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LONGITUDINAL STUDY OF DISEASE COURSE IN PATIENTS WITH X-LINKED RETINITIS PIGMENTOSA DUE TO RPGR GENE MUTATIONS

Oral

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Purpose:

To investigate the disease course of X-linked Retinitis Pigmentosa (RP) caused by mutations in the RPGR gene.

Methods:

In this retrospective, single-center, longitudinal observational study, we evaluated a cohort of 48 male patients (Mean age: 29.6 ± 15.2 years old) from 31 unrelated families with a diagnosis RPGR-associated RP. At each visit, all patients underwent best-corrected visual acuity (BCVA) assessment and were examined with Goldmann visual field (GVF), optical coherence tomography (OCT), fundus autofluorescence (FAF), microperimetry and full-field electroretinography (ERG).

Results:

We analyzed 48 patients throughout a mean follow-up of 6.5 (±0.7) years. Mean BCVA was 0.6 (±0.7) logMAR, mostly with myopic refraction (79.2%). 30 patients (62.5%) showed pigmentary changes, whereas 18 (37.5%) sine pigmento RP. BCVA declined at a mean rate of 0.025 (±0.012) logMAR/year, primarily driven by GVF loss. 13 patients (27.1%) had macular abnormalities. ERG demonstrated rod-cone dysfunction in half of the cohort. Perimacular hyperautofluorescent ring was found in 24 patients (50%), significantly younger and with higher BCVA compared to those with decreased FAF. Pathogenic variants in exons 1-14 resulted in milder phenotypes compared to ORF15 mutation.

Conclusions:

Males with RPGR gene-associated RP displayed a composite spectrum of disease progression. Milder phenotypes were observed in patients with sine pigmento RP, absence of high myopia and mutations in exons 1-14 of the RPGR gene.