Appendix Table A2. The prediction of the variant identified by exome sequencing and confirmed by Sanger sequencing according to ACMG guidelines.

Gene	Position	RefSeq ID	Nucleotide variant	Amino acid change	Region	dbSNP 137	1000 Genomes _all	ExAC_EAS database	ESP6500_ all	PolyPhen -2	SIFT	Mutation Taster	Disease	Genotype IV:1	Genotype V:1	Previous study
CNGB1	Chr 16: 57996772	NM_001297.4	c.385delC	p.(L129WfsTer148)	Exon 6	Novel	Novel	Novel	Novel	N/A	N/A	Disease causing (score: 1)	RP45	MUT/MUT	WT/MUT	No

RP45 = retinitis pigmentosa 45, N/A = not available, WT = wild-type, MUT = mutation.