Research Reports

Investigating the role of BEST1 and PRPH2 variants in the molecular aetiology of adult-onset vitelliform macular dystrophies

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ABSTRACT

Objective: To determine the clinical relevance and frequency of BEST1 and PRPH2 mutations in a clinically diagnosed adult-onset vitelliform macular dystrophy (AVMD) group with Caucasian ethnicity.

Methods: The study comprised 24 patients who had been diagnosed with AVMD via indirect fundus ophthalmoscopy and presented with a dome-shaped appearance between the retinal pigment epithelium and photoreceptors on their spectral-domain optical coherence tomography. They had lesion hyper- autofluorescence on their fundus autofluorescence images and were also investigated for BEST1 and PRPH2 mutations for a probable molecular aetiology.

Results: No pathogenic or likely pathogenic mutation was detected in the BEST1 and PRPH2 genes of any of the clinically diagnosed AVDM patients. A heterozygous NM_000322.5:c.938C>T (p.Pro313Leu) variant of the PRPH2 gene was detected in 2 non-consanguineous patients. According to current guidelines, this variant was classified as a 'variant of uncertain significance'.

Conclusion: In conclusion, AVMD is a genotypic and phenotypic heterogeneous disease. The genetic aetiology could not be explained by sequencing BEST1 and

PRPH2 genes in the AVMD patients; however, the variant of PRPH2 could be a cause of predisposition relevant to the phenotype.

KEYWORDS: Adult vitelliform dystrophy, BEST1, gene, PRPH2