A novel missense variant in IDH3A causes autosomal recessive retinitis pigmentosa

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Inherited retinal degenerations (IRDs) encompass a wide spectrum of genetic ocular diseases characterized by considerable genetic and clinical heterogeneity.; Complete ophthalmic examination and next-generation sequencing.; We describe a patient with no family history of vision loss, who at the age of 28 years developed visual impairment consistent with a severe form of retinitis pigmentosa. Genetic testing by means of whole exome sequencing identified a homozygous variant in the gene IDH3A. To date, only three papers have reported mutations in IDH3A, in families with early-onset retinal degeneration with or without the presence of macular pseudocoloboma.; This study highlights the importance of including this rarely-mutated gene in the molecular diagnostic set-ups for IRDs, and further delineates the phenotypic spectrum elicited by mutations in IDH3A.

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