Supplementary Table 1. Protein-altering biallelic variants shared by two patients in Family 1.

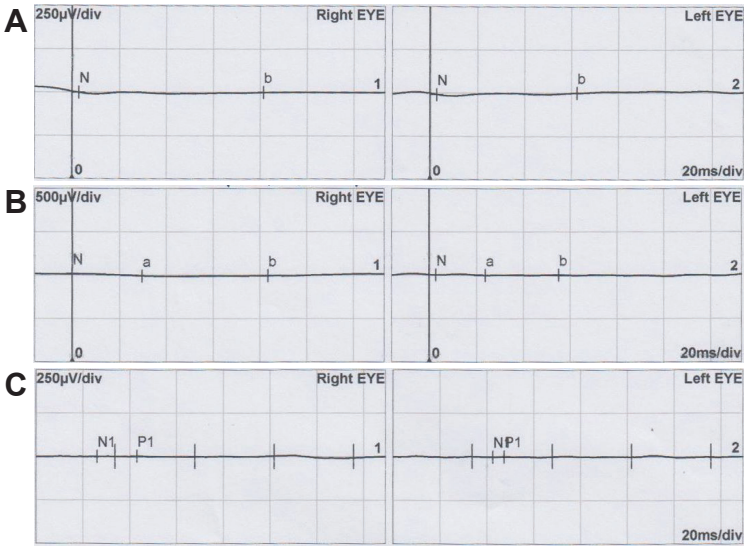
Chromosome Coordinate (hg19)	Change	Genotype	Gene	Variant type	Reason for exclusion
Chr9: 80868198	G > A	Homo	*CEP78*	Sp	/
Chr10: 32860815	A > T	Homo	*CCDC7*	Ns	LoF
Chr1: 175129955	G > T	Homo	*KIAA0040*	Ns	Con
Chr1: 900519	A > G	Homo	*KLHL17*	Ns	Con
Chr1: 228430962	G > C	Homo	*OBSCN*	Ns	LoF
Chr1: 247835881	C > T	Homo	*OR13G1*	Ns	LoF
Chr16: 4935049	G > A	Homo	*PPL*	Ns	LoF
Chr4: 122301586	T > G	Homo	*QRFPR*	Ns	Con
Chr6: 111696550	T > C	Homo	*REV3L*	Ns	Con

Homo, homozygous; Sp, splicing; Ns, nonsynonymous; Con, the exact variant amino acid change occurs in other vertebrates, thus considered not conserved; LoF, the associated gene contains homozygous loss-of-function alleles in ExAC database, thus considered dispensable.

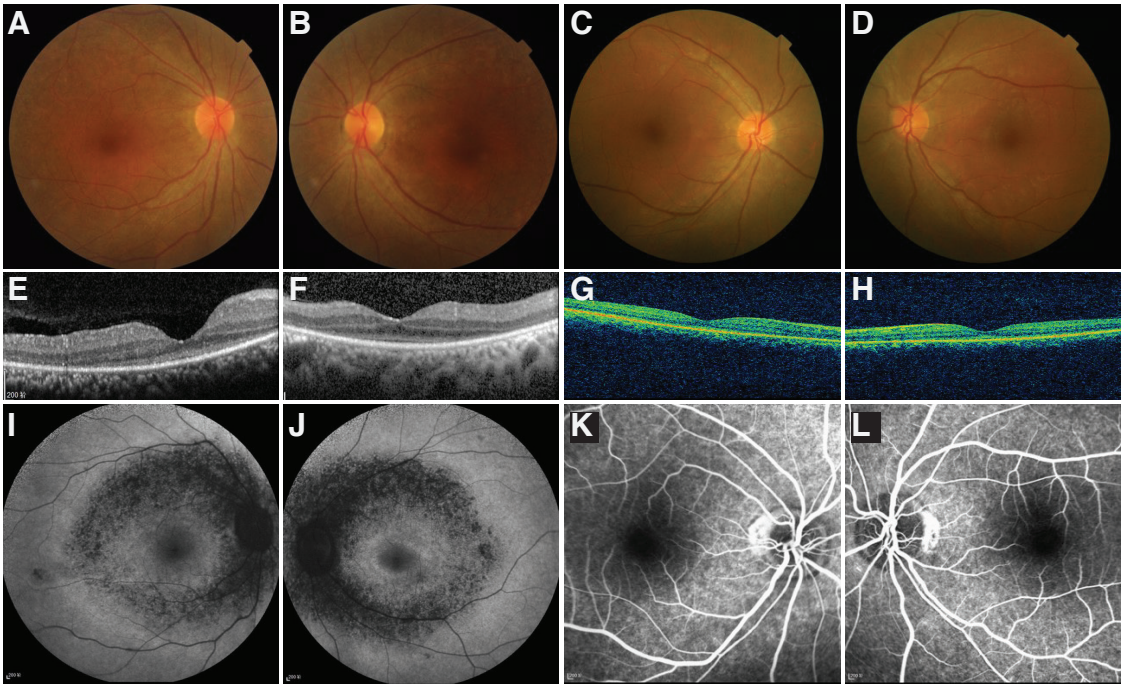
Supplementary Table 2. Biallelic variants identified in Family 2: V-1.

Chromosome Coordinate (hg19)	Change	Genotype	Gene	Variant type
Chr9: 80880286	A > G	Homo	*CEP78*	Sp
Chr16: 88599697	A > C	Homo	*ZFPM1*	Ns
Chr7: 100647511	C > T	Homo	*MUC12*	Ns
Chr16: 70211370	C > G	Homo	*CLEC18C*	Ns
Chr3: 16926607	T > C	Homo	*PLCL2*	Ns
Chr3: 75788260	C > A	Homo	*ZNF717*	Ns
Chr14: 88431969	T > C	Homo	*GALC*	Ns
Chr16: 1279060	C > G	Homo	*TPSB2*	Ns
Chr12: 81066976	A > T	Homo	*PTPRQ*	Ns
Chr3: 184280070	T > G	Homo	*EPHB3*	Ns
Chr11: 123886583	A > G	Homo	*OR10G4*	Ns

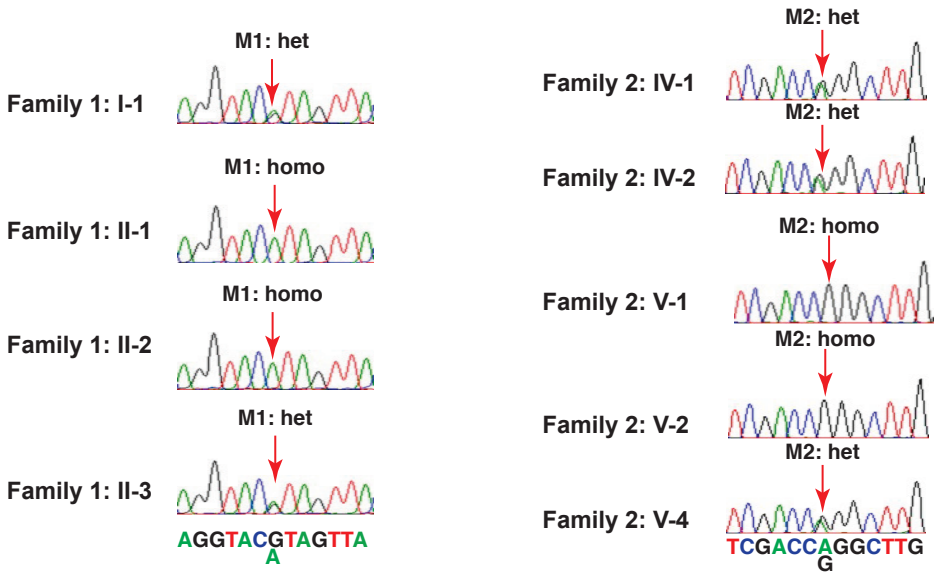
Homo, homozygous; Sp, splicing; Ns, nonsynonymous;



Supplementary Figure 1. Electroretinogram (ERG) data of Family 2: V-1
The results of scotopic ERG 0.01 (A), photopic ERG 3.0 (B), flicker ERG 30Hz (C), funduscopy (D) and fluorescein angiography (E).



Supplementary Figure 2. Ophthalmological findings of the Family 1:II-1 and Family 2: V-2
Fundus images of Family 1: II-1, OD (A) and OS (B), Family 2: V-2, OD (C) and OS (D). OCT images of Family 1: II-1, OD (E) and OS (F), Family 2: V-2, OD (G) and OS (H). Fundus autofluorescence results of Family 1: II-1, OD (I) and OS (J). Fundus fluorescein angiography of Family 2: V-2, OD (K) and OS (L).



CEP78: NM_032171, M1: c.1254+5G>A (p.R403Sfs*7), M2: c.1629-2A>G (p.G545Pfs*6)

Supplementary Figure 3. Sanger sequencing chromatograph and co-segregation analysis. Het, heterozygous; homo, homozygous.