

We report the case of a 45-year-old woman presented with complaints of decreased vision in the left eye (LE). Her best-corrected visual acuity in the RE was 10/10 and left eye was 06/10. Both eyes (BEs) anterior segment examination was unremarkable. There was no history of night blindness or decreased vision in any of the family members. Fundus examination revealed presence of retinal pigment epithelial (RPE) hypopigmentation and atrophy at the posterior pole along with an enhanced golden tapetal sheen seen in posterior pole of the left eye (Fig. 1A). Fundus autofluorescence revealed a crescent-shaped hyperautofluorescence in LE (Fig. 1B). Spectral domain optical coherence tomography (SD-OCT) revealed a normal foveal contour in the left eye with thining of photoreceptor layer to the macula in LE with small drusens (Fig. 2). Based on the multimodal imaging findings, we suspected a starting manifestations of retinitis pigmentosa and the patient was advised for genetic analysis revealing mutation on x-linked RPGR/RP3 GENE then set for observation and a routine follow-up.

A **B**

Fig. 1. A: fundus photo showing tapetal like reflex in left eye, B: fundus autofluorescence showing hyperreflectivity of the posterior pole by atrophy starting of the retinal pigmented epithelium

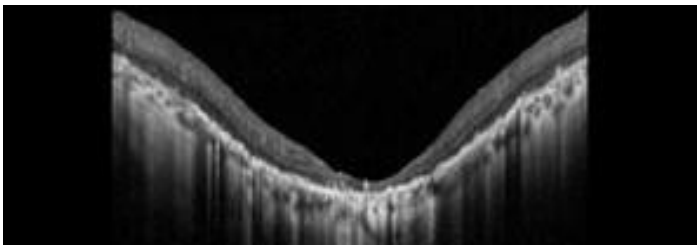

Fig. 2. Macular oct of left eye showing extreme thining of photoreceptor layer with small drusens

3. DISCUSSION

“TLR has been described in female carriers of X-linked RP and has also been observed in a healthy young male” (Meindl et al., 1996). In addition, abnormal fundus reflections in male patients have been reported in Oguchi disease, X-linked retinoschisis, cone-shaped retinal dystrophy, and early X-linked RP. “The TLR has been reported to be located deep to the retinal vasculature and at the level of the outer retina and RPE.3 It may be due to deposits, thickening or degeneration of Bruch's membrane, retinal deposits or an alteration at the level of the RPE-photoreceptor interface” (Meindl et al., 1996). We report the presence of an enhanced TLR located in the midperiphery in a patient with sector RP, confirming its association with inherited retinal degenerations; however, the nature and origin of the phenomenon are still not clear and further studies will shed light on this unique finding.

The worldwide prevalence of RP is 1:3000 to 1:7000 including simple, syndromic and systemic disease. There is usually no gender predilection, but males are slightly more affected than females due to the existence of X-linked RP as seen in our patient. RP has no ethnic specificity, but in certain forms related to specific gene mutations it is more common in conanginous populations (such as the USH3 gene associated with Usher syndrome type III) (Ferrari et al., 2006).

Most cases of retinitis pigmentosa are monogenic, but the disease is still very genetically heterogeneous (Hartong, Berson, & Dryja, 2006). Researchers have identified at least 45 loci at which mutations cause the disease, and these genes together account for the disease in just over half of all patients (Hardcastle et al., 2000).

“Since the first report describing linkage of an RP locus to a DNA marker on human chromosome X in 1984 (Gorbatuyk et al., 2010), more than 40 genes have been associated with RP. Non-syndromic or "simple" cases may be inherited as autosomal dominant (20-25%), autosomal recessive (15-20%), X-linked recessive (10-15%), or sporadic/simple (30%), and 5% may be early-onset and grouped as part of Leber congenital amaurosis (LCA). Rarer forms also exist, such as X-linked dominant, mitochondrial, and digenic (due to mutations in two different genes)” (Ferrari et al., 2006).

XLRP is due to mutation of 6 genes located in chromosome X, but only two have been identified so far: the retinitis pigmentosa gtpase regulator RPGR and the Rp-2 protein RP2 (Vettel & Wittinghoffer, 2009). The mutations in this 2 genes are reposabale for 80/100 of clinical cases of XLRP which make them a good target to small molecules drug and gene therapy approaches (Vervoort et al., 2000; Kalitzeos et al., 2019; Schatz et al., 2012).

4. CONCLUSION

Tapetal like reflex is frequently an incidental fundiscopic finding associated with many retinal diseases like XLRP and may appear years before symptoms and vision loss.

Well understanding of the phenotypes and genes included in RP is the key to be more efficient in developing new treatments of the disease including gene specific approaches, transplantation replacing retinal loss tissue and implanting electrical devices.

DISCLAIMER (ARTIFICIAL INTELLIGENCE)

Author(s) hereby declare that NO generative AI technologies such as Large Language Models (ChatGPT, COPILOT, etc) and text-to-image generators have been used during writing or editing of this manuscript.

CONSENT

As per international standards or university standards, Patient’s written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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