

**Table S1.** Overview of the patient demographics and clinical phenotype. Mutations shaded in grey are inconclusive.

Family history (FH), Transillumination defect (TID), Unaffected (U), Affected (A), Present (P), Absent (-), Inconclusive (I), Declined (D), Fixing & following (FF).

Family ID	Gender	F H	Ethnicity	Consanguinity	Hair & skin	VA	Nystagmus	Iris TID	Foveal hypoplasia	Fundus hypopigmentation	Chiasmal misrouting
25544	M	-	African	-	U	0.74	P	-	P	-	P
25578	F	-	Mixed: White / African	-	U	0.2	P	-	-	P	P
25704	M	-	Mixed: White / Asian	-	U	1.12	P	-	-	P	P
25806	M	P	African	-	U	0.7	P	-	P	-	P
25888-1	F	P	African	-	U	0.3	-	P	P	P	I
25888-2	F	P	African	-	U	0.3	-	P	P	P	P
26015	M	-	White other	-	U	0.78	P	-	-	-	P
26065	M	-	White other	-	U	0.1	P	-	-	P	P
26261	F	-	South Asian	-	U	0.58	P	-	P	-	P
26352	F	-	Middle Eastern	-	A	0.5	P	P	P	P	D
26640	F	-	South Asian	-	U	0.6	P	-	P	P	P
26641	F	-	White other	-	U	0.6	P	-	P	P	P
26680	M	-	White British	-	U	0.64	-	P	P	P	P
26677	M	-	South Asian	-	A	0.6	P	P	P	-	P
25246	F	P	Middle Eastern	-	U	0.4	P	-	P	P	P
26649	M	-	White British	-	A	0.54	P	P	P	P	P
26847	M	-	South Asian	P	U	0.8	P	P	P	P	P
26876	M	-	Mixed: White / African	-	U	0.6	P	P	P	P	P
26905	F	-	South Asian	-	A	FF	P	-	P	P	P
26948-1	F	P	Mixed: White / African	-	A	0.15	-	-	-	-	P
26948-2	F	P	Mixed: White / African	-	A	0.1	-	-	-	-	P
26947	F	-	Mixed: White / African	-	A	0.3	P	-	-	-	P
26984	F	-	Black African	-	U	0.7	P	-	P	-	P
27099	M	-	Black African	-	U	1.12	P	-	-	-	P
27101	M	-	Black African	-	U	0.44	P	-	-	-	P
23192-1	F	P	South Asian	-	A	FF	P	P	P	P	I
23192-2	F	P	South Asian	-	A	0.84	P	P	P	P	P
23192-3	M	P	South Asian	-	A	0.2	-	P	P	P	P
27268	M	-	White British	-	U	0.2	P	-	P	P	P
27321	M	P	Black African	-	A	1.2	P	P	P	P	P

27432	F	-	White other	-	U	0.26	P	-	-	P	-
27431	F	-	White other	-	A	0.4	P	P	P	P	P
27455	M	-	White British	-	U	0.38	P	-	P	P	D
27451	M	-	African	P	U	0.6	P	-	P	P	P
27560	M	-	White British	-	U	0.2	P	P	P	P	D
27655	M	-	South Asian	P	U	0.3	P	-	-	-	P
26903	F	P	South Asian	P	U	1.0	P	P	P	P	D
26956	F	-	White British	-	U	0.6	P	P	P	P	-
27033	F	-	White British	-	A	0.3	P	P	P	P	P
27047	M	-	Mixed: White / African	-	A	0.6	P	P	P	P	P
27079	M	-	White British	-	U	1.0	P	P	P	P	P
13332	M	-	White British	-	A	1.0	P	P	P	P	D
27430	F	-	White British	-	U	0.6	P	P	P	P	D
22151	M	P	White British	-	A	0.78	P	P	P	P	D

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