ID	Result	Gene	Variant c.	Variant p.	Diagnosis	Same kindred	Homozygous
1	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome	1	
1	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
2	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
2	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
3	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	RP		
3	Positive	*USH2A*	c.956G>A	(p.Cys319Tyr)			
4	Positive	*COL18A1*	c.1459C>T	(p.Arg487*)	Knobloch	1	
4	Positive	*COL18A1*	c.929-2A>G	NA			
5	Positive	*COL18A1*	c.1459C>T	(p.Arg487*)	Knobloch	1	
5	Positive	*COL18A1*	c.929-2A>G	NA			
6	Positive	*COL18A1*	c.1459C>T	(p.Arg487*)	Knobloch		
6	Positive	*COL18A1*	c.929-2A>G	NA			
7	Positive	*USH2A*	c.12575G>A	(p.Arg4192His)	RP		
7	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
8	Positive	*COL2A1*	c.3574C>T	(p.Arg1192*)	Stickler	1	
9	Positive	*COL2A2*	c.3574C>T	(p.Arg1192*)	Stickler		
10	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)	RP		
10	Positive	*USH2A*	c.5329C>T	(p.Arg1777Trp)			
11	Positive	*TYR*	c.996G>A	(p.Met332Ile)	OCA		
11	Positive	*TYR*	c.271T>C	(p.Cys91Arg)			
12	Positive	*RS1*	c.577C>T	(p.Pro193Ser)	XLRS		
13	Positive	*COL2A1*	c.1995+1G>T	NA	Stickler		
14	Positive	*PRPH2*	c.774C>G	(p.Tyr258*)	MD		
15	Positive	*LRP5*	c.2555C>T	(p.Thr852Met)	PHPV	1	
15	Positive	*LRP5*	c.3004C>T	(p.Arg1002*)			
16	Positive	*LRP5*	c.2555C>T	(p.Thr852Met)	PHPV		
16	Positive	*LRP5*	c.3004C>T	(p.Arg1002*)			
17	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)	RP		
17	Positive	*USH2A*	c.7454T>A	(p.Leu2485*)			
18	Positive	*ABCA4*	c.3210_3211dup	(p.Ser1071Cysfs*14)	MD		
18	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)			
19	Positive	*RS1*	c.626G>A	(p.Arg209His)	MD	1	
20	Positive	*RS1*	c.626G>A	(p.Arg209His)	XLRS		
21	Positive	*USH2A*	c.1724G>A	(p.Cys575Tyr)	RP		
21	Positive	*USH2A*	c.2035G>T	(p.Gly679*)			
22	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
23	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
24	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
25	Positive	*AHI1*	c.2742del	(p.Leu915Cysfs*64)	RP		
25	Positive	*AHI1*	c.3196C>T	(p.Arg1066*)			
26	Positive	*FZD4*	c.1282_1285del	(p.Asp428Serfs*2)	FEVR		
27	Positive	*CHM*	561T>A	(p.Cys187*	CHM		
28	Positive	*GPR143*	Deletion entire coding sequence	NA	OCA		
29	Positive	*RPGR*	c.2405_2406del	(p.Glu802Glyfs*32	RP	1	
30	Positive	*RPGR*	c.2405_2406del	(p.Glu802Glyfs*32	RP	1	
31	Positive	*RPGR*	c.2405_2406del	(p.Glu802Glyfs*32	RP		
32	Positive	*RS1*	c.214G>A	(p.Glu72Lys)	XLRS		
33	Positive	*ABCA4*	c.5413A>G	(p.Asn1805Asp)	Stargardt		
33	Positive	*ABCA4*	c.5512C>G	(p.His1838Asp)			
33	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)			
34	Positive	*ABCA4*	c.4919G>A	(p.Arg1640Gln)	Stargardt		
34	Positive	*ABCA4*	c.5530G>T	(p.Gly1844Cys)			
35	Positive	*RPGR*	c.356T>C	(p.Leu119Ser)	RP	1	
36	Positive	*RPGR*	c.356T>C	(p.Leu119Ser)	RP		
37	Positive	*CHM*	Deletion entire coding sequence	NA	CHM		
38	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)	RP		
38	Positive	*USH2A*	c.9119G>A	(p.Trp3040*)			
39	Positive	*USH2A*	c.12067-2A>G	NA	RP		
39	Positive	*USH2A*	c.1751G>T	(p.Cys584Phe			
40	Positive	*USH2A*	c.10759C>T	(p.Gln3587*)	RP		
40	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)			
41	Positive	*CRB1*	c.2234C>T	(p.Thr745Met)	EOSRD	1	
41	Positive	*CRB1*	c.2290C>T	(p.Arg764Cys)			
42	Positive	*CRB1*	c.2234C>T	(p.Thr745Met)	EOSRD		
42	Positive	*CRB1*	c.2290C>T	(p.Arg764Cys)			
43	Positive	*CDHR1*	c.1219C>T	(p.Arg407*)	RP		1
44	Positive	*PRPF31*	c.371_375del	(p.Glu124Glyfs*28)	RP		
45	Positive	*USH2A*	c.9119G>A	(p.Trp3040*)	Usher syndrome		
45	Positive	*USH2A*	Deletion exons 69-70	NA			
46	Positive	*RPGR*	C1345C>T	(p.Arg449*)	RP		
47	Positive	*USH2A*	c.9119G>A	(p.Trp3040*)	Usher syndrome		

47	Positive	*USH2A*	c.1417G>A	(p.Trp4725*)			
48	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
49	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
50	Positive	*CNGB1*	c.2030G>A	(p.Arg677His)	RP		
50	Positive	*CNGB1*	c.382-3C>G	NA			
51	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP	1	
52	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP	1	
53	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
54	Positive	*RDH12*	c.806_810del	(p.Ala269Glyfs*2)	EOSRD		
54	Positive	*RDH12*	c.152T>A	(p.Ile51Asn)			
55	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP	1	
56	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
57	Positive	*CDHR1*	c.1219C>T	(p.Arg407*)	RP		1
58	Positive	*RS1*	c.590G>A	(p.Arg197His)	XLRS		
59	Positive	*RS1*	c.590G>A	(p.Arg197His)	XLRS		
60	Positive	*CERKL*	c.847C>T	(p.Arg283*)	RP		1
61	Positive	*USH2A*	c.11864G>A	(p.Trp3955*)	RP		
61	Positive	*USH2A*	c.12575G>A	(p.Arg4192His)			
62	Positive	*PRPF31*	c.23T>G	(p.Leu8*)	RP		
63	Positive	*RDH12*	c.278T>C	(p.Leu93Pro	EOSRD		
63	Positive	*RDH12*	c.464C>T	(p.Thr155Ile)			
64	Positive	*ABCA4*	c.3386G>T	(p.Arg1129Leu)	Stargardt		
64	Positive	*ABCA4*	c.5461-10T>C	NA			
64	Positive	*ABCA4*	c.6718A>G	(p.Thr2240Ala)			
65	Positive	*PDE6A*	Deletion Exon 6	NA	EOSRD		
65	Positive	*PDE6A*	Deletion Exon 9	NA			
66	Positive	*BBS1*	c.217G>T	(p.Gly73*)	RP	1	1
67	Positive	*BBS1*	c.217G>T	(p.Gly73*)	RP		1
68	Positive	*MAK*	c.1356_1357del	(p.Glu454Lysfs*13)	RP		
68	Positive	*MAK*	c.1167del	(p.His389Glnfs*76)			
69	Positive	*RPGR*	c.2819_2837dup	(p.Glu947Argfs*138)	RP		
70	Positive	*RPGRIP1*	c.2910_2911del	(p.Pro971*)	RP		1
71	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)	Usher syndrome		
71	Positive	*USH2A*	Deletion Exons 20-21	NA			
72	Positive	*RPGR*	c.2819_2837dup	(p.Glu947Argfs*138)	RP		
73	Positive	*TTC8*	c.991C>T	(p.Gln331*)	BBS		1
74	Positive	*MYO7A*	c.3508G>A	p.Glu1170Lys	Usher syndrome		
74	Positive	*MYO7A*	c.3764del	(p.Lys1255Argfs*8)			
74	Positive	*MYO7A*	c.338T>C	(p.Ile113Thr)			
75	Positive	*ARL6*	c.506del	(p.Gly169Alafs*6)	RP	1	
75	Positive	*ARL6*	c.344A>G	(p.His115Arg)			
76	Positive	*ARL6*	c.506del	(p.Gly169Alafs*6)	RP		
76	Positive	*ARL6*	c.344A>G	(p.His115Arg)			
77	Positive	*CRB1*	c.4168C>T	(p.Arg1390*)	RP		1
78	Positive	*RP1*	c.368_369dup	(p.Pro124Alafs*20)	RP		
78	Positive	*RP1*	c.532C>T	(p.Gln178*)			
79	Positive	*PRPF31*	c.527+1G>A	NA	RP		
80	Positive	*PRPF31*	c.527+1G>A	NA	RP	1	
81	Positive	*PRPF31*	c.527+1G>A	NA	RP		
82	Positive	*PRPF31*	c.527+1G>A	NA	RP		
83	Positive	*PRPF31*	c.527+1G>A	NA	RP	1	
84	Positive	*PRPF31*	c.527+1G>A	NA	RP		
85	Positive	*BBS7*	c.785_786del	(p.Asp262Glyfs*8)	BBS		
85	Positive	*BBS7*	c.947G>T	(p.Gly316Val)			
86	Positive	*CDH23*	c.336del	(p.Val113*)	Usher syndrome		
86	Positive	*CDH23*	c.2801C>T	(p.Pro934Leu)			
87	Positive	*TYR*	c.1217C>T	(p.Pro406Leu)	OCA		
87	Positive	*TYR*	c.896G>A	(p.Arg299His)			
87	Positive	*TYR*	c.646T>A	(p.Leu216Met)			
88	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
89	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
90	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
91	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
92	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
93	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
94	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
95	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
96	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
97	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
98	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
99	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
100	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	

101	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
102	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
103	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
104	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
105	Positive	*RDH12*	c.295C>A	(p.Leu99Ile)	EOSRD		1
106	Positive	*ABCA4*	c.5461-10T>C	NA	Stargardt		
106	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)			
107	Positive	*PDE6A*	c.1955_1974dup	(p.Ile659Valfs*10)	RP	1	
107	Positive	*PDE6A*	c.1538del	(p.Leu513Glnfs*7)			
108	Positive	*PDE6A*	c.1955_1974dup	(p.Ile659Valfs*10)	RP		
108	Positive	*PDE6A*	c.1538del	(p.Leu513Glnfs*7)			
109	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP	1	
110	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
111	Positive	*EYS*	Deletion Exons13-14	NA	RP		
111	Positive	*EYS*	c.5450G>A	(p.Trp1817*)			
112	Positive	*EYS*	c.5450G>A	(p.Trp1817*)	RP		
112	Positive	*EYS*	c.6794del	(p.Pro2265Glnfs*46)			
113	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
114	Positive	*PRPF8*	c.6928A>G	(p.Arg2310Gly)	RP		
115	Positive	*AGBL5*	c.421dup	(p.His141Profs*23)	RP		1
116	Positive	*CHM*	c.715C>T	(p.Arg239*)	CHM		
117	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
117	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
118	Positive	*CWC27*	c.1101T>G	(p.Tyr367*)	RP	1	
118	Positive	*CWC27*	c.495G>A	(p.Glu165Glu)			
119	Positive	*CWC27*	c.1101T>G	(p.Tyr367*)	RP		
119	Positive	*CWC27*	c.495G>A	(p.Glu165Glu)			
120	Positive	*NR2E3*	c.119-2A>C	NA	RP	1	
120	Positive	*NR2E3*	c.731del	(p.Leu244Argfs*7)			
121	Positive	*NR2E3*	c.119-2A>C	NA	RP		
121	Positive	*NR2E3*	c.731del	(p.Leu244Argfs*7)			
122	Positive	*USH2A*	c.7595-2144A>G	NA	Usher syndrome		
122	Positive	*USH2A*	c.7932G>A	(p.Trp2644*)			
123	Positive	*RP2*	c.465_468dup	(p.Phe157Serfs*18)	RP		
124	Positive	*ABCA4*	c.3617del	(p.Asn1206Metfs*3)	Stargardt		
124	Positive	*ABCA4*	c.302+4A>G	NA			
125	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP	1	
126	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
127	Positive	*COL2A1*	c.233dup	(p.Glu79*)	Stickler	1	
128	Positive	*COL2A1*	c.233dup	(p.Glu79*)	Stickler		
129	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)	Stargardt		
129	Positive	*ABCA4*	c.302+4A>G	NA			
130	Positive	*MYO7A*	c.6228_6232del	(p.Asp2076Glufs*50)	Usher syndrome		
130	Positive	*MYO7A*	c.274del	(p.His92Thrfs*14)			
131	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)	Stargardt		
131	Positive	*ABCA4*	c.1919C>G	(p.Pro640Arg)			
132	Positive	*TUBGCP4*	c.1196C>A	(p.Ser399*)	Chorioretinopaty		
132	Positive	*TUBGCP4*	c.1749G>T	(p.Leu582Leu)			
133	Positive	*PRPH2*	c.634A>G	(p.Ser212Gly)	RP	1	
134	Positive	*PRPH2*	c.634A>G	(p.Ser212Gly)	RP		
135	Positive	*CNGB1*	c.382-3C>G	NA	ACHM		1
136	Positive	*CRB1*	c.1172-2A>G	NA	RP		
136	Positive	*CRB1*	c.2784T>G	(p.Cys928Trp)			
137	Positive	*RPGR*	c.1872_1873del	(p.Glu624Aspfs*5)	RP		
138	Positive	*ABCA4*	c.319C>T	(p.Arg107*)	Stargardt		
138	Positive	*ABCA4*	c.3386G>T	(p.Arg1129Leu)			
138	Positive	*ABCA4*	c.6718A>G	(p.Thr2240Ala)			
139	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
140	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
141	Positive	*CRB1*	c.2843G>A	(p.Cys948Tyr)	RP	1	1
142	Positive	*CRB1*	c.2843G>A	(p.Cys948Tyr)	RP		1
143	Positive	*CDHR1*	c.1720C>G	(p.Pro574Ala)	RP		
143	Positive	*CDHR1*	c.1956del	(p.Trp652Cysfs*3)			
144	Positive	*FAM161A*	c.1003C>T	(p.Arg335*)	RP		1
145	Positive	*ABCA4*	c.1958G>A	(p.Arg653His)	Stargardt		
145	Positive	*ABCA4*	c.5714+5G>A	NA			
146	Positive	*USH2A*	c.100C>T	(p.Arg34*)	Usher syndrome		
146	Positive	*USH2A*	c.1841-2A>G	NA			
147	Positive	*CERKL*	c.847C>T	(p.Arg283*)	RP		1
148	Positive	*CRB1*	c.2291G>A	(p.Arg764His)	RP		1
149	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)	RP		
149	Positive	*USH2A*	c.232T>G	(p.Phe78Val)			

150	Positive	*RP2*	Deletion Exons 1-3	NA	RP	1	
151	Positive	*RP2*	Deletion Exons 1-3	NA	RP	1	
152	Positive	*RP2*	Deletion Exons 1-3	NA	RP		
153	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
154	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
155	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
156	Positive	*PDE6A*	c.1117G>T	(p.Glu373*)	RP		
156	Positive	*PDE6A*	c.2135+1G>T	NA			
157	Positive	*ABCA4*	c.6122G>A	(p.Gly2041Asp)	Stargardt		
157	Positive	*ABCA4*	c.614G>A	(p.Cys205Tyr)			
158	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
158	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
159	Positive	*PRPF31*	c.795del	(p.Ser266Glnfs*55)	RP		
160	Positive	*CDH23*	c.7832_7833del	(p.Phe2611Cysfs*31)	Usher syndrome		1
161	Positive	*CHM*	c.1066A>T	(p.Lys356*)	CHM	1	
162	Positive	*CHM*	c.1066A>T	(p.Lys356*)	CHM		
163	Positive	*PRPF31*	c.455del	(p.Asn152Metfs*46)	RP		
164	Positive	*RHO*	c.512C>A	(p.Pro171Gln)	RP		
165	Positive	*FAM161A*	Deletion Exon 2-7	NA	RP		1
166	Positive	*PRPF31*	c.749dup	(p.Met250Ilefs*29)	RP		
167	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
168	Positive	*RHO*	c.1040C>T	(p.Pro347Leu)	RP		
169	Positive	*USH2A*	c.10712C>T	(p.Thr3571Met)	RP	1	
169	Positive	*USH2A*	c.8834G>A	(p.Trp2945*)			
170	Positive	*USH2A*	c.10712C>T	(p.Thr3571Met)	RP		
170	Positive	*USH2A*	c.8834G>A	(p.Trp2945*)			
171	Positive	*RPGR*	c.2412_2418del	(p.Gly805Lysfs*8)	RP		
172	Positive	*EYS*	c.5928-2A>G	NA	RP		
172	Positive	*EYS*	c.8598del	(p.Gly2867Valfs*5)			
173	Positive	*EYS*	c.5928-2A>G	NA	RP	1	
173	Positive	*EYS*	c.8598del	(p.Gly2867Valfs*5)			
174	Positive	*RHO*	c.1040C>T	(p.Pro347Leu)	RP	1	
175	Positive	*RHO*	c.1040C>T	(p.Pro347Leu)	RP	1	
176	Positive	*RHO*	c.512C>A	(p.Pro171Gln)	RP		
177	Positive	*CHM*	c.546T>A	(p.Cys182*)	CHM		
178	Positive	*PROM1*	c.1956T>G	(p.Tyr652*)	RP		1
179	Positive	*OPA1*	c.635_636del	(p.Lys212Argfs*4)	OA	1	
180	Positive	*OPA1*	c.635_636del	(p.Lys212Argfs*4)	OA		
181	Positive	*CNGB3*	c.1148del	(p.Thr383Ilefs*13)	ACHM		1
182	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)	RP		
182	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
182	Positive	*USH2A*	c.14257G>A	(p.Val4753Ile)			
183	Positive	*NMNAT1*	Deletion Exons 4-5	NA	EOSRD		
183	Positive	*NMNAT1*	c.634G>A	(p.Val212Met)			
184	Positive	*RPGR*	c.2412_2418del	(p.Gly805Lysfs*8)	RP		
185	Positive	*ARL6*	c.506del	(p.Gly169Alafs*6)	RP		
185	Positive	*ARL6*	c.364C>T	(p.Arg122*)			
186	Positive	*RPGR*	c.2412_2418del	(p.Gly805Lysfs*8)	RP		
187	Positive	*RHO*	c.1040C>T	(p.Pro347Leu)	RP		
188	Positive	*PROM1*	c.1354dup	(p.Tyr452Leufs*13)	RP		
188	Positive	*PROM1*	c.1414C>T	(p.Arg472*)			
189	Positive	*USH2A*	c.10197C>A	(p.Cys3399*)	Usher syndrome		
189	Positive	*USH2A*	c.10585+3A>G	NA			
190	Positive	*RHO*	c.1040C>T	(p.Pro347Leu)	RP		
191	Positive	*RP1*	c.2029C>T	(p.Arg677*)	RP		
192	Positive	*RP1*	c.2029C>T	(p.Arg677*)	RP		
193	Positive	*PRPH2*	c.410G>A	(p.Gly137Asp)	RP		
194	Positive	*USH2A*	c.11864G>A	(p.Trp3955*)	Usher syndrome	1	
194	Positive	*USH2A*	c.14225_14232dup	(p.Val4745Argfs*4)			
195	Positive	*USH2A*	c.11864G>A	(p.Trp3955*)	Usher syndrome		
195	Positive	*USH2A*	c.14225_14232dup	(p.Val4745Argfs*4)			
196	Positive	*PRPF31*	c.901_919del	(p.Leu301Valfs*14)	MD		
197	Positive	*CERKL*	c.847C>T	(p.Arg283*)	RP		1
198	Positive	*RPGR*	c.2412_2418del	(p.Gly805Lysfs*8)	RP	1	
199	Positive	*RPGR*	c.2412_2418del	(p.Gly805Lysfs*8)	RP		
200	Positive	*RHO*	c.889A>C	(p.Ser297Arg)	RP		
201	Positive	*RPGR*	c.3348del	(p.Glu1117Serfs*14)	CORD		
202	Positive	*RHO*	c.403C>T	(p.Arg135Trp)	RP		
203	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
203	Positive	*USH2A*	c.14134-3169A>G	NA			
204	Positive	*USH2A*	c.10073G>A	(p.Cys3358Tyr)	RP		
204	Positive	*USH2A*	c.9441G>A	(p.Trp3147*)			

205	Positive	*BBS4*	c.341del	(p.Leu114Trpfs*28)	RP	1	
205	Positive	*BBS4*	c.777_778del	(p.Tyr259*)			
206	Positive	*BBS4*	c.341del	(p.Leu114Trpfs*28)	RP	1	
206	Positive	*BBS4*	c.777_778del	(p.Tyr259*)			
207	Positive	*BBS4*	c.341del	(p.Leu114Trpfs*28)	RP		
207	Positive	*BBS4*	c.777_778del	(p.Tyr259*)			
208	Positive	*RP2*	c.353G>A	(p.Arg118His)	RP		
209	Positive	*ADGRV1*	c.11563G>T	(p.Glu3855*)	Usher syndrome		
209	Positive	*ADGRV1*	c.18261del	(p.Gln6088Serfs*20)			
210	Positive	*PRPF31*	c.523C>T	(p.Gln175*)	MD		
211	Positive	*CRB1*	c.2171A>G	(p.Tyr724Cys)	EOSRD		
211	Positive	*CRB1*	c.3708_3709dup	(p.Ser1237Phefs*46)			
212	Positive	*USH2A*	c.11713C>T	(p.Arg3905Cys)	RP		
212	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)			
212	Positive	*USH2A*	c.14257G>A	(p.Val4753Ile)			
213	Positive	*RHO*	c.251T>C	(p.Leu84Pro)	RP	1	
214	Positive	*RHO*	c.251T>C	(p.Leu84Pro)	RP	1	
215	Positive	*RHO*	c.251T>C	(p.Leu84Pro)	RP		
216	Positive	*CEP290*	c.5254C>T	(p.Arg1752Trp)	EOSRD		
216	Positive	*CEP290*	c.734_735del	(p.Glu245Valfs*10)			
217	Positive	*ABCA4*	c.4316G>A	(p.Gly1439Asp)	Stargardt		
217	Positive	*ABCA4*	c.1018T>C	(p.Tyr340His)			
218	Positive	*RDH12*	c.701G>A	(p.Arg234His)	RP		1
219	Positive	*CHM*	Deletion Entire coding sequence	NA	CHM		
220	Positive	*PRPH2*	c.646C>T	(p.Pro216Ser)	MD		
221	Positive	*PCARE*	c.920T>A	(p.Leu307*)	RP		
221	Positive	*PCARE*	c.1827del	(p.Gln610Argfs*135)			
222	Positive	*MKKS*	c.830T>C	(p.Leu277Pro)	BBS		
222	Positive	*MKKS*	c.1013C>A	(p.Ser338*)			
223	Positive	*ABCA4*	c.4919G>A	(p.Arg1640Gln)	Stargardt		
223	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)			
224	Positive	*PRPF31*	c.901_919del	(p.Leu301Valfs*14)	MD		
225	Positive	*EYS*	c.6812_6813del	(p.Thr2271Argfs*11)	RP		
225	Positive	*EYS*	c.2023+5G>T	NA			
226	Positive	*PRPH2*	c.646C>T	(p.Pro216Ser)	MD		
227	Positive	*RP2*	Deletion Exon 1	NA	RP	1	
228	Positive	*RP2*	Deletion Exon 1	NA	RP		
229	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)	Usher syndrome		
229	Positive	*USH2A*	c.920_923dup	(p.His308Glnfs*16)			
230	Positive	*KIF11*	c.2684dup	(p.Asn895Lysfs*5)	Chorioretinopaty	1	
231	Positive	*KIF11*	c.2684dup	(p.Asn895Lysfs*5)	Chorioretinopaty	1	
232	Positive	*KIF11*	c.2684dup	(p.Asn895Lysfs*5)	Chorioretinopaty		
233	Positive	*EYS*	Deletion Exons 13-29	NA	RP		
233	Positive	*EYS*	c.5283T>A	(p.Tyr1761*)			
234	Positive	*RHO*	c.512C>A	(p.Pro171Gln)	RP	1	
235	Positive	*NR2E3*	c.166G>A	(p.Gly56Arg)	RP		
236	Positive	*RPGR*	c.1872_1873del	(p.Glu624Aspfs*5)	RP		
237	Positive	*RP2*	Deletion Entire coding sequence	NA	RP		
238	Positive	*PDE6B*	c.1726G>A	(p.Gly576Ser)	RP		1
239	Positive	*COL2A1*	c.1995+1G>T	NA	Stickler		
240	Positive	*ABCA4*	c.1240-1G>A	NA	Stargardt		
240	Positive	*ABCA4*	c.3564_3566delinsAAG	(p.Cys1188*)			
241	Positive	*RPGR*	c.2234_2237del	(p.Arg745Lysfs*69)	RP		
242	Positive	*MYO7A*	c.3892G>A	(p.Gly1298Arg)	Usher syndrome		
242	Positive	*MYO7A*	c.211_215dup	(p.Leu73Serfs*35)			
243	Positive	*ABCA4*	c.3113C>T	(p.Ala1038Val)	Stargardt		
243	Positive	*ABCA4*	c.4436G>A	(p.Trp1479*)			
244	Positive	*CDHR1*	c.1219C>T	(p.Arg407*)	RP		1
245	Positive	*PDE6A*	c.1705C>A	(p.Gln569Lys)	RP		
245	Positive	*PDE6A*	c.304C>T	(p.Arg102Cys)			
246	Positive	*RHO*	c.403C>T	(p.Arg135Trp)	RP		
247	Positive	*PDE6A*	c.1705C>A	(p.Gln569Lys)	RP		
247	Positive	*PDE6A*	c.1749C>G	(p.Tyr583*)			
248	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
249	Positive	*FAM161A*	c.1355_1356del	(p.Thr452Serfs*3)	RP		
249	Positive	*FAM161A*	c.1567C>T	(p.Arg523*)			
250	Positive	*RDH12*	c.278T>C	(p.Leu93Pro)	EOSRD		
250	Positive	*RDH12*	c.716G>A	(p.Arg239Gln)			
251	Positive	*RHO*	c.562G>A	(p.Gly188Arg)	RP		
252	Positive	*RDH12*	c.464C>T	(p.Thr155Ile)	EOSRD		
252	Positive	*RDH12*	c.716G>T	(p.Arg239Leu)			
253	Positive	*EYS*	c.1299+5_1299+8del	NA	RP		1

254	Positive	*RDH12*	c.295C>A	(p.Leu99Ile)	EOSRD		
254	Positive	*RDH12*	c.464C>T	(p.Thr155Ile)			
255	Positive	*RHO*	c.330C>G	(p.Cys110Trp)	RP		
256	Positive	*IQCB1*	c.1090C>T	(p.Arg364*)	EOSRD		
256	Positive	*IQCB1*	c.1465C>T	(p.Arg489*)			
257	Positive	*CRB1*	c.2843G>A	(p.Cys948Tyr)	EOSRD		1
258	Positive	*ARL6*	c.506del	(p.Gly169Alafs*6)	RP		1
259	Positive	*CNGB3*	c.1148del	(p.Thr383Ilefs*13)	ACHM		
259	Positive	*CNGB3*	c.852+1G>C	NA			
260	Positive	*RPGR*	c.2527del	(p.Glu843Lysfs*246)	RP		
261	Positive	*CNGB3*	c.1148del	(p.Thr383Ilefs*13)	ACHM		1
262	Positive	*ABCA4*	c.3210_3211dup	(p.Ser1071Cysfs*14)	Stargardt		
262	Positive	*ABCA4*	c.2588G>C	(p.Gly863Ala)			
263	Positive	*BEST1*	c.887A>G	(p.Asn296Ser)	MD		
264	Positive	*COL2A1*	c.192C>A	(p.Cys64*)	Stickler		
265	Positive	*PRPF31*	c.1263dup	(p.Lys422Glnfs*53)	RP		
266	Positive	*ARL6*	c.506del	(p.Gly169Alafs*6)	RP		
266	Positive	*ARL6*	c.350-2A>C	NA			
267	Positive	*ABCA4*	c.4919G>A	(p.Arg1640Gln)	Stargardt		
267	Positive	*ABCA4*	c.5929G>A	(p.Gly1977Ser)			
268	Positive	*USH2A*	c.1000C>T	(p.Arg334Trp)	RP		
268	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)			
268	Positive	*USH2A*	c.5858C>G	(p.Ala1953Gly)			
269	Positive	*CHM*	Deletion Exons 3-15	NA	CHM		
270	Positive	*RDH12*	c.524C>T	(p.Ser175Leu)	EOSRD		
270	Positive	*RDH12*	c.806_810del	(p.Ala269Glyfs*2)			
271	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
271	Positive	*USH2A*	c.8224-1G>A	NA			
272	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
273	Positive	*ABCA4*	c.6089G>A	(p.Arg2030Gln)	Stargardt		
273	Positive	*ABCA4*	c.5835+1G>T	NA			
274	Positive	*USH2A*	c.1841-2A>G	NA	RP		
274	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)			
275	Positive	*RPGR*	c.2405_2406del	(p.Glu802Glyfs*32)	RP	1	
276	Positive	*RPGR*	c.2405_2406del	(p.Glu802Glyfs*32)	RP		
277	Positive	*PDE6B*	c.1927_1969delinsGG	(p.Asn643Glyfs*29)	RP		
277	Positive	*PDE6B*	c.1258-2A>G	NA			
278	Positive	*ABCA4*	c.1804C>T	(p.Arg602Trp)	Stargardt	1	
278	Positive	*ABCA4*	c.3322C>T	(p.Arg1108Cys)			
278	Positive	*ABCA4*	c.6320G>A	(p.Arg2107His)			
279	Positive	*ABCA4*	c.1804C>T	(p.Arg602Trp)	Stargardt		
279	Positive	*ABCA4*	c.3322C>T	(p.Arg1108Cys)			
279	Positive	*ABCA4*	c.6320G>A	(p.Arg2107His)			
280	Positive	*RPGR*	c.2405_2406del	(p.Glu802Glyfs*32)	RP		
281	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
282	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
283	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
284	Positive	*ABCA4*	c.3386G>T	(p.Arg1129Leu)	Stargardt		
284	Positive	*ABCA4*	c.5959_5964delinsTG	(p.Gly1987*)			
285	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
286	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
287	Positive	*TTLL5*	c.1270C>T	(p.Gln424*)	RP		1
288	Positive	*TYR*	c.1037-7T>A	NA	OCA		
288	Positive	*TYR*	c.221_222del	(p.Val74Glyfs*2)			
289	Positive	*EYS*	c.5450G>A	(p.Trp1817*)	RP		1
290	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
291	Positive	*PAX6*	c.377T>A	(p.Val126Asp)	Aniridia	1	
292	Positive	*PAX6*	c.377T>A	(p.Val126A sp)	Aniridia		
293	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
294	Positive	*CHM*	Deletion Entire coding sequence	NA	CHM	1	
295	Positive	*CHM*	Deletion Entire coding sequence	NA	CHM	1	
296	Positive	*CHM*	Deletion Entire coding sequence	NA	CHM	1	
297	Positive	*CHM*	Deletion Entire coding sequence	NA	CHM	1	
298	Positive	*CHM*	Deletion Entire coding sequence	NA	CHM	1	
299	Positive	*CHM*	Deletion Entire coding sequence	NA	CHM		
300	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
301	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
302	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
303	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
304	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
305	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
306	Positive	*PROM1*	c.1956T>G	(p.Tyr652*)	CORD		

306	Positive	*PROM1*	c.2489+1G>A	NA			
307	Positive	*PAX6*	c.52G>C	(p.Gly18Arg)	Aniridia		
308	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
309	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
310	Positive	*ABCA4*	c.5512C>G	(p.His1838Asp)	Stargardt		1
310	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)			1
311	Positive	*ABCA4*	c.5461-10T>C	NA	Stargardt		
311	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)			
312	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
313	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
313	Positive	*USH2A*	c.2809+1G>A	NA			
314	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
315	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
316	Positive	*BEST1*	c.887A>G	(p.Asn296Ser)	MD		
317	Positive	*CERKL*	c.847C>T	(p.Arg283*)	RP		1
318	Positive	*FRMD7*	Deletion entire coding sequence	NA	Congenital nystagmus		
319	Positive	*USH2A*	c.10759C>T	(p.Gln3587*)	RP		
319	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)			
320	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
321	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
322	Positive	*RDH12*	c.716G>T	(p.Arg239Leu)	RP		1
323	Positive	*RPGR*	c.2236_2237del	(p.Glu746Argfs*23)	RP		
324	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP	1	
325	Positive	*ABCA4*	c.5413A>G	(p.Asn1805Asp)	Stargardt	1	
325	Positive	*ABCA4*	c.2588G>C	(p.Gly863Ala)			
326	Positive	*ABCA4*	c.4926C>G	(p.Ser1642Arg)	Stargardt		
326	Positive	*ABCA4*	c.5044_5058del	(p.Val1682_Val1686del)			
327	Positive	*ABCA4*	c.2041C>T	(p.Arg681*)	Stargardt		
327	Positive	*ABCA4*	c.5512C>G	(p.His1838Asp)			
327	Positive	*ABCA4*	c.5882G>A	(p. Gly1961Glu)			
328	Positive	*PRPF31*	c.221_224dup	(p.Lys76SerfsTer15)	RP		
329	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
330	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)	Stargardt		
330	Positive	*ABCA4*	c.3210_3211dup	(p.Ser1071CysfsTer14)			
331	Positive	*WFS1*	c.631G>A	(p.Asp211Asn)	Wolfram-like syndrome		1
332	Positive	*CERKL*	c.847C>T	(p.Arg283*)	CORD		1
333	Positive	*CRB1*	c.750T>A	(p.Cys250*)	RP		
333	Positive	*CRB1*	c.3307G>A	(p.Gly1103Arg)			
334	Positive	*USH2A*	c.10342G>A	(p.Glu3448Lys)	RP	1	
334	Positive	*USH2A*	c.1606T>C	(p.Cys536Arg)			
335	Positive	*USH2A*	c.10342G>A	(p.Glu3448Lys)	RP		
335	Positive	*USH2A*	c.1606T>C	(p.Cys536Arg)			
336	Positive	*CRX*	c.263A>G	(p.Lys88Arg)	RP		
337	Positive	*CHM*	c.1287_1288delTG	(p.Glu430Alafs*11)	CHM		
338	Positive	*USH2A*	c.2299delG	(p.Glu767Serfs*21)	RP		
338	Positive	*USH2A*	c.907C>A	(p.Arg303Ser)			
339	Positive	*PDE6B*	c.1572del	(p.Tyr525Thrfs*50)	RP		
339	Positive	*PDE6B*	c.1860del	(p.His620GlnfsTer23)			
340	Positive	*RHO*	c.512C>A	(p.Pro171Gln)	RP	1	
341	Positive	*RHO*	c.512C>A	(p.Pro171Gln)	RP		
342	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
342	Positive	*USH2A*	c.1876C>T	(p.Arg626*)			
343	Positive	*ABCA4*	c.6147+2T>A	NA	Stargardt		
343	Positive	*ABCA4*	c.5332A>G	(p.Met1778Val)			
344	Positive	*BBS2*	c.471G>A	(p.Thr157Thr)	BBS		1
345	Positive	*ABCA4*	c.6147+2T>A	NA	Stargardt		1
346	Positive	*GUCY2D*	c.2513G>A	(p.Arg838His)	CORD	1	
347	Positive	*GUCY2D*	c.2513G>A	(p.Arg838His)	CORD	1	
348	Positive	*GUCY2D*	c.2513G>A	(p.Arg838His)	CORD	1	
349	Positive	*GUCY2D*	c.2513G>A	(p.Arg838His)	CORD		
350	Positive	*ABCA4*	c.5882G>A	(p.Gly1961Glu)	Stargardt		
350	Positive	*ABCA4*	c.2032_2034del	(p.Lys678del)			
351	Positive	*RS1*	c.78+5G>C	NA	MD		
352	Positive	*PHYH*	c.683dup	(p.Val229SerfsTer2)	Refsum syndrome		
352	Positive	*PHYH*	c.380A>G	(p.Asp127Gly)			
353	Positive	*USH2A*	Deletions exons 4-72	NA	RP	1	
353	Positive	*USH2A*	c.9428A>G	(p.Tyr3143Cys)			
354	Positive	*USH2A*	Deletions exons 4-72	NA	RP		
354	Positive	*USH2A*	c.9428A>G	(p.Tyr3143Cys)			
355	Positive	*CACNA1F*	c.2269G>C	(p.Glu811Gln)	CSNB		
356	Positive	*RPGR*	c.823G>T	(p.Gly275Cys)	RP		
357	Positive	*RP1L1*	c.133C>T	(p.Arg45Trp)	MD		

358	Positive	*PRPF3*	c.1426+1G>A	NA	RP		
359	Positive	*PRPF8*	c.6337_6339del	(p.Lys2113del)	RP		
360	Positive	*RPGR*	c.1573-3C>G	NA	RP		
361	Positive	*USH2A*	c.10712C>T	(p.Thr3571Met)	RP		
361	Positive	*USH2A*	c.9424G>T	(p.Gly3142*)			
362	Positive	*PRPF8*	c.6994G>T	(p.Asp2332Tyr)	RP		
363	Positive	*CRX*	c.205C>T	(p.Arg69Cys)	RP		
364	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
365	Positive	*EYS*	c.6191+1G>A	NA	RP		
365	Positive	*EYS*	c.618_619del	(p.Ser207Trpfs*8)			
365	Positive	*EYS*	c.615_616delinsCC	(p.Phe206Leu)			
366	Positive	*EYS*	c.6191+1G>A	NA	RP	1	
366	Positive	*EYS*	c.618_619del	(p.Ser207Trpfs*8)			
366	Positive	*EYS*	c.615_616delinsCC	(p.Phe206Leu)			
367	Positive	*USH2A*	c.9119G>A	(p.Trp3040*)	RP		
367	Positive	*USH2A*	c.232T>G	(p.Phe78Val)			
367	Positive	*USH2A*	c.11927C>T	(p.Thr3976Met)			
368	Positive	*USH2A*	c.9119G>A	(p.Trp3040*)	Usher syndrome		1
369	Positive	*KIZ*	c.226C>T	(p.Arg76*)	RP		1
370	Positive	*ABCA4*	c.179C>T	(p.Ala60Val)	Stargardt		
370	Positive	*ABCA4*	c.3299T>C	(p.Ile1100Thr)			
371	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
372	Positive	*RPGR*	c.2442_2445del	(p.Gly817Lysfs*2)	RP		
373	Positive	*MERTK*	Deletion Exons 3-19	NA	RP		1
374	Positive	*USH2A*	c.11864G>A	(p.Trp3955*)	Usher syndrome		
374	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)			
375	Positive	*CHM*	c.757C>T	(p.Arg253*)	CHM		
376	Positive	*CNGB1*	c.2544dup	(p.Leu849Alafs*3)	RP		
376	Positive	*CNGB1*	c.382-3C>G	NA			
377	Positive	*PDE6B*	c.1107+3A>G	NA	RP	1	
377	Positive	*PDE6B*	c.892C>T	(p.Gln298*)			
378	Positive	*PDE6B*	c.1107+3A>G	NA	RP		
378	Positive	*PDE6B*	c.892C>T	(p.Gln298*)			
379	Positive	*RPGR*	c.1345C>T	(p.Arg449*	RP		
380	Positive	*USH2A*	c.9119G>A	(p.Trp3040*)	Usher syndrome		1
381	Positive	*PDE6A*	c.1955_1974dup	(p.Ile659Valfs*10)	RP		1
382	Positive	*SNRNP200*	c.2042G>A	(p.Arg681His)	RP	1	
383	Positive	*SNRNP200*	c.2042G>A	(p.Arg681His)	RP		
384	Positive	*EYS*	c.6131_6134del	(p.Asn2044Thrfs*11)	RP		
384	Positive	*EYS*	c.6416G>A	(p.Cys2139Tyr)			
385	Positive	*USH2A*	c.9119G>A	(p.Trp3040*)	Usher syndrome		1
386	Positive	*USH2A*	c.13217T>C	(p.Leu4406Pro)	RP		
386	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)			
387	Positive	*USH2A*	c.13374del	(p.Glu4458Aspfs*3)	RP		
387	Positive	*USH2A*	c.1531G>A	(p.Glu511Lys)			
388	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome	1	
388	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
389	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		
389	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
390	Positive	*RP1*	c.5564del	(p.Lys1855Argfs*42)	RP		1
391	Positive	*ABCA4*	c.3386G>T	(p.Arg1129Leu)	Stargardt		
391	Positive	*ABCA4*	c.5531_5557dup	(p.Gly1844_Gln1852dup)			
392	Positive	*USH2A*	c.2809+1G>A	NA	Usher syndrome		
392	Positive	*USH2A*	c.8917_8918del	(p.Leu2973Lysfs*79)			
393	Positive	*RS1*	c.626G>A	(p.Arg209His)	XLRS	1	
394	Positive	*RS1*	c.626G>A	(p.Arg209His)	MD		
395	Positive	*RPGR*	c.3218_3236dup	p.Glu1075Valfs*10	CORD		
396	Positive	*RHO*	c.1040C>T	(p.Pro347Leu)	RP		
397	Positive	*RP1L1*	c.133C>T	(p.Arg45Trp)	MD		
398	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)	Usher syndrome		1
399	Positive	*MYO7A*	c.3612delC	(p.Ser1205Profs*27)	Usher syndrome		
399	Positive	*MYO7A*	c.1623dupC	(p.Lys542Glnfs*5)			
400	Positive	*SNRNP200*	c.3260C>T	(p.Ser1087Leu)	RP		
401	Positive	*EYS*	c.3938T>A	(p.Leu1313*)	RP		
401	Positive	*EYS*	c.514C>T	(p.Gln172*)			
402	Positive	*ABCA4*	c.1648G>A	(p.Gly550Arg)	MD		
402	Positive	*ABCA4*	c.5714+5G>A	NA			
403	Positive	*RPGR*	c.2543delA	(p.Glu848Glyfs*241)	RP		
404	Positive	*PRPH2*	c.535T>G	(p.Trp179Gly)	RP		
405	Positive	*RPGR*	c.2234_2237delGAGA	(p.Arg745Lysfs*69)	RP		
406	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)	RP		
406	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			

406	Positive	*USH2A*	c.14257G>A	(p.Val4753Ile)			
407	Positive	*EYS*	c.6812_6813del	(p.Thr2271Argfs*11)	RP		
407	Positive	*EYS*	c.2023+5G>T	NA			
408	Positive	*MERTK*	Deletion exon 9	NA	RP		
408	Positive	*MERTK*	c.280_81del	(p.Leu94Cysfs*13)			
409	Positive	*RPGR*	c.2412_2418del	(p.Gly805Lysfs*8)	RP		
410	Positive	*PROM1*	c.1956T>G	(p.Tyr652*)	CORD		1
411	Positive	*RHO*	c.251T>C	(p.Leu84Pro)	RP		
412	Positive	*USH2A*	c.11713C>T	(p.Arg3905Cys)	RP		
412	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)			
413	Positive	*RHO*	c.403C>T	(p.Arg135Trp)	RP		
414	Positive	*PDE6A*	c.1117G>T	(p.Glu373*)	RP		
414	Positive	*PDE6A*	c.2135+1G>T	NA			
415	Positive	*USH2A*	c.12575G>A	(p.Arg4192His)	RP		
415	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)			
416	Positive	*CRB1*	c.2234C>T	(p.Thr745Met)	EOSRD		
416	Positive	*CRB1*	c.2053G>A	(p.Gly685Arg)			
417	Positive	*VPS13B*	c.5998_5999del	(p.Leu2000Alafs*2)	Cohen syndrome		
417	Positive	*VPS13B*	c.6614T>G	(p.Ile2205Arg)			
418	Positive	*USH2A*	c.12575G>A	(p.Arg4192His)	RP		
418	Positive	*USH2A*	c.15089C>A	(p.Ser5030*)			
419	Positive	*CHM*	c.702+3_702+12del	NA	CHM		
420	Positive	*RDH12*	c.278T>C	(p.Leu93Pro)	RP	1	
420	Positive	*RDH12*	c.148G>A	(p.Gly50Ser)			
421	Positive	*RDH12*	c.278T>C	(p.Leu93Pro)	RP		
421	Positive	*RDH12*	c.148G>A	(p.Gly50Ser)			
422	Positive	*CNGB1*	c.1276del	(p.Glu426Argfs*77)	RP		1
423	Positive	*CHM*	c.1603G>T	(p.Glu535*)	CHM		
424	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)	Usher syndrome		
424	Positive	*USH2A*	c.1551-9T>A	NA			
425	Positive	*PRPF8*	c.6337_6339del	(p.Lys2113del)	RP		
426	Positive	*RPGR*	c.823G>T	(p.Gly275Cys)	RP	1	
427	Positive	*RPGR*	c.823G>T	(p.Gly275Cys)	RP	1	
428	Positive	*RPGR*	c.823G>T	(p.Gly275Cys)	RP		
429	Positive	*CLRN1*	c.189C>A	(p.Tyr63*)	Usher syndrome		
429	Positive	*CLRN1*	c.144T>G	(p.Asn48Lys)			
430	Positive	*RS1*	c.305G>A	(p.Arg102Gln)	XLRS	1	
431	Positive	*RS1*	c.305G>A	(p.Arg102Gln)	XLRS		
432	Positive	*RPE65*	c.202C>T	(p.His68Tyr)	EOSRD		
432	Positive	*RPE65*	c.314C>T	(p.Thr105Ile)			
433	Positive	*RS1*	c.637C>T	(p.Arg213Trp)	XLRS		
434	Positive	*CRX*	c.591_594dup	(p.Ser199GlyfsTer38)	EOSRD	1	
435	Positive	*CRX*	c.591_594dup	(p.Ser199GlyfsTer38)	EOSRD	1	
436	Positive	*CRX*	c.591_594dup	(p.Ser199GlyfsTer38)	EOSRD		
437	Positive	*BEST1*	c.13T>A	(p.Tyr5Asn)	MD		
438	Positive	*IMPG1*	c.1543_1544dup	(p.Met515IlefsTer6)	MD		
439	Positive	*PROM1*	c.1354dup	(p.Tyr452Leufs*13)	CORD		
439	Positive	*PROM1*	Deletion Exon 16	NA			
440	Positive	*USH2A*	c.2809+1G>A	NA	RP		
440	Positive	*USH2A*	c.13018G>A	(p.Gly4340Arg)			
441	Positive	*RLBP1*	c.25C>T	(p.Arg9Cys)	RP		1
442	Positive	*RPGR*	c.2964_2965del	(p.Glu989Glyfs*89)	RP	1	
443	Positive	*RPGR*	c.2964_2965del	(p.Glu989Glyfs*89)	RP		
444	Positive	*RHO*	c.560G>T	(p.Cys187Phe)	RP		
445	Positive	*RPGR*	c.3092_3093del	(p.Glu1031Glyfs*47)	CORD		
446	Positive	*GUCY2D*	c.2512C>T	(p.Arg838Cys)	MD		
447	Positive	*ABCA4*	c.4457C>T	(p.Pro1486Leu)	MD		
447	Positive	*ABCA4*	c.6383A>G	(p.His2128Arg)			
448	Positive	*RDH12*	c.278T>C	(p.Leu93Pro)	EOSRD		1
449	Positive	*RHO*	c.1033G>C	(p.Val345Leu)	RP		
450	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
451	Positive	*ALMS1*	c.542_545dup	(p.Asp182Glufs*4)	Alstrom syndrome		
451	Positive	*ALMS1*	c.9784+1G>C	NA			
452	Positive	*RHO*	c.760_762dup	(p.Val254dup)	RP	1	
453	Positive	*RHO*	c.760_762dup	(p.Val254dup)	RP	1	
454	Positive	*RHO*	c.760_762dup	(p.Val254dup)	RP		
455	Positive	*RTN41P1*	c.968_972dup	(p.Gly325Leufs*2)	RP		
455	Positive	*RTN41P1*	c.1084A>T	(p.Ile362Phe)			
456	Positive	*ABCA4*	c.5822A>C	(p.His1941Pro)	Stargardt		1
457	Positive	*RP1*	c.2555del	(p.Lys852Argfs*4)	RP		
458	Positive	*TSPAN12*	c.916del	(p.*306Lysext*19)	FEVR		
459	Positive	*EYS*	c.7943delC	(p.Thr2648LysfsTer34)	RP		

459	Positive	*EYS*	c.2527G>A	(p.Gly843Arg)			
460	Positive	*CRB1*	c.2234C>T	(p.Thr45Met)	EOSRD		
460	Positive	*CRB1*	c.596C>A	(p.Ala199Asp)			
461	Positive	*RPGR*	c.2501del	(p.Glu834Glyfs*255)	RP	1	
462	Positive	*RPGR*	c.2501del	(p.Glu834Glyfs*255)	RP	1	
463	Positive	*RPGR*	c.2501del	(p.Glu834Glyfs*255)	RP		
464	Positive	*CNGB3*	c.1148del	(p.Thr383Ilefs*13)	ACHM		1
465	Positive	*GPR143*	c.360G>A	(p.Ala120Ala)	Ocular albinism		
466	Positive	*OCA2*	Deletion Exon 14	NA	OCA		
466	Positive	*OCA2*	Deletion Exon 7	NA			
467	Positive	*RPGR*	c.1345C>T	(p.Arg449*)	RP		
468	Positive	*COL18A1*	c.2673dup	(p.Gly892Argfs*9)	Knobloch		
468	Positive	*COL18A1*	c.1765G>T	(p.Gly589*)			
469	Positive	*RP1*	c.1719_1723del	(p.Ser574Cysfs*7)	RP		
469	Positive	*RP1*	c.3157del	(p.Tyr1053Thrfs*4)			
470	Positive	*EYS*	c.7943del	(p.Thr2648Lysfs*34)	RP	1	
470	Positive	*EYS*	c.2527G>A	(p.Gly843Arg)			
471	Positive	*EYS*	c.7943del	(p.Thr2648Lysfs*34)	RP		
471	Positive	*EYS*	c.2527G>A	(p.Gly843Arg)			
472	Positive	*USH2A*	c.10636G>A	(p.Gly3546Arg)	RP		
472	Positive	*USH2A*	c.2809+1G>A	NA			
473	Positive	*EYS*	Deletion Exon 17-22	NA	RP		
473	Positive	*EYS*	c.4955C>G	(p.Ser1652*)			
474	Positive	*USH2A*	c.13621C>T	(p.Gln4541*)	Usher syndrome		1
475	Positive	*CERKL*	c.847C>T	(p.Arg283*)	RP		1
476	Positive	*PRPF31*	c.221_224dup	(p.Lys76Serfs*15)	RP	1	
477	Positive	*PRPF31*	c.221_224dup	(p.Lys76Serfs*15)	RP	1	
478	Positive	*ABCA4*	c.3386G>T	(p.Arg1129Leu)	Stargardt		
478	Positive	*ABCA4*	c.1390_1391del	(p.Leu464Glufs*2)			
479	Positive	*ABCA4*	c.3386G>T	(p.Arg1129Leu)	Stargardt	1	
479	Positive	*ABCA4*	c.1390_1391del	(p.Leu464Glufs*2)			
480	Positive	*CNGB1*	c.382-3C>G	NA	RP		1
481	Positive	*CNGB1*	c.382-3C>G	NA	RP	1	1
482	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)	RP		
482	Positive	*USH2A*	c.8681+2T>C	NA			
483	Positive	*USH2A*	c.14287G>A	(p.Gly4763Arg)	RP		
483	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)			
484	Positive	*PRPF6*	c.514C>T	(p.Arg172Trp)	RP		
485	Positive	*GUCY2D*	c.2512C>T	(p.Arg838Cys)	MD		
486	Positive	*USH2A*	c.12575G>A	(p.Arg4192His)	RP		
486	Positive	*USH2A*	c.14885dup	(p.Glu4963Glyfs*38)			
487	Positive	*CNGB3*	c.852+1G>C	NA	ACHM		1
488	Positive	*CHM*	c.1674del	(p.Asp559Thrfs*24)	CHM		
489	Positive	*BEST1*	c.140G>A	(p.Arg47His)	MD		
490	Positive	*BEST1*	c.652C>T	(p.Arg218Cys)	MD		
491	Positive	*USH2A*	c.2276G>T	(p.Cys759Phe)	RP		
491	Positive	*USH2A*	c.2299del	(p.Glu767Serfs*21)			
492	Positive	*CNGB1*	c.382-3C>G	NA	RP		1
493	Positive	*RPGR*	c.356T>C	(p.Leu119Ser)	RP		
494	VUS	*COL11A1*	c.740C>G	(p.Ala247Gly)	Stickler		
495	VUS	*PROM1*	c.2490-2A>G	NA	RP		
495	VUS	*RPGR*	c.436T>C	(p.Ser146Pro)			
496	VUS	*CACNA1F*	c.3965A>T	(p.Lys1322Met)	Nyctalopia, high myopia and decreased acuity		
497	VUS	*RPE65*	c.1444G>T	(p.Asp482Tyr)	EOSRD	1	
497	VUS	*RPE65*	c.998G>A	(p.Gly333Glu)			
498	VUS	*RPE65*	c.1444G>T	(p.Asp482Tyr)	EOSRD		
498	VUS	*RPE65*	c.998G>A	(p.Gly333Glu)			
499	VUS	*EYS*	Deletion Exon 22	NA	RP		
499	VUS	*EYS*	c.6602T>C	(p.Leu2201Pro)			
500	VUS	*USH2A*	c.15089C>A	(p.Ser5030*)	RP		
500	VUS	*USH2A*	c.8371G>C	(p.Val2791Leu)			
501	VUS	*PDE6C*	c.2036+1G>C	NA	ACHM		
501	VUS	*PDE6C*	c.295T>C	(p.Phe99Leu)			
502	VUS	*USH2A*	c.9799T>C	(p.Cys3267Arg)	RP		
502	VUS	*USH2A*	c.13139C>T	(p.Thr4380Ile)			
503	VUS	*BBS2*	c.1015C>T	(p.Arg339Ter)	RP		
503	VUS	*BBS2*	c.117G>A	(p.Lys39Lys)			
504	VUS	*USH2A*	c.14219C>A	(p.Ala4740Asp)	RP		
504	VUS	*USH2A*	c.5519G>A	(p.Gly1840Glu)			
505	VUS	*EYS*	c.4120C>T	(p.Arg1374*)	RP		
505	VUS	*EYS*	c.8981T>A	(p.Leu2994Gln)			

506	VUS	*ARSG*	c.284G>A	(p.Arg95Gln)	Usher syndrome		
506	VUS	*ARSG*	c.1010G>A	(p.Trp337*)			
507	VUS	*USH2A*	c.14134-3169A>G	NA	RP		
507	VUS	*USH2A*	c.6163G>A	(p.Ala2055Thr)			
508	VUS	*CDHR1*	c.1219C>T	(p.Arg407*)	RP		
508	VUS	*CDHR1*	c.2119G>C	(p.Gly707Arg)			
509	VUS	*ADGRV1*	c.13758_13761del	(p.Gly4587Glufs*2)	Usher syndrome		
509	VUS	*ADGRV1*	c.17461T>C	(p.Ser5821Pro)			
510	VUS	*USH2A*	c.11816_11822dup	(p.Val3942Ilefs*7)	RP		
510	VUS	*USH2A*	c.13531G>A	(p.Ala4511Thr)			
511	VUS	*EYS*	c.6131_6134del	(p.Asn2044ThrfsTer11)	RP	1	
511	VUS	*EYS*	c.8959A>G	(p.Ser2987Gly)			
512	VUS	*EYS*	c.6131_6134del	(p.Asn2044ThrfsTer11)	RP		
512	VUS	*EYS*	c.8959A>G	(p.Ser2987Gly)			
513	VUS	*IFT74*	c.466-2A>G	NA	BBS		
513	VUS	*IFT74*	c.120+2dup	NA			
514	VUS	*USH2A*	c.11597C>T	(p.Ala3866Val)	RP		1
515	VUS	*CERKL*	c.497C>T	(p.Pro166Leu)	RP		1
516	VUS	*RPGR*	c.1481G>T	(p.Gly494Val)	RP		
517	VUS	*PRPF8*	c.4103T>C	(p.Leu1368Ser)	RP		
518	VUS	*PDE6B*	c.1151T>C	(p.Leu384Pro)	RP		
518	VUS	*PDE6B*	c.515C>A	(p.Thr172Lys)			
519	VUS	*ABCA4*	c.5882G>A	(p.Gly1961Glu)	Stargardt		
519	VUS	*ABCA4*	c.4877C>A	(p.Ala1626Asp)			
520	VUS	*PRPF3*	c.424-20A>G	NA	RP		
521	VUS	*IMPG1*	c.2162G>C	(p.Ser721Thr)	MD		
521	VUS	*PROM1*	c.1780A>C	(p.Ile594Leu)			
521	VUS	*PROM1*	c.604C>G	(p.Arg202Gly)			
522	VUS	*IFT172*	c.3426del	(p.Glu1143ArgfsTer174)	RP		
522	VUS	*IFT172*	c.3925G>A	(p.Gly1309Ser)			
523	VUS	*RP2*	c.535C>T	(p.Pro179Ser)	RP		
524	VUS	*EYS*	c.9022A>G	(p.Ser2987Gly)	RP	1	
524	VUS	*EYS*	c.6131_6134delATAA	(p.Asn2044ThrfsTer11)			
525	VUS	*EYS*	c.9022A>G	(p.Ser2987Gly)	RP		
525	VUS	*EYS*	c.6131_6134delATAA	(p.Asn2044ThrfsTer11)			
526	VUS	*MERTK*	c.518A>G	(p.Tyr173Cys)	RP		
526	VUS	*MERTK*	c.734C>A	(p.Ser245Tyr)			
527	VUS	*IMPG2*	c.533G>A	(p.Ser178Asn)	RP		1
528	VUS	*HK1*	c.1550G>A	(p.Arg517Gln)	RP		
529	VUS	*PROM1*	c.1441G>T	(p.Val481Phe)	MD		
530	VUS	*USH2A*	c.9799T>C	(p.Cys3267Arg)	RP		
530	VUS	*USH2A*	c.2723T>C	(p.Leu908Ser)			
530	VUS	*USH2A*	c.8846-3C>G	NA			
531	VUS	*RP1*	c.3416del	(p.Val1139GlyfsTer69)	RP		
531	VUS	*BBS2*	c.345+5G>A	NA			
531	VUS	*BBS2*	c.815G>A	(p.Arg272Gln)			
532	VUS	*ARHGEF18*	c.1276A>C	(p.Ile426Leu)	RP		
532	VUS	*ARHGEF18*	c.305C>T	(p.Ser102Phe)			
533	VUS	*USH2A*	c.12094G>A	(p.Gly4032Arg)	RP		
533	VUS	*USH2A*	c.235T>C	(p.Cys79Arg)			
534	VUS	*USH2A*	c.2809+1G>A	NA	RP		
534	VUS	*USH2A*	c.457T>C	(p.Trp153Arg)			
535	VUS	*CDHR1*	c.1219C>T	(p.Arg407*)	RP		
535	VUS	*CDHR1*	c.862+4A>C	NA			
536	One candidate	*CRB1*	c.2843G>A	(p.Cys948Tyr)	EOSRD		
537	One candidate	*ADGRV1*	c.8995C>G	(p.Gln2999Glu)	Usher syndrome		
538	One candidate	*REEP6*	c.481C>T	(p.Arg161Ter)	RP		
539	One candidate	*NPHP1*	Deletion entire coding sequence	NA	Senior Loken		
540	One candidate	*RPE65*	c.1022T>C	(p.Leu341Ser)	EOSRD		
541	One candidate	*TCTN3*	c.3G>A	(p.Met1Ile)	Joubert Syndrome		
542	One candidate	*IFT172*	c.402+2T>G	NA	BBS		
543	One candidate	*RDH12*	c.295C>A	(p.Leu99Ile)	EOSRD		
544	One candidate	*CYP4V2*	c.1199G>A	(p.Arg400His)	Bietti		
545	One candidate	*USH2A*	c.2299del	(p.Glu767SerfsTer21)	RP		
546	One candidate	*CNGB3*	c.1148del	(p.Thr383Ilefs*13)	ACHM		
547	One candidate	*CEP290*	c.4945C>T	(p.Gln1649*)	EOSRD		
548	One candidate	*KIZ*	c.226C>T	(p.Arg76*)	RP		
549	One candidate	*CDH23*	Deletion Exons 17-19	NA	Usher syndrome		
550	One candidate	*EYS*	Deletion Exon 12	NA	RP		
551	One candidate	*ARL6*	c.506del	(p.Gly169Alafs*6)	RP		
552	One candidate	*PHYH*	c.823C>T	(p.Arg275Trp)	RP		
553	One candidate	*HGSNAT*	Gain Exons 6-18	NA	RP		

554	One candidate	*NMNAT1*	Deletion Exon 4-5	NA	EOSRD		
555	One candidate	*KCNV2*	c.889_901del	(p.Asp297Serfs*21)	RP		
556	One candidate	*PDE6B*	c.1655G>A	(p.Arg552Gln)	RP		
557	One candidate	*ABCA4*	c.5077G>A	(p.Val1693Ile)	MD		
558	One candidate	*CNGB1*	c.2030G>A	(p.Arg677His)	EOSRD		
559	One candidate	*RP1*	c.1299_1306dup	(p.Gln436Leufs*22)	RP		
560	One candidate	*LRP5*	c.2543C>T	(p.Pro848Leu)	Stickler	1	
561	One candidate	*LRP5*	c.2543C>T	(p.Pro848Leu)	Stickler		
562	One candidate	*RDH12*	c.716G>T	(p.Arg239Leu)	RP		
563	One candidate	*BBS1*	c.1169T>G	(p.Met390Arg)	RP		
564	One candidate	*RDH12*	c.152T>A	(p.Ile51Asn)	RP		
565	One candidate	*RDH12*	c.806_810del	(p.Ala269Glyfs*2)	RP		
566	One candidate	*CEP290*	c.734_735del	(p.Glu245Valfs*10)	EOSRD		
567	One candidate	*MAK*	c.170_171del	(p.Leu57Glnfs*7)	RP		
568	One candidate	*PDE6B*	c.1726G>A	(p.Gly576Ser)	RP		
569	One candidate	*ABCA4*	Deletion Exons 39-40	NA	Stargardt		
570	One candidate	*IMPG2*	Partial Deletion Exons 13-14	NA	RP		
571	One candidate	*TYR*	Deletion Exon 3	NA	OCA		
572	One candidate	*USH2A*	c.1724G>A	(p.Cys575Tyr)	RP		
573	One candidate	*RDH12*	c.295C>A	(p.Leu99Ile)	RP	1	
574	One candidate	*RDH12*	c.295C>A	(p.Leu99Ile)	RP		
575	One candidate	*RDH12*	c.701G>A	(p.Arg234His)	EOSRD		
576	One candidate	*ABCA4*	c.3050+5G>A	NA	Stargardt		
577	One candidate	*RDH12*	c.701G>A	(p.Arg234His)	RP		
578	One candidate	*USH2A*	c.9119G>A	(p.Trp3040*)	RP		
579	One candidate	*ABCA4*	c.5882G>A	(p.Gly1961Glu)	Stargardt		
580	One candidate	*IMPG2*	Partial Deletion Exons 13-14	NA	RP	1	
581	One candidate	*IMPG2*	Partial Deletion Exons 13-14	NA	RP	1	
582	One candidate	*IMPG2*	Partial Deletion Exons 13-14	NA	RP		

Abbreviations: RP: retinitis pigmentosa, OCA: oculo-cutaneous albinism, XLRS: X-linked retinoschisis, MD: macular dystrophy, PHPV: persistent hyperplastic primary vitreous, FEVR: Familial exudative vitreoretinopathy, CHM: choroideremia, EOSRD: Early onset severe retinal dystrophy, BBS: Bardet-Biedl syndrome, ACHM: achromatopsia, OA: optic atrophy, CORD: cone-rod dystrophy, CSNB: congenital stationary night blindness.