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Mutations in *IMPG1* cause autosomal dominant and recessive retinitis pigmentosa

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Abstract

Purpose : To identify the causative mutation in a large family with autosomal dominant retinitis pigmentosa (adRP) and consequently to genetically characterize a cohort of RP patients without mutation in known genes.

Methods : A large adRP family without mutation in the known RP genes was screened for mutations using whole-exome sequenging (WES). Following the identification of an *IMPG1* mutation, additional dominant and recessive RP probands were screened for mutation in this gene by targeted next-generation sequencing (tNGS). Clinical investigations included visual acuity and visual field testing, fundus examination, high-resolution spectral-domain optical coherence tomography (OCT), fundus autofluorescence imaging, full-fields and multifocal electroretinogram (ERG) recording.

Results : By WES, a heterozygous splicing mutation in the *IMPG1* gene, c.1824+1G>A was identified in a large family with adRP. Three more families with different novel missense mutations were identified. All the variants segregated with the disease phenotype and are predicted to be pathogenic.

Conclusions : We identified a novel causative gene, *IMPG1*, responsible for autosomal dominant and recessive retinitis pigmentosa (RP). *IMPG1* was previously associated with vitelliform macular dystrophy (VMD). In conclusion, mutations in the same gene can lead to two clinical entities, RP and VMD as it was reported previously for *IMPG2*, paralog gene of *IMPG1*.

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