

RESEARCH ARTICLE				Journal of Bio-X Research		
Table S1						
Summary of the probands' clinical diagnoses information						
Sample name	Gene	Gender	Age	Vision (left eye/right eye)	Disease history	Disease
19093	ABCA4	M	16	0.3/0.5	Sporadic	Recessive Stargardt disease
19179	ABCA4	F	38	0.1/0.1	Sporadic	Recessive Stargardt disease
19159	AHI1	M	25	0.2/0.3	Sporadic	Recessive Joubert syndrome
19169	AIP1L1	M	43	0.1/0.1	Sporadic	Cone or cone-rod dystrophy
19156	ALMS1	M	5	0.2/0.2	Sporadic	Recessive Alström syndrome
CDRP007	C2orf71	M	28	FC/BE	Sporadic	Recessive retinitis pigmentosa
19071	CA4	M	31	FC/BE	Autosomal dominant	Dominant retinitis pigmentosa
19141	CACNA1F	M	7	0.5/0.4	Sporadic	Congenital stationary night blindness
19141	CACNA1F	M	4	0.5/0.4	Sporadic	Cone or cone-rod dystrophy
19182	CDHR1	M	27	0.2/0.3	Autosomal recessive	Recessive cone-rod dystrophy
19182	CDHR1	M	15	0.2/0.3	Autosomal recessive	Cone or cone-rod dystrophy
CDRP008	CHM	F	20	0.1/0.07	Sporadic	Choroideremia
19175	CNGA1	F	46	0.07/0.2	Autosomal recessive	Recessive retinitis pigmentosa
19105	COL11A1	F	20	0.3/0.3	Sporadic	Dominant Stickler syndrome
19159	COL11A1	M	25	0.2/0.3	Sporadic	Dominant Stickler syndrome
19171	CRB1	F	44	0.5/0.6	Autosomal dominant	Dominant pigmented paravenous chorioretinal atrophy
CDRP003	CRB1	M	35	0.3/0.1	Sporadic	Dominant pigmented paravenous chorioretinal atrophy
19160	CRX	M	5	0.03/0.03	Sporadic	Dominant retinitis pigmentosa
19095	CYP4V2	M	31	0.04/BE	Sporadic	Recessive retinitis pigmentosa
19087	EYS	M	53	0.1/no LP	Sporadic	Recessive retinitis pigmentosa
19090	EYS	M	32	0.3/0.2	Sporadic	Recessive retinitis pigmentosa
19091	EYS	F	36	0.6/0.9	Sporadic	Recessive retinitis pigmentosa
19106	EYS	M	35	0.3/0.2	Sporadic	Recessive retinitis pigmentosa
19150	EYS	M	32	0.8/0.6	Sporadic	Recessive retinitis pigmentosa
XHRP004	EYS	F	25	FC/BE	Sporadic	Recessive retinitis pigmentosa
19096	HMCN1	M	35	0.3/0.1	Sporadic	Dominant macular dystrophy
19121	HMCN1	M	24	-/0.04	Sporadic	Dominant macular dystrophy
19140	HMCN1	M	6	0.1/0.2	Sporadic	Dominant macular dystrophy
19161	HMCN1	M	30	0.2/0.2	Sporadic	Dominant macular dystrophy
19177	HMCN1	M	25	0.07/0.1	Autosomal dominant	Dominant macular dystrophy
CDRP020	HMCN1	M	24	0.1/0.2	Sporadic	Dominant macular dystrophy
Avastin8	IMP2	M	25	0.2/0.2	Sporadic	Recessive retinitis pigmentosa
19109	KCNJ13	M	28	0.3/0.6	Sporadic	Dominant vitreoretinal degeneration
CDRP020	KIF11	M	25	0.1/0.4	Sporadic	Dominant microcephaly
19145	MERTK	F	20	0.3/0.3	Autosomal dominant	Recessive retinitis pigmentosa
19148	NPHP1	F	7	0.1/0.05	Sporadic	Recessive Senior-Loken syndrome
19148	NPHP4	F	7	0.1/0.05	Sporadic	Recessive Senior-Loken syndrome
19167	PDE6A	M	42	0.7/0.6	Sporadic	Recessive retinitis pigmentosa
19172	PDE6A	M	33	0.1/0.3	Autosomal recessive	Recessive retinitis pigmentosa
19151	PDE6B	F	45	0.2/0.2	Sporadic	Recessive retinitis pigmentosa
19083	PDE6B	M	32	0.02/0.04	Sporadic	Congenital stationary night blindness
19158	PDE6B	F	23	0.2/0.2	Sporadic	Congenital stationary night blindness
19098	PITPNM3	M	25	0.1/0.1	Sporadic	Cone or cone-rod dystrophy
19176	RHO	F	30	0.1/0.2	Autosomal dominant	Dominant retinitis pigmentosa
19185	RHO	M	52	FC/BE	Sporadic	Dominant retinitis pigmentosa
19120	RP1	M	29	0.6/0.6	Sporadic	Dominant retinitis pigmentosa
19139	RP1	M	35	FC/BE	Autosomal recessive	Recessive retinitis pigmentosa
CDRP011	RP1	F	23	0.4/0.6	Sporadic	Dominant retinitis pigmentosa
19129	RP1L1	F	28	0.7/0.5	Sporadic	Recessive retinitis pigmentosa
CDRP014	RPE65	F	30	0.2/0.03	Sporadic	Recessive retinitis pigmentosa
CDRP009	RPGR	F	8	0.3/0.03	X-linked	X-linked retinitis pigmentosa
19143	RS1	M	3	0.2/0.03	Sporadic	Other retinopathy, X-linked
19157	RS1	M	40	-/-	Sporadic	Other retinopathy, X-linked
19138	SNRNP200	M	11	0.3/0.3	Sporadic	Dominant retinitis pigmentosa
19153	TIMP3	M	42	0.6/0.7	Autosomal dominant	Dominant Sorsby's fundus dystrophy
19128	VCAN	M	43	0.1/0.7	Sporadic	Dominant Wagner disease and erosive vitreoretinopathy