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Identification of causal variants in North Indian families with ocular disorders

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Understanding the molecular genetics of hereditary eye disorders is essential not only for early diagnosis but also to arrest the progression of the disease. North Indian population encompassing Eastern Uttar Pradesh and Western Bihar has been untouched in terms of understanding the genetics of eye disorders. We have recruited several families afflicted with various ocular diseases like X-linked idiopathic congenital nystagmus (XLICN), X-linked retinoschisis (XLR), macular dystrophy etc. We have identified a novel missense mutation c.556A>G (p.M186V) in the *FRMD7*. A recurrent missense mutation c.242T>A (p.I81N) in the gene Retinoschisin (RS1) was also identified in XLR. The c.242T>A mutation leads to an Ile81Asn substitution in the Discoidin domain of the RS1protein which is likely to interfere with the function of the RS1 protein. Detailed *in silico* analysis also supports the pathogenecity of these variants. We have also carried out WES (whole exome sequencing) in families with macular dystrophy and identified causal mutations. Collectively, this study will help us in identification of rare and novel genetic variants in our population that could be used as diagnostic genetic markers. This will also improve our understanding towards molecular mechanism and genetic basis of various eye disorders. Variants identified in this manner will enable us predict the predisposition towards a genetic ocular disorder and will facilitate early diagnosis and better management of such diseases.

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