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Analysis of *ELOVL4* and *PRPH2* genes in Turkish Stargardt disease patients

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ABSTRACT. Stargardt disease (STGD) is an inherited genetic eye condition involving bilateral macular dystrophy leading to progressive central vision loss. It is the most common form of autosomal recessive juvenile macular dystrophy. In this study, *ELOVL4* and *PRPH2* genes were analyzed in 30 STGD probands for genetic variations using next-generation sequencing. In the patient group, two genetic variants in exon 6 of *ELOVL4*, and three in exon 3 of *PRPH2* were detected. All sequence modifications in both *ELOVL4* and *PRPH2* were recorded, including

those of a non-pathogenic nature. In the control group, four different genetic variations were detected in *ELOVL4*, and five in *PRPH2*. STGD patients of different ethnicities may carry distinct *ELOVL4* and *PRPH2* sequence variants. We believe that the genetic variations identified in this study may be related to STGD etiopathogenesis.

Key words: *ELOVL4* gene; *PRPH2* gene; Stargardt disease