

Supplementary table 2: Clinical Data

Subject	Age of onset	Principal Symptom(s)*	Age (y)	Fundus Appearance	VA (LogMAR)	Latest refractive error
P1	< 3mo	Nystagmus	34	Unremarkable Disc pallor Thin vessels	R: 1.3 L: 1.2	R: +1.00/-2.00 x 70° L: +1.00
			56	Unchanged	R: 1.3 L: 1.3	
P2A	1-3yo	Nystagmus Photophobia	53	Poorly defined macula, Central RPE atrophy. Hyperpigmented area inferotemporal to the right macula	R: 1.8 L: 1.5	R: +1.50/-0.50 x 20° L: +1.75/-1.75 x 160°
			63	Unchanged	R: 1.8 L: 1.6	
P2B	3-12mo	Nystagmus Photophobia	54	Poorly defined macula RPE atrophy Small foci of hypertrophy	BE: HM	Emmetropia
			58	Unchanged	BE: HM	
P3	< 3mo	Nystagmus Photophobia	4	Central and mid peripheral RPE atrophy	BE: 0.78	R: -0.50/-4.00 x 10° L: +0.50/-4.00 x 170°
			60	Unchanged	R: 1.3 L: 1.0	
P4	< 3mo	Nyctalopia Reduced color vision	14	Blonde fundus	R: 0.4 L: 0.5	R: -3.75/-1.00 x 5° L: -3.75/-2.50 x 180°
			23	Unchanged	R: 0.48 L: 0.6	
P5	< 3mo	Nystagmus No eye contact Nyctalopia Glare in daytime	8	Peripheral hypopigmentation	R: 0.48 L: 0.62	R: +2.00/-2.25 x 5° L: +2.50/-2.25 x 180°
			11	Unchanged	R: 0.48 L: 0.54	
P6	< 3mo	Nystagmus Eye poking Nyctalopia	17	Unremarkable	BE: NPL	NA
			27	Unchanged	BE: NPL	
P7	< 3mo	Nystagmus Eye poking Nyctalopia	1	Blonde fundus	BE: NPL	BE: +7.00
			2	Unchanged	BE: NPL	
P8A	< 3mo	Nystagmus Nyctalopia Eye poking	14	Unremarkable fundus apart from thin vessels	BE: HM	NA
			12	Unchanged	BE: HM	
P8B	< 3mo	Nystagmus Not fixing Eye poking	1	Unremarkable	BE: fixate at large object	BE: +7.00
			11	Unchanged	BE: HM	
P9	3-12mo	Nystagmus Photophobia Nyctalopia	1	Unremarkable	BE: fixate at large object	BE +5.00
			16	Waxy optic discs and thin retinal vessels	BE: 1.2	

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P10	3-12mo	Nystagmus Eye poking	17	Unremarkable	BE: NPL	NA
			27	Unchanged	BE: NPL	
P11	3-12mo	Nystagmus Eye poking	<1	Unremarkable	BE: PL	BE +4.50/+1.50 x 90°
			1	Unchanged	BE: PL	
P12	< 3mo	Nystagmus Poor vision	2	Unremarkable	BE: HM	RE +6.50/-1.00 x 25° LE +6.50/1.00 x 160°
			16	Minimal retinal atrophy	BE: CF	
P13	< 3mo	Nystagmus	60	Peripheral RPE atrophy Pigment hypertrophy with large lacunae of chorioretinal atrophy peripherally Disc pallor Thin vessels	BE: HM	NA
			79	Unchanged	BE: PL	
P14	3-12mo	Nystagmus Nyctalopia	1	Pale discs Thin vessels Fine pigment changes	BE: fixate at large object	Emmetropia
			14	Unchanged	BE: PL	
P15	< 3mo	Nystagmus Eye poking	1	NA	BE: NPL	R +10.00/-2.00 x 30° LE +6.50/1.00 x 160°
			2	Pale disc Otherwise unremarkable	BE: NPL	
P16	< 3mo	Poor color vision Nystagmus Photophobia	10	Unremarkable	R: 1.56 L: 1.76	R: +1.25 L: +1.00
			19	Unchanged	R: 2.00 L: 1.76	
P17	< 3mo	Nystagmus Eye poking	1	Unremarkable Thin vessels	BE: PL	BE: +8.50
			3	Unchanged	BE: PL	
P18	< 3mo	Nystagmus Eye poking Photophobia	6	Blonde fundus	BE: PL	R: -7.00 L: -8.00
			10	Unchanged	BE: PL	
P19	< 3mo	Nystagmus	8	Unremarkable	BE: PL	R: +7.50 L: +6.50/+3.00 x 125°
			18	Unchanged	BE: PL	

*All patients presented with reduced visual acuity

R; right eye, L; left eye, BE; both eyes, mo; months old, yo; years old, y; years, PL; light perception, NPL = no perception of light; HM = hand motions; NA= not available