

Table S1: Clinical findings of probands

Patient ID	Age at Examination / Gender	Disease	Initial Symptom	Age of Onset	BCVA ^a (Right/Left)	Fundus Examination	OCT / FAF	OCTA	Fundus Photography	Additional Findings	Causative Gene	Genetic Diagnosis	Sanger Confirmation
P1	21 / M	Stargardt disease	DVA	11	0.1/0.1	Foveal atrophy and flecks	✓	✓	-	-	<i>ABCA4</i> <i>CLRN1</i>	Solved	✓
P2	42 / F	Usher syndrome	DVA	12	p+p-/p+p-	Diffuse retinal atrophy	✓	-	-	Bilateral HL, right ear hearing aid, bilateral cataract surgery	<i>USH2A</i>	Solved	✓
P3	59 / M	Retinitis pigmentosa	Nyctalopia	<10	HM/HM	Bone spicule pigmentations, optic disc pallor	✓	✓	-	Bilateral cataract surgery	-	Unsolved	-
P4	26 / F	Usher syndrome	DVA	7	0.2/0.2	Bone spicule pigmentations, optic disc pallor	✓	✓	-	Congenital bilateral SNHL, right ear cochlear implant, vestibular disturbance	<i>USH1C</i>	Solved	✓
P5	32 / F	Retinitis pigmentosa	DVA	14	0.2/0.3	Normal	✓	✓	-	-	-	Unsolved	✓
P6	27 / M	Retinoschisis	DVA	0	0.05/0.2	Retinoschisis	✓	✓	-	-	<i>RS1</i>	Solved	✓
P7	26 / F	Retinitis pigmentosa	DVA	7	0.4/0.4	Bone spicule pigmentations, optic disc pallor	✓	✓	-	History of speech delay, bilateral posterior subcapsular cataract	<i>IFT81</i>	Potentially solved	✓
P8	30 / M	Usher syndrome	DVA	17	0.5/0.5	Bone spicule pigmentations, optic disc pallor	✓	✓	-	Bilateral SNHL (age of onset 3)	-	Unsolved	✓
P9	51 / F	Retinitis pigmentosa	DVA	9	HM/HM	Diffuse atrophy, bone spicule pigmentations	✓	✓	-	Bilateral posterior subcapsular cataract	<i>ABCA4</i>	Solved	✓
P10	32 / F	Leber's congenital amaurosis	DVA	0	-	Rod-cone dystrophy	✓	-	-	Nystagmus	<i>RPGRIP1</i>	Solved	✓
P11	48 / M	Retinitis pigmentosa	Nyctalopia	20	FC(2)M/ FC(2)M	Retinitis pigmentosa	✓	✓	-	Bilateral cataract surgery	<i>RP2</i> <i>BEST1</i>	Solved	✓
P12	16 / M	Retinoschisis	DVA	0	0.3/0.2	Retinoschisis	✓	-	✓	Left eye laser photocoagulation	<i>RS1</i>	Solved	✓
P13	61 / F	Retinitis pigmentosa	Central vision loss	31	HM / FC(0.5)M	Diffuse chorioretinal atrophy	✓	✓	-	Bilateral cataract surgery	<i>PCARE</i>	Solved	✓
P14	48 / F	Pattern dystrophy	DVA	42	0.4/0.1	Pattern dystrophy	✓	✓	-	-	-	Unsolved	✓

P15	37 / F	Cone dystrophy	DVA	7	0.2/0.16	Macular scar	✓	✓	✓	✓	✓	ERG: supernormal rod responses	KCNJ2	Solved	✓
P16	37 / M	Cone dystrophy	DVA	33	0.1/0.1	-	-	-	-	-	-	-	-	Unsolved	-
P17	18 / M	Bietti crystalline dystrophy	DVA	10	-	Early-stage Bietti crystalline dystrophy	✓	✓	-	-	-	-	CYP4V2	Potentially solved	-
P18	24 / F	Bietti crystalline dystrophy	Abnormal color vision	24	-	Stage 2 Bietti crystalline dystrophy, hyperreflective crystals	✓	✓	-	-	-	-	CYP4V2	Solved	-
P19	18 / M	Optic atrophy	DVA	0	0.1/0.1	Pallor optic disc, cupping of the optic nerve head	✓	✓	-	-	-	-	OPAI IMPG1	Solved	-
P20	8 / F	Stargardt disease	DVA	8	-	Macular atrophy	✓	-	-	-	-	-	ABCA4	Solved	-
P21	47 / M	Cone-rod dystrophy	Abnormal color vision	39	-	-	-	-	-	-	-	Bilateral HL	DRAM2	Solved	-
P22	31 / M	Stargardt disease	Visual field loss	10	-	-	-	-	-	-	-	-	ABCA4	Solved	-
P23	51 / M	Retinitis pigmentosa	Peripheral visual field loss	6	0.4/0.4	Central retina preserved	✓	✓	✓	✓	✓	-	RPGR	Potentially solved	-
P24	30 / F	Bietti crystalline dystrophy	DVA	26	0.5/0.5	Diffuse hyperreflective crystals	✓	✓	-	-	-	-	CYP4V2	Solved	✓
P25	51 / M	Retinitis pigmentosa	Nyctalopia	30	0.4/0.8	Peripheral bone spicule pigmentations	✓	-	-	-	-	Behçet disease, uveitis, and cataract in the right eye	-	Unsolved	-
P26	42 / F	Usher syndrome	Nyctalopia	37	0.6/0.6	Peripheral bone spicule pigmentations	✓	✓	✓	✓	✓	HL	ARSG	Potentially solved	✓
P27	28 / F	Retinitis pigmentosa	Nyctalopia	7	-	Peripheral bone spicule pigmentations	✓	-	-	-	-	-	RPE65	Solved	-
P28	26 / F	Retinitis pigmentosa	DVA	5	0.7/0.8	Bone spicule pigmentations, optic disc pallor	✓	✓	-	-	-	Cataract	-	Unsolved	-
P29	37 / M	Enhanced S-cone syndrome	DVA	10	0.05/0.05	Bone spicule pigmentations, optic disc	✓	✓	✓	✓	✓	Cataract (age of onset 24), retinal detachment (age 27)	NR2E3	Solved	✓

P30	16 / F	Retinitis pigmentosa	Nyctalopia	14	0.1/0.2	✓	✓	-	-	Epilepsy, MR	MFSD8	Solved	-
P31	52 / M	Retinitis pigmentosa	Nyctalopia	35	1.0/1.0	✓	✓	✓	-	ERG: bilateral rod response unrecordable, cone response subnormal	-	Unsolved	-
P32	24 / M	Leber's congenital amaurosis	DVA	NA	HM/HM (at 24 age)	✓	✓	✓	✓	Slow amplitude nystagmus	RPE65	Solved	-
P33	49 / M	Retinitis pigmentosa	Nyctalopia	38	0.9/0.6	✓	✓	-	-	Bilateral cataract surgery	-	Unsolved	-
P34	71 / M	Retinitis pigmentosa	DVA	<10	p-p-	✓	✓	-	-	Bilateral cataract surgery	-	Unsolved	-

Abbreviations: AF: autofluorescence, BCVA: Best corrected visual acuity, DVA: decreased visual acuity, ERG: electroretinography, F: female, FAF: fundus autofluorescence, FCM: finger counting from () meter, HL: hearing loss, HM: hand motion, M: male, MR: mental retardation, NA: not available, OCT: optical coherence tomography, OCTA: optical coherence tomography angiography, p-p-: perception is present/projection is absent, SNHL: sensorineural hearing loss

^a: The Snellen test was used