

Appendix 1. Clinical features of patients identified with *RDH12* mutations

Family	Gender	Age initial visual loss, further central visual deterioration (yr)	Age of nyctalopia (yr)	Age at diagnosis (yr)	Presenting symptoms	Visual Acuity in logMAR (age, yr)	Refraction	Anterior segment	Fundus
1	F	3, 34	7	3	Reduced vision	PL EE (38)	+7.50 DS EE	PSCLO	Dense retinal pigment clumping with para arteriolar sparing, atrophic macula with pigment, arteriolar attenuation
2	F	1.5, 19	Birth	8	Reduced vision	2.30 EE (28)	-1.63/+1.73, 97 RE -6.63/+1.38, 75 LE	Normal	Dense retinal pigment clumping, macular atrophy, pale discs, arteriolar attenuation
3	F	5, 7	5	5	Reduced vision	0.78 RE HM LE (27)	N/A	PSCLO	Dense retinal pigment clumping with para arteriolar sparing, atrophic yellow macula with pigment, arteriolar attenuation
4	M	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
5	F	0.5, 26	1	2	VF constriction	PL EE (36)	-0.25/+1.5, 83 RE 0.00/+1.75, 96 LE	Polar cataracts	Moderate retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation
6	F	3	3	4	Reduced vision	0.70 EE (5)	+1.50/+0.75, 100 RE +2.25/+0.50, 90 LE	Normal	Minimal retinal pigment clumping, widespread RPE atrophy, atrophic macula
7	M	2	N/A	5	Reduced vision, VF constriction	HM EE (34)	N/A	PSCLO	Dense retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation
8	F	7	6	6	Reduced vision, nyctalopia	1.00 EE (27)	0.00/+1.5, 90 EE	Normal	Moderate retinal pigment clumping, atrophic macula with pigment, arteriolar attenuation
9	F	1.5, 20	10	3	VF constriction	PL EE (32)	N/A	PSCLO	Dense retinal pigment clumping, atrophic macula, arteriolar attenuation
10	M	4	4	5	VF constriction	0.60 EE (6)	N/A	Normal	Retinal pigment clumping, atrophic macula
11	F	Birth	Birth	3.5	Reduced vision	1.48 RE PL LE (11)	+1.00/-0.75, 90 RE +1.50/-2.00, 100 LE	Normal	Moderate retinal pigment clumping, RPE atrophy, atrophic yellow macula, arteriolar attenuation
12	F	Birth	N/A	18	Reduced vision, VF constriction	HM EE (22)	N/A	Normal	Dense retinal pigment clumping with para arteriolar sparing, atrophic macula with pigment, arteriolar attenuation
13	M	2.5	8	6	Nyctalopia	CF EE (29)	N/A	PSCLO	Moderate retinal pigment clumping, atrophic macula, arteriolar attenuation
14	F	3.5	N/A	3.5	Strabismus	0.80 EE (13)	+ 0.50/+4.50, 80 EE	Normal	Dense retinal pigment clumping, atrophic macula with pigment, arteriolar attenuation
	F	3.5, 7	3.5	3.5	Strabismus	1.08 RE CF LE(17)	-1.00/+2.50, 120 RE +1.50/+1.00, 80 LE	PSCLO	Dense retinal pigment clumping, atrophic macula with pigment, arteriolar attenuation

15	F M	Birth, 14 4, 29	Birth 4	15 21	Reduced vision Reduced vision, nyctalopia	CF RE HM LE (27) 1.00 EE (34)	-1.50/+2.00, 85 RE -1.50/+2.25, 95 LE + 0.25/-3.50, 5 RE -1.00/-3.00, 175 LE	Normal PSCLO	Moderate retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation Moderate retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation, mid-peripheral pericentric spared zone
16	M	4	5	6	Reduced vision, strabismus	1.00 RE 1.20 LE (12)	+5.50/-2.00, 15 RE +5.00/-1.00, 180LE	Normal	Minimal retinal pigment clumping, widespread RPE atrophy, atrophic yellow macula
17	M	0.75	0.5	2	Reduced vision, VF constriction, nyctalopia	1.00 RE 0.90 LE (8)	N/A	Normal	No retinal pigment clumping, widespread RPE atrophy, atrophic yellow macula
18	M	0.25	2	7	Reduced vision	1.00 RE 1.30 LE (8)	+3.50 DS RE +4.00 DS LE	Normal	Moderate retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation
19	M F	Teens 8	4 N/A	4 6	N/A N/A	HM EE (27) HM EE (25)	N/A N/A	RE PCIOL; LE PSCLO Normal	Widespread retinal pigment clumping, macular atrophy Dense retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation
20	F	Birth, 27	Birth	30	Reduced vision	1.00 RE 0.78 LE (33)	N/A	PSCLO	Dense retinal pigment clumping with para arteriolar sparing, atrophic yellow macula with pigment, arteriolar attenuation
21	M	N/A	N/A	10	Reduced vision	0.78 RE 1.00 LE (18)	+0.25/-1.50, 15 RE +1.00/-1.50, 170 LE	Normal	Moderate retinal pigment clumping, RPE atrophy, atrophic pigmented macula arteriolar attenuation mid-peripheral pericentric spared zone
22	F	2	3	7	Strabismus	1.00 RE 0.78 LE (20)	+1.25/-0.50, 180 RE +1.00/-0.75, 180 LE	Normal	Dense retinal pigment clumping with para arteriolar sparing, atrophic yellow macula with pigment, arteriolar attenuation
23	F	1	N/A	N/A	Reduced vision, nyctalopia	PL EE (52)	N/A	PSCLO	Dense retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation
24	M	2	2	2	Reduced vision, nyctalopia	HM EE (29)	N/A	Keratoconus PSCLO	Dense retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation
25	M	Birth	N/A	N/A	Reduced vision	PL EE (21)	N/A	PSCLO	Dense retinal pigment clumping with para arteriolar sparing, atrophic yellow macula with pigment, arteriolar attenuation
26	F	11	11	12	Reduced vision	PL EE (30)	N/A	PSCLO	Moderate retinal pigment clumping, widespread macular atrophy and yellowing, arteriolar attenuation
27	M	10,35	18	32	Reduced vision	1.78 RE 1.48 LE (40)	N/A	PSCLO	Dense retinal pigment clumping, atrophic macula with pigment, arteriolar attenuation
28	M	4, 35	4	5	Reduced vision, VF constriction	HM EE (35)	N/A	Normal	Dense retinal pigment clumping, atrophic yellow macula with pigment, arteriolar attenuation
29	M	5	5	5	N/A	1.30 RE 1.00 LE (33)	N/A	Bilateral PCIOL	Dense retinal pigment clumping with para arteriolar sparing, atrophic yellow macula with pigment, arteriolar attenuation

Abbreviations in table; CF- counting fingers; DS- dioptre sphere; EE – either eye; F-Female; HM-Hand movements; LE – left eye; M-Male, N/A – not available; PCIOL- posterior chamber intra ocular lens; PL-Perception of light; PSCLO – posterior subcapsular lens opacification; RE- right eye; VF-Visual field.