

Supplementary Table 1. Potential causative variants and clinical characteristics of 42 patients with suspected retinitis pigmentosa

Patient	Gender	Gene	Age of loss vision (y)	Current age (y)	Current signals and symptoms	Current BCVA (OD/OS)	Biomicroscopy	Fundus Photography
1	F	<i>RHO</i>	27	32	Visual loss	20/30 (-4,50 -2,50 x 10) 20/30 (-5,00 -2,00 x 155)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in periphery
2	F	<i>RHO</i>	26	51	Visual loss	20/30 (+3,25 -1,00 x 154) 20/30 (+3,25 -1,50 x 148)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in inferior periphery
3	M	<i>RHO</i>	5	49	Visual loss	20/30 (-0,25 -0,50 x 30) 20/30 (-0,50 -0,75 x 170)	OU: Normal	OU: Pallor of optic disc, narrow vessels, pigmentations in periphery
4	M	<i>RHO</i>	37	47	Visual loss, photophobia	20/160 (-7,50 -0,00 x 00) 20/400 (-7,50 -0,00 x 00)	OU: Posterior subcapsular cataract	OU: Pallor of optic disc with peripapillary atrophy, narrow vessels, pigmentations in periphery
5	F	<i>RHO</i>	16	45	Visual loss, nyctalopia, photophobia	20/64 (+2,00 -1,00 x 11) 20/50 (+2,00 -1,25 x 160)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in periphery OD: Macular macroaneurysm with lipid exudation surround the fovea
6	F	<i>RHO</i>	8	28	Visual loss, nyctalopia	20/30 (0,00 -2,50 x 7) 20/30 (0,00 -2,75 x 166)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in the periphery
7	F	<i>RHO</i>	13	18	Visual loss, nyctalopia	20/40 (0,00 -1,50 x 14) 20/50 (0,00 -0,50 x 163)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in the periphery
8	F	<i>RHO</i>	5	9	Visual loss, nyctalopia	20/50 (-0,75 -0,75 x 12) 20/50 (-0,50 -2,50 x 178)	OU: Normal	OU: Normal optic disc, narrow vessels, pigmentations in the periphery
9	F	<i>RPGR</i>	19	20	Visual loss, nyctalopia	20/30 (0,00 -2,00 x 5) 20/25 (0,00 -1,75 x 180)	OU: Normal	OU: Normal optic disc, normal vessels, RPE rarefaction in periphery
10	M	<i>RPGR</i>	5	23	Visual loss, nyctalopia	20/125 (-7,00 -1,00 x 40) 20/100 (-6,50 -1,25 x 150)	OU: Normal	FP: NP Fundus examination of OU: Mild pallor of optic disc, narrow vessels, pigmentations in the periphery
11	F	<i>RPGR</i>	19	46	Visual loss, nyctalopia	20/80 (-11,00 -1,00 x 150) 20/80 (-11,00 -1,00 x 180)	OU: Posterior subcapsular cataract	OU: Pallor of optic disc, narrow vessels, pigmentations in the periphery
12	M	<i>RPGR</i>	15	23	Visual loss, nyctalopia	20/50 (NP) 20/400 (NP)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in the periphery
13	M	<i>RPGR</i>	29	39	Visual loss, nyctalopia, photophobia	20/63 (-3,25 -1,50 x 60) 20/80 (-3,25 -0,00 x 00)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in the periphery
14	M	<i>RPGR</i>	17	27	Visual loss, nyctalopia	20/100 (-2,50 -4,50 x 60) 20/125 (-3,00 -3,50 x 80)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in the periphery
15	M	<i>RPGR</i>	15	20	Visual loss	20/32 (-2,50 -0,75 x 35) 20/40 (-1,50 -1,00 x 115)	OU: Posterior subcapsular cataract	OU: Mild pallor of optic disc, narrow vessels, pigmentations in periphery OS: Lattice degeneration in in the nasal periphery
16	M	<i>USH2A</i> <i>USH2A</i>	39	49	Visual loss, photophobia	20/32 (-0,50 -0,25 x 70) 20/32 (-0,75 -0,00 x 00)	OU: Normal	OU: Pallor of optic disc, narrow vessels, pigmentations in the periphery
17	M	<i>USH2A</i> <i>USH2A</i>	30	40	Visual loss, nyctalopia	20/25 (0,00 -0,75 x 90) 20/32 (0,00 -0,50 x 73)	OU: Normal	OU: Pallor of optic disc, narrow vessels, pigmentations in the periphery
18	M	<i>USH2A</i>	30	38	Visual loss, nyctalopia	20/30 (-0,25 -1,25 x 102) 20/30 (-0,75 -0,25 x 79)	OU: Cataract	OU: Pallor of optic disc, narrow vessels, pigmentations in the periphery
19	M	<i>USH2A</i> <i>USH2A</i>	40	45	Visual loss, nyctalopia	20/50 (+0,75 -1,00 x 83) 20/30 (0,00 -0,50 x 39)	OU: Posterior subcapsular cataract	OU: Pallor of optic disc, narrow vessels, pigmentations in the periphery OD: Vitreous opacity
20	M	<i>AH11</i> <i>AH11</i>	24	36	Visual loss, nyctalopia	20/50 (-1,50 -1,25 x 23) 20/50 (-1,50 -0,25 x 48)	OU: Posterior subcapsular cataract	OU: Pallor of optic disc, narrow vessels, pigmentations in periphery
21	M	<i>AH11</i> <i>AH11</i>	24	34	Visual loss, nyctalopia	20/100 (-0,75 -0,75 x 87) 20/100 (-0,50 -0,25 x 132)	OU: Normal	OU: Pallor of optic disc, narrow vessels, pigmentations in periphery
22	M	<i>CLN3</i>	5	13	Visual loss, nyctalopia	Light perception (NP) Light perception (NP)	OU: Normal	OU: Pallor of optic disc, macula with focal pigmentations, narrow vessels, pigmentations in periphery
23	M	<i>CLRN1</i>	10	10	Visual loss, nyctalopia	20/30 (0,00 -1,50 x 30) 20/30 (0,00 -1,50 x 172)	OU: Normal	OU: Mild pallor of optic disc, normal vessels, pigmentations in periphery
24	F	<i>CRB1</i> <i>CRB1</i>	8	18	Visual loss, nyctalopia	20/200 (0,00 -0,25 x 180) 20/125 (0,00 -1,00 x 180)	OU: Pigmented cells into the anterior vitreous	OU: Mild pallor of optic disc, narrow vessels, few pigmentations in periphery, atrophic chorioretinal regions in the far periphery
25	M	<i>CRB1</i> <i>CRB1</i>	6	11	Visual loss, nyctalopia, photophobia	20/63 (-4,00 -3,00 x 20) 20/50 (-3,50 -4,00 x 180)	OU: Normal	OU: Mild pallor of optic disc, normal vessels with hypopigmented area around the arterial arcades, few pigmentations in periphery
26	F	<i>EYS</i> <i>EYS</i>	46	56	Visual loss, nyctalopia	20/50 (-9,50 -0,00 x 00) 20/50 (-9,50 -0,00 x 00)	OU: Nuclear and posterior subcapsular cataract	OU: Pallor of optic disc and peripapillary atrophy, narrow vessels, pigmentations in periphery
27	M	<i>EYS</i> <i>EYS</i>	40	55	Visual loss, nyctalopia	20/125 (-1,50 -1,50 x 10) 20/100 (-1,75 -1,50 x 170)	OU: Normal	OU: Pallor of optic disc, narrow vessels, pigmentations in periphery
28	M	<i>EYS</i> <i>EYS</i>	49	59	Visual loss, nyctalopia	20/25 (-0,75 -0,00 x 00) 20/32 (-0,75 -0,00 x 170)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in periphery more concentrated inferiorly
29	M	<i>FSCN2</i>	11	90	Visual loss, nyctalopia, photophobia	Count fingers (+1,25 -4,50 x 91) Count fingers (-0,50 -0,50 x 130)	OD: Nuclear cataract OS: pseudophakia	OU: Pallor and increased excavation of optic disc, narrow vessels, pigmentations in periphery, multi atrophic chorioretinal patches in periphery
30	F	<i>OPA1</i>	20	25	Visual loss, nyctalopia	20/80 (-2,50 -1,00 x 160) 20/32 (-1,00 -0,75 x 90)	OU: Posterior subcapsular cataract	OU: Mild pallor of optic disc, narrow vessels, ghost vessels in OS, pigmentations in periphery
31	F	<i>PCARE</i>	19	49	Visual loss, nyctalopia	20/400 (0,00 -1,00 x 90) 20/800 (0,00 -0,75 x 90)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, few pigmentations in periphery, multi atrophic chorioretinal regions in the far periphery OS: asteroid hyalosis

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32	F	<i>PCARE</i>	29	44	Visual loss, nyctalopia	20/50 (+1,00 -1,75 x 30) 20/63 (+1,50 -2,50 x 170)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, few pigmentations in periphery
33	F	<i>PDE6A</i> <i>PDE6A</i>	25	50	Visual loss, nyctalopia, photophobia	20/100 (-1,00 -0,00 x 00) 20/200 (-1,00 -0,00 x 00)	OU: pseudophakia	OU: Pallor of optic disc, narrow vessels, pigmentations in periphery
34	F	<i>PHYC</i>	48	49	Visual loss, nyctalopia	20/30 (0,00 -1,00 x 90) 20/70 (0,00 -2,50 x 90)	OU: Shallow anterior chamber	OU: Pallor of optic disc and peripapillary atrophy, narrow vessels, pigmentations and multi atrophic chorioretinal regions in the periphery
35	F	<i>PRCD</i>	33	56	Visual loss, nyctalopia, photophobia	20/400 (-1,75 -3,25 x 70) Count fingers (-1,25 -3,00 x 120)	OD: Pseudophakia OS: nuclear and posterior subcapsular cataract	OU: Pallor of optic disc, narrow vessels, pigmentations in periphery
36	F	<i>PRCD</i>	40	50	Visual loss, nyctalopia	Count fingers (-1,50 -1,50 x 62) 20/120 (-2,00 -1,50 x 131)	OU: Posterior subcapsular cataract	OU: Pallor of optic disc, narrow vessels, pigmentations in periphery
37	F	<i>PRPF8</i>	21	39	Visual loss, nyctalopia	20/25 (+1,50 -1,00 x 175) 20/25 (+1,25 -0,25 x 165)	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in periphery
38	M	<i>PRPF31</i>	36	46	Visual loss, nyctalopia	20/25 (0,00 -0,00 x 00) 20/32 (0,00 -0,00 x 00) Before refractive surgery: +1,00 -3,00 x 10 +1,00 -3,00 x 10	OU: Normal	OU: Mild pallor of optic disc, narrow vessels, pigmentations in periphery
39	F	<i>PRPH2</i>	49	57	Visual loss, nyctalopia	20/40 (-2,75 -1,25 x 80) 20/40 (-1,00 -0,00 x 00) Before facectomy: -2,75 -2,25 x 17 -1,00 -0,75 x 170	OU: Normal	FP: NP Fundus examination of OU: Mild pallor of optic disc, macula without reflex, narrow vessels, pigmentations in periphery
40	F	<i>RPE65</i> <i>RPE65</i>	9	12	Visual loss, nyctalopia	20/60 (+0,75 -1,00 x 20) 20/60 (+0,75 -1,00 x 160)	OU: Normal	OU: Normal optic disc, normal vessels, few pigmentations and rarefaction of RPE in the periphery
41	F	<i>RPE65</i> <i>RPE65</i>	25	59	Visual loss, nyctalopia	20/60 (-6,00 -1,00 x 160) 20/60 (-5,50 -1,00 x 165)	OU: Pseudophakia	FP: NP Fundus examination of OU: Pallor of optic disc, narrow vessels, pigmentations in the periphery
42	M	<i>SDCCAG8</i> <i>SDCCAG8</i>	52	62	Visual loss, nyctalopia	20/60 (+0,50 -1,50 x 20) 20/800 (0,00 -0,00 x 00)	OU: Pseudophakia OS: subluxated IOL	OU: Pallor of optic disc, narrow vessels, pigmentations in the periphery

Legend: F: Female; M: Male; Y: Years; BCVA: Best Corrected Visual Acuity; OD: Right Eye; OS: Left Eye; OU: Both Eyes; RPE: Retinal Pigment Epithelium; NP: Not Performed.