Appendix 1. Summary of clinical findings and probands *BEST1* mutations.

PATIENT	MUTATION	POSITION	MISSENSE EFFECT	AGE - GENDER	AGE OF ONSET	LESION TYPE RE	LESION TYPE LE	BCVA RE	BCVA LE	COMPLICATIONS
FG01 (FAMILY FG I)	C>T728 heterozygous	exon 7	A243V	49-M	41-	atrophy	atrophy	20/125	20/160	-
FG02 (FAMILY FG I)	C>T728 heterozygous	exon 7	A243V	45-F	37-	vitelliruptive	vitelliruptive	20/25	20/25	-
FG03 (FAMILY FG I)	C>T728 heterozygous	exon 7	A243V	75-M	67-	pseudohypopion	vitelliruptive	20/50	20/125	-
FG04 (FAMILY FG I)	C>T728 heterozygous	exon 7	A243V	13-F	-	none	none	20/20	20/20	-
FG05 (FAMILY FG I)	C>T728 heterozygous	exon 7	A243V	17-F	-	none	none	20/20	20/20	-
FG06 (FAMILY FG II)	G>A275 heterozygous	exon 4	R92G	16-F	11-	fibrosis	fibrosis	20/160	20/160	CNV RLE
FG07 (FAMILY FG II)	G>A275 heterozygous	exon 4	R92G	3-M	2-	vitelliform	vitelliform	20/32	20/32	-
FG08 (FAMILY FG III)	C>T 274 homozygous	exon 4	R92C	16-F	15-	vitelliruptive+multifocal	vitelliruptive+multifocal	20/32	20/40	-
FG09 (FAMILY FG IV)	G>A44 heterozygous	exon 2	G15D	3-F	2-	vitelliform	vitelliform	20/25	20/25	-
FG10 (FAMILY FG IV)	G>A44 heterozygous	exon 2	G15D	30-M	-	none	none	20/20	20/20	-
CT01 (FAMILY CT I)	C>T274 heterozygous	exon 4	R92C	14-M	8-	fibrosis	fibrosis	20/50	20/40	CNV RLE
CT02 (FAMILY CT II)	T>C791 heterozygous	exon 7	I230T	11-M	10-	pre-vitelliform	pre-vitelliform	20/20	20/25	-
CT03 (FAMILY CT II)	T>C791 heterozygous	exon 7	I230T	42-F	41-	pre-vitelliform+multifocal	pre-vitelliform+multifocal	20/32	20/25	-
CT04 (FAMILY CT II)	T>C791 heterozygous	exon 7	I230T	9-M	6-	vitelliruptive	vitelliruptive	20/125	20/125	-
CT05 (FAMILY CT III)	C>T272 heterozygous	exon 4	T91I	44-M	36-	atrophy	atrophy	20/125	20/40	-
CT06 (FAMILY CT III)	C>T272 heterozygous	exon 4	T91I	19-F	11-	fibrosis	fibrosis	20/200	20/40	CNV RE

CT07 (FAMILY CT IV)	A>G10 heterozygous	exon 2	T4A	27-F	20-	atrophy	none	20/50	20/25	-
CT08 (FAMILY CT IV)	A>G10 heterozygous	exon 2	T4A	23-F	16-	pseudohypopion	atrophy	20/32	20/50	CNV LE
CT09 (FAMILY CT V)	C>T73 heterozygous	exon 2	R25W	10-F	9	vitelliruptive	fibrosis	20/20	20/200	-
CT10 (FAMILY CT V)	C>T73 heterozygous	exon 2	R25W	36-F	30-	vitelliruptive	vitelliruptive	20/63	20/63	-
CT11 (FAMILY CT V)	C>T73 heterozygous	exon 2	R25W	70-M	60-	pseudohypopion	none	20/50	20/20	-
CT12 (FAMILY CT VI)	T>C26 heterozygous	exon 2	V9A	44-M	7-	atrophy	fibrosis	20/50	20/200	-
CT13 (FAMILY CT VI)	T>C26 heterozygous	exon 2	V9A	12-F	12-	pre-vitelliform	pre-vitelliform	20/20	20/20	-

M: male; F: female; RE: right eye; LE: left eye; BCVA: best corrected visual acuity; CNV: choroidal neovascularization.