

Panel-based NGS Reveals Novel Pathogenic Mutations in Autosomal Recessive Retinitis Pigmentosa

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Supplementary Table 2. Clinical findings identified in the index cases of the characterized families.

Family	Symptoms and course	Fundus	ERG	Other features
RP-1147	NB (16y), VF constriction (30y) and VA diminished (30y)	Bone spicule pigmentation	ND	
RP-2066	NB (6y), loss of VF (6y) and VA diminished (10y)	Bone spicule pigmentation in mid periphery	ND	
RP-2114	NB (3y), loss of VF (3y) and VA diminished (4y)	Attenuated vessels and bone spicule pigmentation.	NR	Photofobia
RP-1319	NB (20y), VF constriction (22y) and loss of VA (28y)	Bone spicule pigmentation and macular edema.	ND	
RP-1412	NB (18y), VF constriction (30y) and loss of VA (38y)	ND	ND	
RP-1646	NB (48y), VF constriction (48y) and loss of VA (53y)	Bone spicule pigmentation	ND	
RP-1695	NB (7y), loss of VF (13y) and VA diminished (13y)	ND	NR	Photofobia
RP-1735	NB (30y), loss of VF (30y) and VA diminished (43y)	Pale optic disc. Bone spicule pigmentation and attenuated vessels.	Reduced a-wave and b-wave amplitudes	Bilateral sensorineural hearing loss
RP-1802	NB (48y), VF constriction (48y) and loss of VA (53y)	Bone spicule pigmentation	ND	Posterior subcapsular cataract (45y)
RP-1976	NB (40y), VF constriction (50y) and loss of VA (56y)	Pale optic disc. Bone spicule pigmentation and attenuated vessels.	NR	Posterior subcapsular cataract (66y) Photofobia
RP-1979	NB (30y), VF constriction (30y) and loss of VA (33y)	Pale optic disc. Bone spicule pigmentation and attenuated vessels.	NR	Posterior subcapsular cataract (49y). Bilateral sensorineural hearing loss
RP-2112	NB (30y), loss of VF (36y) and VA diminished (39y)	Bone spicule pigmentation	NR	
RP-2113	NB (35y), loss of VF (50y) and VA diminished (55y)	Pale optic disc. Bone spicule pigmentation	ND	Posterior subcapsular cataract (62y). Photofobia
RP-1543	NB (20y), VF constriction (23y) and loss of VA (8y)	Pale optic disc. Bone spicule pigmentation	NR	Nystagmus
RP-1056	NB (21y), loss of VF (28y) and VA diminished (30y)	ND	ND	Posterior subcapsular cataract (38y)
RP-1998	NB (12y), loss of VF (12y) and VA diminished (27y)	Attenuated vessels and initial pigment deposits	Reduced a-wave and b-wave amplitudes	
RP-1706	NB (10y), VF constriction (6y) and loss of VA (15y)	Pale optic disc. Bone spicule pigmentation and attenuated vessels.	NR	Posterior subcapsular cataract (42y)
RP-1929	NB (10y), VF constriction (10y) and loss of VA (10y)	Pale optic disc. Bone spicule pigmentation	ND	Posterior subcapsular cataract (70y)
RP-1142	NB (<10y), loss of VF (<10y) and VA diminished (<10y)	Pale optic disc. Bone spicule pigmentation and attenuated vessels.	NR	Posterior subcapsular cataract (70y). Dyschromatopsia
RP-0372	NB (5y), loss of VF (5y) and VA diminished (5y)	Bone spicule pigmentation and attenuated vessels.	Reduced a-wave and b-wave amplitudes	
RP-0040	NB (25y), VF constriction (25y) and loss of VA (28y)	Pale optic disc. Bone spicule pigmentation and attenuated vessels.	NR	Posterior subcapsular cataract (34y)
RP-1772	NB (20y), VF constriction (35y) and loss of VA (45y)	Bone spicule pigmentation and attenuated vessels.	Reduced a-wave and b-wave amplitudes	Photofobia
RP-1988	NB (6y), VF constriction (6y) and loss of VA (6y)	Bone spicule pigmentation and attenuated vessels.	ND	Photofobia. Dyschromatopsia
RP-1201	NB (16y), loss of VF (20y) and VA diminished (26y)	ND	ND	
RP-0338	NB (35y), loss of VF (35y) and VA diminished (35y)	Bone spicule pigmentation in mid periphery and attenuated vessels	NR	Bilateral sensorineural hearing loss
RP-0344	NB (21y), VF constriction (28y)	Pale optic disc. Bone spicule pigmentation and attenuated vessels.	NR	Photofobia. Bilateral sensorineural hearing loss
RP-0456	NB (38y), VF constriction (40y) and loss of VA (42y)	ND	ND	Posterior subcapsular cataract (40y)

Abbreviations: NB(night blindness); VF(VF); VA(visual acuity);ERG(electroretinogram); NR(non-recordable);ND(no data).