

Multimodal structural disease progression of retinitis pigmentosa according to mode of inheritance

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Supplementary Table 1. Correlations between the repeated measurements from both independent graders for horizontal diameter, vertical diameter, and ring area of the hyperautofluorescent rings at visit 1 and 2.

	EZ line width		Horizontal diameter		Ring area	
Pearson correlation	Visit 1	Visit 2	Visit 1	Visit 2	Visit 1	Visit 2
r	0.999	0.993	0.966	0.997	0.997	0.999
P-value	< 0.001	< 0.001	< 0.001	< 0.001	< 0.001	< 0.001

EZ = ellipsoid zone.

Supplementary Table 2. Complete characterization of the disease-causing genetic variants of the patients along with length of follow-up and best-corrected visual acuity at both visit 1 and visit 2.

Patient ID	Disease	Gene	Variants			Length of Follow-up (yrs)	Visit 1 BCVA	Visit 2 BCVA
			cDNA Change	Protein Change	Zygosity			
1	adRP	<i>RHO</i>	c.404G>T	p.R135L	<i>hetero</i>	3.7	20/30; 20/25	20/40; 20/40
2	adRP	<i>RHO</i>	c.568G>A	p.D190N	<i>hetero</i>	7.1	20/25; HM	20/40; HM
3	adRP	<i>RHO</i>	c.266G>A	p.G89D	<i>hetero</i>	7.1	20/125; 20/25	20/150; 20/40
4	adRP	<i>RHO</i>	c.316G>A	p.G106R	<i>hetero</i>	5.4	20/40; 20/40	20/50; 20/50
5	adRP	<i>RHO</i>	c.800C>T	p.P267L	<i>hetero</i>	8.1	20/20; 20/25	20/40; 20/40
6	adRP	<i>RHO</i>	c.102G>A	p.T342M	<i>hetero</i>	7.5	20/50; 20/40	20/50; 20/60
7	adRP	<i>RHO</i>	c.328T>C	p.C110R	<i>hetero</i>	4.2	20/50; 20/40	20/200; LP
8	adRP	<i>RHO</i>	c.937-27_- 19delCCCTGACTC	n/a	<i>hetero</i>	5.6	20/20; 20/20	20/40; 20/40
9	adRP	<i>RHO</i>	c.568G>A	p.D190N	<i>hetero</i>	3.0	20/25; 20/20	20/40; 20/40
10	adRP	<i>RHO</i>	c.404_4005delinsGG >TT	p.R135L	<i>hetero</i>	1.7	20/300; 20/250	CF 3ft; 20/400
11	adRP	<i>RHO</i>	c.328T>C	p.C110R	<i>hetero</i>	2.8	20/30; 20/20	20/40; 20/25
12	adRP	<i>RHO</i>	c.50C>T	p.T17M	<i>hetero</i>	1.3	20/20; 20/20	20/30; 20/30
13	adRP	<i>RHO</i>	c.328T>C	p.C110R	<i>hetero</i>	2.8	20/20; 20/20	20/25; 20/25
14	adRP	<i>RP1</i>	c.2105_2108del	p.I702Tfs*10	<i>hetero</i>	1.1	20/20; 20/30	20/25; 20/30
15	adRP	<i>RP1</i>	c.5107delT	n/a	<i>hetero</i>	2.4	20/20; 20/20	20/25; 20/30
16	adRP	<i>RP1</i>	c.2105_2108del	p.I702Tfs*10	<i>hetero</i>	1.1	20/20; 20/20	20/25; 20/25
17	adRP	<i>RP1</i>	c.2285_2289delTAA AT	p.L762Yfs*17	<i>hetero</i>	6.5	20/30; 20/25	20/40; 20/30
18	adRP	<i>RP1</i>	c.2479G>C	p.E827Q	<i>hetero</i>	7.3	20/30; 20/30	20/40; 20/40
19	adRP	<i>RP1</i>	c.2029C>T	p.R677*	<i>hetero</i>	5.0	20/20; 20/20	20/25; 20/25
20	adRP	<i>RP1</i>	c.2285_2289delTAA AT	p.L762Yfs*17	<i>hetero</i>	3.7	20/25; 20/40	20/40; 20/50
21	adRP	<i>RP1</i>	c.2285_2289delTAA AT	p.L762Yfs*17	<i>hetero</i>	4.6	20/25; 20/30	20/30; 20/40

22	adRP	<i>RP1</i>	c.1437G>T	p.M479I	<i>hetero</i>	1.9	20/20; 20/25	20/20; 20/25
23	adRP	<i>PRPF31</i>	c.383T>A	p.L128*	<i>hetero</i>	4.6	20/25; 20/20	20/40; 20/25
24	adRP	<i>PRPF31</i>	c.420+6C>T	n/a	<i>hetero</i>	2.2	20/25; 20/25	20/25; 20/25
25	adRP	<i>PRPF31</i>	c.1222C>T	p.R408W	<i>hetero</i>	2.5	20/80; 20/150	20/150; 20/400
26	adRP	<i>PRPF31</i>	c.1066G>A	p.G356S	<i>hetero</i>	1.3	20/25; 20/50	20/50; 20/50
27	adRP	<i>KLHL7</i>	c.433A>G	p.N145D	<i>hetero</i>	6.9	20/30; 20/30	20/40; 20/40
28	adRP	<i>KLHL7</i>	c.472T>C	p.C158R	<i>hetero</i>	2.8	20/20; 20/25	20/30; 20/40
29	adRP	<i>KLHL7</i>	c.433A>G	p.N145Y	<i>hetero</i>	1.8	20/30; 20/30	20/30; 20/30
30	adRP	<i>IMPDH1</i>	c.962C>T	p.R321V	<i>hetero</i>	2.5	20/50; 20/70	20/60; 20/70
31	adRP	<i>IMPDH1</i>	c.1280C>T	p.P427L	<i>hetero</i>	1.1	20/40; 20/30	20/40; 20/60
32	adRP	<i>GUCA1B</i>	c.153_155delTGA	p.D51del	<i>hetero</i>	5.2	20/20; 20/20	20/40; 20/40
33	adRP	<i>NRL</i>	c.149C>T	p.S50L	<i>hetero</i>	6.1	20/25; 20/30	20/40; 20/40
34	adRP	<i>PRPF8</i>	c.6991delG	p.E2331fs	<i>hetero</i>	1.9	20/20; 20/30	20/25; 20/40
35	adRP	<i>PRPH2</i>	c.652T>C	p.S218P	<i>hetero</i>	1.8	20/25; 20/40	20/40; 20/50
36	arRP	<i>USH2A</i>	c.9371+A>G c.11048-2A>G	n/a n/a	<i>hetero</i> <i>hetero</i>	1.0	20/20; 20/20	20/20; 20/20
37	arRP	<i>USH2A</i>	c.13378A>T c.15178T>C	p.I4460L p.S5060P	<i>hetero</i> <i>hetero</i>	6.6	20/125; 20/40	20/400; 20/60
38	arRP	<i>USH2A</i>	c.10073G>A c.920_923dupGCCA	p.C3358Y p.H308Qfs	<i>hetero</i> <i>hetero</i>	4.0	20/20; 20/20	20/30; 20/30
39	arRP	<i>USH2A</i>	c.12575G>A c.7863delA	p.R4192H p.2623Hfs*18	<i>hetero</i> <i>hetero</i>	3.9	20/50; 20/25	20/50; 20/30
40	arRP	<i>USH2A</i>	c.10073G>A c.9110G>A	p.C3358Y p.R3037H	<i>hetero</i> <i>hetero</i>	3.9	20/25; 20/25	20/30; 20/30
41	arRP	<i>USH2A</i>	c.12575G>A c.12575G>A	p.R4192H p.R4192H	<i>hetero</i> <i>hetero</i>	2.1	20/30; 20/25	20/50; 20/50
42	arRP	<i>USH2A</i>	c.9676C>T c.1478A>G	p.R3226* p.Y493C	<i>hetero</i> <i>hetero</i>	3.5	20/20; 20/20	20/30; 20/30
43	arRP	<i>USH2A</i>	c.12575G>A c.2802T>G	p.R4192H p.C934W	<i>hetero</i> <i>hetero</i>	2.6	20/20; 20/20	20/25; 20/25
44	arRP	<i>USH2A</i>	c.13223T>C c.13231C>G; c.4251+1G>A	p.V4408A p.L4411V; n/a	<i>hetero</i> <i>hetero</i> <i>hetero</i>	2.4	20/30; 20/50	20/40; 20/60
45	arRP	<i>USH2A</i>	c.2276G>T	p.C759F	<i>homo</i>	1.4	20/20; 20/20	20/40; 20/30

46	arRP	<i>USH2A</i>	c.12575G>A c.7595-2144A>G	p.R4192H n/a	<i>hetero</i> <i>hetero</i>	7.3	20/20; 20/20	20/30; 20/30
47	arRP	<i>USH2A</i>	c.2522C>A c.5975A>G	p.S841Y p.Y1992C	<i>hetero</i> <i>hetero</i>	2.9	20/20; 20/50	20/70; 20/100
48	arRP/USH2	<i>USH2A</i>	c.3713C>G c.9459C>A	p.T1238R p.C3153*	<i>hetero</i> <i>hetero</i>	2.9	20/40; 20/40	20/60; 20/60
49	arRP/USH2	<i>USH2A</i>	c.13577G>A c.5775A>G	p.R4526Q p.T1925T	<i>hetero</i> <i>hetero</i>	4.9	20/20; 20/20	20/30; 20/30
50	arRP/USH2	<i>USH2A</i>	c.10712C>T c.4711G>C	p.T3571M p.A1571P	<i>hetero</i> <i>hetero</i>	1.8	20/30; 20/50	20/50; 20/60
51	arRP	<i>PDE6β</i>	c.1927_1967del41	n/a	<i>homo</i>	1.8	20/20; 20/40	20/30; 20/50
52	arRP	<i>PDE6β</i>	c.1923_1969ins6del47	p.T641Tfs*31	<i>homo</i>	1.1	20/25; 20/25	20/25; 20/25
53	arRP	<i>PDE6β</i>	c.1923_1969ins6del47	p.T641Tfs*31	<i>homo</i>	3.2	20/20; 20/20	20/40; 20/40
54	arRP	<i>PDE6β</i>	c.1540delC	p.L514Wfs	<i>homo</i>	4.5	20/50; 20/40	20/60; 20/50
55	arRP	<i>PDE6β</i>	c.1488delC c.1669C>T	p.T497Pfs*78 p.H557Y	<i>hetero</i> <i>hetero</i>	2.9	20/60; 20/50	20/60; 20/60
56	arRP	<i>PDE6β</i>	c.1655G>A	p.R552Q	<i>homo</i>	5.3	20/100; 20/100	20/200; 20/200
57	arRP	<i>EYS</i>	c.6528C>A	p.Y2176*	<i>homo</i>	3.5	20/40; 20/50	20/50; 20/70
58	arRP	<i>EYS</i>	c.8111T>G c.9316_9336delCCA ATTTTGTGGCAA AATT	p.L2740* p.T3106Kfs*13	<i>hetero</i> <i>hetero</i>	2.8	20/50; 20/60	20/70; 20/70
59	arRP	<i>EYS</i>	c.4120C>T	p.R1374*	<i>homo</i>	1.3	20/20; 20/25	20/20; 20/30
60	arRP	<i>EYS</i>	c.1645G>T c.2992+1G>A	p.E549* n/a	<i>hetero</i> <i>hetero</i>	3.9	20/25; 20/25	20/50; 20/50
61	arRP	<i>EYS</i>	c.4402G>C c.3250A>C	p.D1468H p.T1084P	<i>homo</i> <i>homo</i>	2.0	20/200; 20/400	20/800; 20/800
62	arRP	<i>PDE6α</i>	c.304C>A c.769C>T	p.R102S p.R257*	<i>hetero</i> <i>hetero</i>	1.8	20/150; 20/150	20/200; 20/200
63	arRP	<i>PDE6α</i>	c.304C>A c.908C>G	p.R102S p.S303C	<i>hetero</i> <i>hetero</i>	1.6	20/50; 20/50	20/100; 20/100
64	arRP	<i>PDE6α</i>	c.304C>A c.908C>G	p.R102S p.S303C	<i>hetero</i> <i>hetero</i>	2.9	20/40; 20/50	20/50; 20/60
65	arRP	<i>PDE6α</i>	c.1705C>A c.2263C>T	p.Q569K p.Q755*	<i>hetero</i> <i>hetero</i>	1.0	20/25; 20/25	20/30; 20/30
66	arRP	<i>CDHR1</i>	c.2522_2528delTCT CTGA	p.I841Sfs*119	<i>homo</i>	1.7	20/40; 20/40	20/60; 20/60
67	arRP	<i>CDHR1</i>	c.1463delG	p.G488fs	<i>homo</i>	2.1	20/50; 20/50	20/150; 20/150
68	arRP	<i>CNGB1</i>	c.3150delG	p.F105Lfs*12	<i>homo</i>	4.9	20/25; 20/25	20/40; 20/40
69	arRP	<i>CNGB1</i>	c.1896C>A c.3150delG	p.C632* p.G1050Gfs*13	<i>hetero</i> <i>hetero</i>	1.4	20/40; 20/40	20/50; 20/50

70	arRP	<i>DHDDS</i>	c.124A>G	p.K42E	<i>homo</i>	3.9	20/25; 20/40	20/30; 20/40
71	arRP	<i>DHDDS</i>	c.124A>G	p.K42E	<i>homo</i>	1.1	20/30; 20/50	20/30; 20/70
72	arRP	<i>KIZ</i>	c.119_122delAACT	p.K10Ifs*14	<i>homo</i>	2.1	20/30; 20/80	20/50; 20/100
73	arRP	<i>KIZ</i>	c.226C>T	p.R76*	<i>homo</i>	2.2	20/40; 20/50	20/40; 20/50
74	arRP	<i>MAK</i>	c.1291A>T	p.K431*	<i>homo</i>	2.3	20/20; 20/25	20/30; 20/30
75	arRP	<i>MAK</i>	c.1297_1298ins	p.K433Rfs*31	<i>homo</i>	1.2	20/20; 20/25	20/25; 20/30
76	arRP	<i>MERTK</i>	c.1133C>T	p.T378M	<i>homo</i>	1.4	20/25; 20/25	20/30; 20/30
77	arRP	<i>MERTK</i>	c.2189+1G>T	n/a	<i>homo</i>	2.1	20/40; 20/40	20/50; 20/50
78	arRP/USH1	<i>MYO7A</i>	c.4544_4551delAGA TCATGinsCA	p.E1515_M1517 delinsA	<i>homo</i>	1.9	20/60; 20/40	20/80; 20/40
79	arRP/USH1	<i>MYO7A</i>	c.634C>T c.999T>G	p.R212C p.Y333*	<i>hetero</i> <i>hetero</i>	2.2	20/40; 20/50	20/60; 20/60
80	arRP	<i>C21ORF2</i>	c.670G>T	p.A224S	<i>homo</i>	4.0	20/50; 20/60	20/80; 20/150
81	arRP	<i>CERKL</i>	c.847C>T	p.R283*	<i>homo</i>	1.6	20/50; 20/60	20/50; 20/60
82	arRP	<i>FAM161A</i>	c.1321dupC	p.H441Pfs*15	<i>homo</i>	2.2	20/25; 20/40	20/30; 20/40
83	arRP	<i>GPR98</i>	c.9430G>T c.10769+2T>A	p.E3144* n/a	<i>hetero</i> <i>hetero</i>	2.2	20/40; 20/40	20/50; 20/50
84	arRP	<i>IFT140</i>	c.2788C>T c.3141+13C>T	p.H930Y n/a	<i>hetero</i> <i>hetero</i>	2.4	20/25; 20/80	20/40; 20/100
85	arRP	<i>NPHP1</i>	c.1078C>G c.1434+9G>A	p.L360V n/a	<i>hetero</i> <i>hetero</i>	7.4	20/50; 20/60	20/200; 20/200
86	arRP	<i>REEP6</i>	c.295G>A	p.E99K	<i>homo</i>	1.7	20/20; 20/20	20/30; 20/25
87	arRP	<i>SPATA7</i>	c.1100A>G c.1101_1102delTC	p.Y367C p.L368Efs*4	<i>hetero</i> <i>hetero</i>	3.5	20/25; 20/50	20/50; 20/100
88	arRP	<i>TULP1</i>	c.349G>A	p.E117K	<i>homo</i>	3.5	20/80; 20/50	20/100; 20/60
89	XLRP	<i>RPGR</i>	c.202G>A	p.G68R	<i>hemi</i>	5.8	20/20; 20/30	20/25; 20/40
90	XLRP	<i>RPGR</i>	c.2194del	p.E732Rfs*83	<i>hemi</i>	3.2	20/40; 20/40	20/40; 20/50
91	XLRP	<i>RPGR</i>	c.2752G>T	p.G918*	<i>hemi</i>	2.5	HM; 20/40	HM; 20/40
92	XLRP	<i>RPGR</i>	c.1059_1059+2delG GT	n/a	<i>hemi</i>	1.3	20/25; 20/25	20/30; 20/30
93	XLRP	<i>RPGR</i>	c.1307G>A	p.G436D	<i>hemi</i>	1.4	20/20; 20/30	20/25; 20/40
94	XLRP	<i>RPGR</i>	c.2752G>T	p.G918*	<i>hemi</i>	1.2	20/50; 20/30	20/60; 20/40

95	XLRP	<i>RPGR</i>	c.2997_2998delGG	p.E1000Gfs*78	<i>hemi</i>	1.6	20/70; 20/40	20/70; 20/50
96	XLRP	<i>RPGR</i>	c.2236_2237delGA	p.E746Rfs*23	<i>hemi</i>	1.1	20/80; 20/70	20/80; 20/70

adRP = autosomal dominant retinitis pigmentosa; arRP = autosomal recessive retinitis pigmentosa; XLRP = x-linked retinitis pigmentosa; BCVA = best-corrected visual acuity; CF = counting fingers; hemi = hemizygous; hetero = heterozygous; homo = homozygous; HM = hand motion; yrs = years.