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**EVOLIUCINĖ MEDICINA:  
ŽMOGAUS RAIDOS PERSPEKTYVOS IR SVEIKATA  
BESIKEIČIANČIOJE APLINKOJE**

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**EVOLUTIONARY MEDICINE:  
PERSPECTIVES ON HUMAN DEVELOPMENT AND HEALTH  
CONSEQUENCES IN A CHANGING ENVIRONMENT**

Rengėjas:

Vilniaus universiteto Medicinos ir sveikatos mokslų  
Doktorantūros mokykla, VU Medicinos fakultetas

Organized by

the Doctoral School of Medicine and Health sciences  
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# COMPARATIVE ANALYSIS OF MACULAR STRUCTURAL PARAMETERS AMONG GENETIC SUBTYPES OF ROD-CONE DYSTROPHIES

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**Background and Aim:** Macular structure is closely associated with best corrected visual acuity (BCVA) in patients with rod-cone dystrophies (RCDs). This study aimed to investigate whether macular structural alterations in RCDs differ by genotype.

**Materials and Methods:** Individuals with suspected RCDs underwent comprehensive ophthalmic and genetic examination at a tertiary referral center between 2013 and 2026. BCVA and macular optical coherence tomography parameters were compared across different genotypic groups. The analysed parameters included central retinal thickness (CRT), subfoveal choroidal thickness (SCT), ellipsoid zone length (EZL), outer nuclear layer thickness (ONLT), as well as the presence of hyperreflective foci (HRF), intraretinal fluid (IRF) and epiretinal membrane (ERM). Due to non-normal distribution of the data, group comparisons were performed using the Kruskal-Wallis test in R software. Linear regression models adjusted for age were used for continuous variables. Categorical variables were compared using the chi-square test.

**Results:** 91 probands with a confirmed molecular diagnosis were included. The most frequently affected genes were *USH2A* (n = 19), *RP1* (n = 17), and *EYS* (n = 14), while 41 individuals carried disease-causing variants in other genes. The median BCVA in the *RP1* group was significantly higher compared with the other groups (p < 0.05). In addition, the *RP1* group demonstrated a significantly greater EZL (p < 0.05). The *USH2A* and *RP1* groups showed a significantly higher prevalence of ERM (p < 0.05). No significant differences were observed in CRT, SCT, ONLT, or in the presence of HRF and IRF among the groups.

**Conclusions:** Individuals with *RP1* variants demonstrated a milder disease course, characterised by better-preserved EZ in the macula and higher BCVA. Both the *RP1* and *USH2A* groups showed a higher risk of developing ERM, which may warrant surgical intervention.

**Keywords:** macular structure, ophthalmic genetics, rod-cone dystrophies