

## Appendix 1: Summary of clinical features

Family # patient/age	Disease history			Refraction; visual acuity		Fundus	OCT	Goldman perimetry	ERG
	NB age*	PP age*	PV age*	OD	OS				
<b>PB15</b> <b>II:4/13</b>	? nd	? nd	? nd	-7 0.2	-6 0.2	No pigment deposits, alteration of macula	ND	Restriction of VF central scotoma OD	No ERG responses
<b>PB74</b> <b>II:1/46</b>	yes Ch	yes nd	yes nd	nd 0.8	nd 0.8	Pigment deposits 360° in peripheral retina	ND	Tubular VF (< 10°)	No ERG responses
<b>RP517</b> <b>II:1/32</b>	yes 3	? nd	no	-0.50(-1.00;15°) 0.33	(-1.50;140°) 0.5	Atrophic spots in peripheral retina, rare pigment clumps, whitish aspect of fundus	foveal thinning, fragmented IS/OS line OD, absent OS	Normal peripheral VF Large central scotoma OS	No scotopic responses barely detectable 3-Hz fl
<b>RP670</b> <b>II:2/66</b>	yes 40	yes 50	yes 50	-1.75(-0.75 ;170°) 0.6	-1.00(-1.50;145°) 0.3	Macular and peripheral retina pigment deposits as clumps & bone spicules, perifoveal atrophy	remaining foveal IS/OS line but absent outside fovea	Tubular VF 10-15° OD, 5-8° OS	No scotopic responses photopic responses at 20 % of nl value
<b>RP854</b> <b>III:2/37</b>	yes Ch	yes 14	yes 10	-9.00(-1.00;65°) HM	-9.75(-1.00;95°) HM	Macular and peripheral retina is atrophic, bone spicule pigment deposits 360° in peripheral retina	ND	Not detectable	No ERG responses
<b>RP1013</b> <b>III:2/29</b>	yes 18	yes 18	yes 14	-9.00(-2.00;25°) 0.3	-7.00(-1.00;135°) 0.4	Foveal atrophy and pigment deposits 360° in peripheral retina	ND	Tubular VF at 30°, annular scotoma at 20° OS	No scotopic responses barely detectable 30-Hz fl
<b>RP1324</b> <b>II:3/57</b>	yes 10	yes 10	yes 10	intraocular lens LP	intraocular lens LP	Whole retina is atrophic with pigment deposits overall retina	ND	Not detectable	ND
<b>RP1324</b> <b>II:8/44</b>	yes 10	yes 10	no 20	-1.25(-1.75;5°) 0.3	ocular phtisis no LP	Few pigment deposits in retinal periphery	Remaining IS/OS in fovea only, moderate macular edema	Tubular VF at 20-30°	No ERG responses
<b>RP1361</b> <b>II:1/19</b>	yes 10	yes 14	no	+0.50(-1.50;20°) 0.5	(-1.50;150°) 0.5	Macular edema, epiretinal membrane and very few pigment deposits in peripheral retina for both II:1 and II:2	Fragmented IS/OS line important macular edema	Normal peripheral VF	No scotopic responses barely detectable 30-Hz fl
<b>RP1361</b> <b>II:2/17</b>	yes 10	yes 13	yes 16	+0.25 0.6	(-0.75;175°) 0.7		Decreased IS/OS line and important macular edema	ND	No scotopic responses 30-Hz fl at 10% nl value
<b>RP1625</b> <b>II:2/10</b>	yes 4	yes Ch	yes Ch	-4.50(-2.75;5°) 0.6	-5.00(-2.50;165°) 0.6	Abnormal fovea, small pigment deposits	Thin retina and fovea, IS/OS visible at fovea	Tubular VF at 10° OD, 20° OS	No ERG responses
<b>RP1682</b> <b>II:3/58</b>	yes 1	yes 42	yes 30	+2.00(-1.25;180°) 0.4	+1.75(-1.25;140°) 0.3	Very large atrophic spots in retinal periphery, retinal vessels only moderately attenuated	IS/OS line visible in fovea only thin retina	Tubular VF at 3°	No ERG responses

\*: apparent age at onset; Ch: childhood; fl: flickers; NB: night blindness; nd: not determined; ND: not done; nl: normal; PP: photophobia; PV: peripheral vision impairment; OD: ocular dextra; OS: ocular sinistra  
HM: hand motion; LP: light perception