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Targeted next generation sequencing identifies novel mutations in Indian patients with retinal dystrophies

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Abstract

Retinal dystrophies (RD) are a group of inherited ocular disorders of the retina causing blindness in more than 2 million people worldwide. RDs are characterized by clinical variability and progressive vision loss. It is associated with high degree of genetic heterogeneity. In order to correlate RDs clinically and genetically and to develop novel therapeutic approaches, genetic testing is of utmost importance. Prior requisite of a genetic test is genetic counselling. The proband and family members underwent genetic counselling including a detailed family history. Pre-test education was vital to help these families understand the importance of genetic test for the proband and validation of the report by testing the parents/siblings blood samples to confirm the genetic mutation. We performed targeted next-generation sequencing (NGS) in clinically confirmed 21 unrelated patients who showed different forms of RD and validated in their family members using panel comprising 184 genes, which covered previously associated genes with retinal disease. The sequencing analysis revealed a total of 21 different mutations in patients with RDs including Leber's Congenital Amaurosis, Cone-Rod dystrophy, Retinitis Pigmentosa, Achromatopsia and Stargardt disease. Among these, seven mutations were unreported and fourteen variants were reported. We found five novel mutations with existing spectrum of gene mutations identified in Indian patients with the characteristic features of RDs. The knowledge of the pathogenic gene mutation in the affected family member was used to correlate with the proband's clinical diagnosis, to screen other family members suspected of having similar symptoms and also for carrier testing. In some cases of retinal dystrophy with overlapping clinical symptoms, the genetic report was used to confirm the RD. Post-test genetic counselling was done to discuss the implications of the genetic mutation on the prognosis and management of the RD.

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