Supplementary table 2 Genetic profile of patients with inherited retinal diseases and associated families

No.	Family	Relationship	Symptom	Ocular history	Genotype	Inheritance	Gene	Nucleotide change	Amino acid change	Variant effect
1	1	Mother	Unaffected	Myopia	Heterozygous	AD	*PROM1*	c.139del	p.His47IlefsTer12	Frameshift
2	2	Father	Unaffected	Nil	Heterozygous	AD	*PROM1*	c.139del	p.His47IlefsTer12	Frameshift
		Mother	Unaffected	Nil	Heterozygous	AD	*PROM1*	c.139del	p.His47IlefsTer12	Frameshift
5	5	Father	Unaffected	Nil	N/A	N/A	N/A	N/A	N/A	N/A
		Mother	Unaffected	Nil	Heterozygous	AD	*PROM1*	c.139del	p.His47IlefsTer12	Frameshift
					Hemizygous	X-linked recessive	*CACNA1F*	c.3115G>T	p.Glu1039Ter	Stop gained
		Younger brother	Unaffected	Myopia Amblyopia	Hemizygous	X-linked recessive	*CACNA1F*	c.3115G>T	p.Glu1039Ter	Stop gained
6	6	Older son	Unaffected	Myopia	Heterozygous	AD	*PROM1*	c.794del	p.Glu265GlyfsTer9	Frameshift
		Younger son	Unaffected	Myopia	Heterozygous	AD	*PROM1*	c.794del	p.Glu265GlyfsTer9	Frameshift

Abbreviation: AD, autosomal dominant; AR, autosomal recessive.