

Appendix 1. Clinical details of all cases.

Patient	Gender	Family History	Visual Acuity	Final Ocular Diagnosis	Other Ocular Diagnoses	Sequence Variations
30	M	Negative	20/310 OU	Colobomatous microphthalmia OU	Epiphora OS	E44/D44
45	M	Negative	1/200 OD 20/40 OS	Coloboma (iris OS, chorioretinal OD)	None	Q294Q and E44/D44
66	M	Negative	20/15 OU	Coloboma, retinal OD	Hyperopia	R66T, Q294Q, and E44/D44
124	M	Negative	20/30 OD CF 1' OS	Microphthalmia and cataract OU	Ectopic pupils OU	None
138	M	Negative	F and F OU	Colobomatous microphthalmia with cyst OU	Orbital cyst OD	None
159	M	Negative	F and F OU	Iris coloboma OU, cataract OU	None	Q294Q and E44/D44
176	F	Positive (father)	20/40 OD 20/65 OS	Coloboma (iris OS, retina OU)	None	None
182	F	Negative	20/25 OD 20/30 OS	Colobomatous microphthalmia (iris OS, retina OU)	Heterochromia	None
187	M	Negative	Prosthesis OD LP OS	Anophthalmia OD, Microphthalmia OS	None	E44/D44
230	M	Negative	20/400 OD 20/200 OS	Nanophthalmos OU	Cataracts OU, recurrent retinal detachment OD	None
231	M	Negative	6/200 OD 20/40 OS	Colobomatous microphthalmia OU (iris, uvea)	None	None
274	F	Negative	HM OD 20/50 OS	Microphthalmia, coloboma (iris, retinal) OU	None	None
277	F	Negative	6"/200 OD LP OS	Microphthalmia OU	Glaucoma OU, band keratopathy OD, persistent fetal vasculature	None
312	F	Negative	F and F OU	Microphthalmia OS	None	None
349	F	Positive (son)	20/400 OD NLP OS	Coloboma OU	None	None
353	F	Negative	20/20 OD 20/400 OS	Optic nerve coloboma OS	None	Q294Q
415	M	Negative	1/200 OD 20/15 OS	Optic nerve coloboma OU	Exotropia, optic nerve hypoplasia	None
437	F	Negative	LP OD 1/200 OS	Microphthalmia OU	Retinal detachment OD	Q294Q and E44/D44
444	M	Negative	20/20 OU	Optic nerve coloboma OD	None	None
475	F	Positive (AD)	20/30 OD F and F	Microphthalmia OS	None	E44/D44
702	F	Negative	20/80 OD 20/100 OS	Macular coloboma OU	Ocular albinism with Hermansky Pudlak syndrome, exotropia	Q294Q
737	M	Negative	20/20 OD 20/400 OS	Colobomatous microphthalmia OD	Anterior polar cataract	None
738	M	Negative	20/20 OU	Iris coloboma OS	None	None
918	F	Negative	NLP OU	Anophthalmia OU with brain malformations	None	None

M = male, F = female, AD = autosomal dominant, OD = right eye, OS = left eye, OU = both eyes