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Tapetal-like reflex in X-linked RPGR-associated retinopathy

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Abstract

Purpose: To investigate the clinical, imaging, electrophysiological, and genetic characteristics of male patients with *RPGR*-associated retinopathy exhibiting tapetal-like reflex (TLR) versus those without (non-TLR).

Methods: This retrospective observational study included 9 Indian males from 7 unrelated families with genetically confirmed pathogenic *RPGR* variants. Patients were divided into TLR (*n*=6) and non-TLR (*n*=3) groups based on fundus appearance. Multimodal imaging (fundus photography, fundus autofluorescence [FAF], optical coherence tomography [OCT]) and full-field electroretinography (ERG) were analyzed. Molecular genetic testing was performed to identify *RPGR* variants.

Results: The TLR group showed a characteristic golden, scintillating sheen with radial streaks on fundus exam. Median age of onset was 35 years with worse best-corrected visual acuity (BCVA) (0.66 LogMAR) and higher myopia (median -5.50 D) compared to the non-TLR group (23 years, 0.26 LogMAR, -1.00 D). FAF in the TLR group revealed central confluent or patchy macular atrophy surrounded by hyper-autofluorescence, whereas the non-TLR group exhibited widespread mid-peripheral degeneration and bull's eye maculopathy. OCT showed complete outer retinal atrophy (cRORA) in most TLR eyes and incomplete atrophy (iRORA) with preserved ellipsoid zone in the youngest patient. Non-TLR eyes demonstrated milder retinal atrophy with limited ellipsoid zone preservation. Full-field ERG demonstrated preserved scotopic but extinguished photopic responses in TLR eyes, while non-TLR eyes had extinguished photopic and severely reduced scotopic responses. All variants were hemizygous RPGR mutations in exon 15, predominantly frameshift or stop-gain mutations.

Conclusions: Tapetal-like reflex in male *RPGR*-associated retinopathy correlates with a cone-rod dystrophy-like phenotype featuring later onset, severe central atrophy, and predominant photopic dysfunction. In contrast, absence of TLR associates with rod-cone dystrophy-like

features. Recognition of TLR may aid clinical classification, early diagnosis, and prognosis in RPGR-related retinal disease.

Keywords: RPGR- associated retinopathy; Tapetal- like reflex; macular atrophy; male.