

Appendix 1. Results of genetic testing and clinical features covering laterality, age at diagnosis, staging at diagnosis

samples	sex	laterality	staging (L/R)	age (months)	Location	cDNA change	protein	mutational type	genotype
12F373	F	B	D/Extra-ocular	14	Exon11	c.1072C>T	p.R358X	nonsense	heterozygote
12F1020	M	B	C/B	18	Exon12	c.1183C>T	p.Q395X	nonsense	heterozygote
13F0569	F	B	B/E	2	Exon13	c.1332G>C	p.Q444H	missense	heterozygote
11F104	F	B	C/B	9	Exon14	c.1333C>T	p.R445X	nonsense	heterozygote
12F031	F	B	E/D	10	Exon14	c.1346G>A	p.G449E	missense	heterozygote
12F174	M	B	A/B	2	Exon14	c.1363C>T	p.R455X	nonsense	heterozygote
11F122	M	B	E/C	3	Exon15	c.1399C>T	p.R467X	nonsense	heterozygote
11F121	M	B	E/B	8	Exon2	c.142_170del	p.E48YfsX52	frameshift	heterozygote
-	F	B	A/E	3	Exon16	c.1468G>A	p.A490T	missense	heterozygote
13F0937	F	B	E/Extra-ocular	25	Exon17	c.1654C>T	p.R552X	nonsense	heterozygote
13F0958	M	B	D/C	8	Exon17	c.1689G>A	p.W563X	nonsense	heterozygote
12F1048	M	B	E/C	17	Exon18	c.1735C>T	p.R579X	nonsense	heterozygote
12F805	F	B	E/C	14	Exon18	c.1735C>T	p.R579X	nonsense	heterozygote
13F0020	F	B	E/B	5	Exon18	c.1735C>T	p.R579X	nonsense	heterozygote
12F875	F	B	D/E	18	Exon2	c.179T>A	p.L60X	nonsense	heterozygote
12F628	M	U	N/E	5	Exon19	c.1861C>A	p.R621S	missense	heterozygote
S101801	F	B	B/Extra-ocular	11	Exon19	c.1947delG	p.L649LfsX9	frameshift	heterozygote
13F412	F	B	C/C	7	Exon19	c.1954_1959dupA	p.V654MfsX14	frameshift	heterozygote
12F1028	M	B	E/D	9	Exon20	c.1981C>T	p.R661W	missense	heterozygote
13F1190	F	B	D/E	21	Exon20	c.1994T>C	p.L665P	missense	heterozygote
12F840	F	B	E/N	9	Exon2	c.224G>A	p.W75X	nonsense	heterozygote
12F1015	F	B	C/A	12	Exon23	c.2359C>T	p.R787X	nonsense	heterozygote
12F037	F	B	E/D	12	Exon23	c.2403delA	p.G801GfsX9	frameshift	heterozygote
13F0100	F	U	E/N	31	Exon23	c.2455C>G	p.L819V	missense	homozygote
13F0473	M	B	E/D	8	Exon24	c.2490-1G>A	-	splice	heterozygote
12F245	M	B	B/E	22	Exon24	c.2501C>A	p.S834X	nonsense	heterozygote
13F0168	M	B	D/Extra-ocular	13	Exon6	c.584G>T	p.W195L	missense	heterozygote
13F0091	M	U	N/Extra-ocular	12	Exon6	c.607+1G>C	-	splice	heterozygote
12F1002	M	B	C/E	8	Exon8	c.751C>T	p.R251X	nonsense	heterozygote
12F030	M	U	E/N	27	Exon9	c.920C>T	p.T307I	missense	heterozygote
12F650	M	B	B/E	20	Exon10	c.963C>G	p.Y321X	nonsense	heterozygote
12F1080	M	B	E/C	15	Exon10	c.967G>T	p.E323X	nonsense	heterozygote
12F022	F	B	D/C	1	Exon8 del	mlpa (+)	all exon del		heterozygote
12F545	M	B	E/C	9	sequencing (-)	mlpa (+)	all exon del		heterozygote
12F301	M	B	E/C	12	sequencing (-)	mlpa (+)	all exon del		heterozygote
12F0800	M	B	E/D	16	sequencing (-)	mlpa (+)	all exon del		heterozygote
13F1325	F	B	E/D	8	sequencing (-)	mlpa (+)	all exon del		heterozygote
12F144	F	B	A/E	5	sequencing (-)	mlpa (+)	exon 13-16 del		heterozygote
13F0085	F	B	C/D	2	sequencing (-)	mlpa (+)	exon 7-17 del		heterozygote
13F0113	F	B	E/C	2	sequencing (-)	mlpa (+)	exon 7-9 del		heterozygote
13F0285	M	B	C/B	12	sequencing (-)	mlpa (-)			
12F196	M	B	B/E	27	sequencing (-)	mlpa (-)			
12F1023	M	B	A/C	17	sequencing (-)	mlpa (-)			

13F1240	M	U	E/N	41	sequencing	(-)	mlpa	(-)
13F1128	M	U	N/E	16	sequencing	(-)	mlpa	(-)
13F0350	F	U	N/E	27	sequencing	(-)	mlpa	(-)
13F1168	F	U	E/N	55	sequencing	(-)	mlpa	(-)
12F354	M	U	E/N	12	sequencing	(-)	mlpa	(-)
13F0949	F	U	E/N	8	sequencing	(-)	mlpa	(-)
12F117	M	U	D/N	15	sequencing	(-)		
S101802	M	U	N/E	24	sequencing	(-)		
12F0001	M	U	N/E	18	sequencing	(-)		
12F872	M	U	N/E	13	sequencing	(-)		
12F1075	M	U	E/N	28	sequencing	(-)		
12F1054	M	U	N/E	22	sequencing	(-)		
S102902	M	U	N/E	39	sequencing	(-)		
13F0504	M	U	N/D	31	sequencing	(-)		
12F092	M	U	N/Extra-ocular	22	sequencing	(-)		
12F221	M	U	Extra-ocular/N	19	sequencing	(-)		
12F283	M	U	Extra-ocular/N	15	sequencing	(-)		
12F810	M	U	N/E	18	sequencing	(-)		
11F115	F	U	E/N	20	sequencing	(-)		
12F024	F	U	N/E	18	sequencing	(-)		
12F698	F	U	E/N	20	sequencing	(-)		
S101201	F	U	C/N	13	sequencing	(-)		
12F961	F	U	E/N	33	sequencing	(-)		
13F0634	F	U	E/N	19	sequencing	(-)		
12F463	F	U	N/D	62	sequencing	(-)		
13F1138	F	U	N/E	49	sequencing	(-)		
13F0881	F	U	N/E	33	sequencing	(-)		
12F148	F	U	N/E	28	sequencing	(-)		
12F069	F	U	N/E	28	sequencing	(-)		
12F512	F	U	E/N	25	sequencing	(-)		
13F990	F	U	N/Extra-ocular	41	sequencing	(-)		
13F0276	F	U	N/Extra-ocular	39	sequencing	(-)		
S092401	F	U	N/Extra-ocular	20	sequencing	(-)		
13F0416	F	U	Extra-ocular/N	31	sequencing	(-)		
13F1212	F	U	Extra-ocular/N	13	sequencing	(-)		
13F0215	F	U	N/Extra-ocular	15	sequencing	(-)		
13F0211	F	U	Extra-ocular/N	52	sequencing	(-)		
13F0488	F	U	N/Extra-ocular	51	sequencing	(-)		
12F1041	F	U	N/Extra-ocular	47	sequencing	(-)		
12F364	F	U	Extra-ocular/N	40	sequencing	(-)		
12F119	F	U	Extra-ocular/N	28	sequencing	(-)		
12F038	F	U	Extra-ocular/N	24	sequencing	(-)		

F: female. M: male. B: bilateral. U: unilateral. L: left eye. R: right eye. N: normal.