

Supplementary table . List of mutations for each patient and visual acuity.

Patient	Age	Mutation	Type of mutation	BCVA (LogMAR)	
				Right eye	Left eye
1	54	c.877C>T; p.R293X	Nonsense	3.00	3.00
2	29	c.1245_1246delins14; p.K415NfsX	Deletion	0.00	0.00
3	12	c.1584_1587delTGTT; p.P529Hfs	Deletion	0.08	3.00
4	30	c.116+1G>A (intron 2)	Splice site	0.08	0.08
5	33	c.759delA; p.R253Rfs	Deletion	0.17	0.30
6	32	Deletion of exons12-13	Deletion	0.17	0.17
7	36	c.116+1G>A (intron 2)	Splice site	0.08	0.00
8	20	Deletion of exon 3	Deletion	0.30	0.17
9	57	c.1347C>G; p.Y449X	Nonsense	N/A	0.47

BCVA: Best corrected visual acuity, N/A not available