Supplementary material for Sodi et al., "CFH Y402H polymorphism in Italian patients with age-related macular degeneration, retinitis pigmentosa, and Stargardt disease," *Ophthalmic Genetics,* 2018.

Supplementary Table 1. The genotypes of Retinitis pigmentosa patients included in the study

Patient	Gene	Allele 1	Allele 2
1	PRPF8	c.5804G>A (p.Arg1935His)	Wt
2	NR2E3	c.119-2A>C (p.?)	c.328C>T (p.Gln110*)
3	USH2A	c.14379G>A (p.Gly1460Glu)	c.9878del (p.Lys3293Serfs*8)
4	EYS	c.1682_1686del (p.Asn561Ilefs*7)	c.1682_1686del (p.Asn561Ilefs*7)
5	PRPF31	c.420+2T>G (p.?)	wt
6	PDE6A	c.2053G>A (p.Val685Met)	c.2053G>A (p.Val685Met)
7	CRB1	c.221G>A 8p.Cys74Tyr)	c.1623C>A (p.Tyr541*)
8	USH2A	c.9811del (p.Met3271Cysfs*30)	c.9959-1G>C (p.?)
9	PDE6B	c.719T>C (p.Leu240Pro)	c.1107+3A>G (p.?)
10	CRB1	c.221G>A 8p.Cys74Tyr)	c.1623C>A (p.Tyr541*)
11	PDE6B	c.1929+2T>C (p.?)	c.1929+2T>C (p.?)
12	RPGR	c.1591G>T (p.Glu531*)	-
13	RHO	c.568G>A (p.Asp190Asn)	wt
14	RPGR	c.1234C>T (p.Arg412*)	-
15	RP2	c.409_411del (p.Ile37del)	wt
16	RPGR	c.1234C>T (p.Arg412*)	-
17	CRB1	c.1445_1453del (p.Ile482_Thr484del)	c.1445_1453del (p.Ile482_Thr484del)
18	RP2	c.892_893del (p.Ile298Cysfs*5)	-
19	RHO	c.403C>T (p.Arg135Trp)	wt
20	RHO	c.568G>A (p.Asp190Asn)	wt
21	RPGR	c.1234C>T (p.Arg412*)	-
22	RHO	c.568G>A (p.asp190Asn)	wt
23	RPGR	c.1060-?_3459+?	-
24	RHO	c.1040C>T (p.Pro347Leu)	wt
25	RHO	c.568G>A (p.Asp190Asn)	wt
26	CEP290	c.1666del (p.Ile556Phefs*17)	c.2991+1655A>G (p.?)
27	PRPF8	c.5804G>A (p.Arg1935His)	wt
28	RPGR	c.439G>C (p.Ala147Pro)	-
29	RHO	C.569A>G (p.Asp190Gly)	wt
30	C2ORF71	c.8G>A (p.Cys3Tyr)	c.3604C>T (p.Arg1202*)
31	PDE6A	c.304C>A (p.Arg102Ser)	c.1966G>T (p.Glu656*)
32	CNGB1	c.2296T>C (p.Cys766Arg)	c.2296T>C (p.Cys766Arg)
33	RDH12	c.146C>A (p.Thr49Lys)	c.184C>T (p.Arg62*)
34	USH2A	c.12168G>A (p.Trp4056*)	c.13166A>G (p.Tyr4389Cys)
35	RP2	c.352C>T (p.Arg118Cys)	-
36	RHO	c.644C>T (p.Pro215Leu)	wt
37	USH2A	c.2276G>T (p.Cys759Phe)	c.7685T>C (p.Val2562Ala)
38	IMPG2	c.851T>G (p.Leu284*)	c.851T>G (p.Leu284*)
39	CRX	c.122G>A (p.Arg41Gln)	wt

40	USH2A	c.7955G>C (p.Gly2652Ala)	c.3317-?_4251+?
41	RPGR	c.1060-?_3459+?	-
42	RDS	c.535T>G (p.Trp179Gly)	wt
43	EYS	c.2137+1G>A (p.?)	c.3131A>G (p.Asn1044Ser)
44	CERKL	c.481+2G>A (p.?)	c.481+2G>A (p.?)
45	RP2	c.409_411del (p.Ile37del)	-
46	RPGR	c.2169_2172del (p.Arg723Serfs*91)	-
47	PITPNM3	c.2792A>C (p.Gln931Pro)/c.2793G>C (p.Gln931His)	wt
48	ABCR	c.5196+1G>A (p.?)	c.5882G>A (p.Gly1961Glu)
49	C2ORF71	c.1525del (p.Thr509Leufs*32)	c.1525del (p.Thr509Leufs*32)
50	RHO	c.644C>T (p.Pro215Leu)	wt
51	RP2	c.892_893del (p.Ile298Cysfs*5)	-
52	PRPF8	c.7000T>A (p.Tyr2334Asn)	wt
53	RPGR	c.1234C>T (p.Arg412*)	-

Supplementary Table 2. ABCA4 gene sequence variants of patients affected by Stargardt disease included in the study

Patient	Allele 1	Allele 2	Allele 3
1	c.2099G>A (p.Trp700X)	c.4457C>T (p.Pro1486Leu)	
2	c.5882G>A (p.Gly1961Glu)	c.571-2A>T (p.?)	
3	c.2461T>A (p.Trp821Arg)	c.2461T>A (p.Trp821Arg)	
4	c.5882G>A (p.Gly1961Glu)	c.2791G>A (p.Val931Met)	c.288C>A (p.Asn96Lys)
5	c.4297G>A (p.Val1433Ile)	c.4297G>A (p.Val1433Ile)	
6	c.2933G>A (p.Gly978Asp)	c.4577C>T (p.Thr1526Met)	
7	c.634C>T (p.Arg212Cys)	c.3056C>T (p.T1hr019Met)	
8	c.634C>T (p.Arg212Cys)	c.3056C>T (p.T1hr019Met)	
9	c.5882G>A (p.Gly1961Glu)	c.5417G>A (p.Ser1806Asn)	
10	c.247_250dup (p.Ser84Thrfs*16)	c.5087G>A (p.Ser1696Asn)	
11	c.1239+1G>C (p.?)	c.3296C>G (p.Ser1099*)	
12	c.2345G>A (p.Trp782*)	c.6320G>A (p.Arg2107His)	
13	c.2300T>A (p.Val767Asp)	c.6088C>T (p.Arg2030*)	
14	c.3610G>A (p.Asp1204Asn)	c.5527C>T (p.Arg1843Trp)	c.6079C>T (p.Leu2027Phe)
15	c.2461T>A (p.Trp821Arg)	c.3323G>A (p.Arg1108His)	c.4297G>A (p.Val1433Ile)
16	c.2791G>A (p.Val931Met)	c.3322C>T (p.Arg1108Cys)	
17	c.571-1G>T (p.?)	c.3322C>T (p.Arg1108Cys)	
18	c.5882G>A (p.Gly1961Glu)	c.6658C>T (p.Gln2220*)	
19	c.2791G>A (p.Val931Met)	c.4234C>T (p.Gln1412*)	
20	c.1937+1G>A (p.?)	c.4128G>A (p.=) (p.Gln1376Gln)	
21	c.2069G>T (p.Gly690Val)	c.3994C>T (p.Gln1332*)	
22	c.5882G>A (p.Gly1961Glu)	c.4450C>T (p.Pro1484Ser)	c.203C>T (p.Pro68Leu)
23	c.4437G>A (p.Trp1479*)	c.6419T>A (p.Leu2140Gln)	
24	c.3292C>T (p.Arg1098Cys)	c.5908C>T (p.Leu1970Phe)	c.6545_6548dup (p.Pro2184Glufs*68)
25	c.1846G>A (p.Glu616Lys)	c.4739T>C (p.Leu1580Ser)	c.6515A>G (p.Lys2172Arg)
26	c.4417C>A (p.Leu1473Met)	c.1714C>T (p.Arg572*)	
27	c.5882G>A (p.Gly1961Glu)	c.5836-2delA (p.?)	
28	c.2461T>A (p.Trp821Arg)	c.5714+5G>A (p.?)	
29	c.2894A>G (p.Asn965Ser)	c.5714+5G>A (p.?)	
30	c.2887G>C (p.Gly963Arg)	c.5714+5G>A (p.?)	
31	c.5882G>A (p.Gly1961Glu)	c.5018+2T>C (p.?)	
32	c.4667+1G>A (p.?)	c.5512C>A (p.His1838Asn)	
33	c.288C>A (p.Asn96Lys)	c.2933G>A (p.Gly978Asp)	
34	c.4383G>C (p.Trp1461Cys)	c.5929G>A (p.Gly1977Ser)	
35	c.5714+5G>A (p.?)	c.5929G>A (p.Gly1977Ser)	
36	c.5882G>A (p.Gly1961Glu)	c.4793C>A (p.Ala1598Asp)	
37	c.5882G>A (p.Gly1961Glu)	c.514G>A (p.Gly172Ser)	
38	c.5882G>A (p.Gly1961Glu)	c.4139C>T (p.Pro1380Leu)	
39	c.5882G>A (p.Gly1961Glu)	c.3323G>A (p.Arg1108His)	
40	c.5882G>A (p.Gly1961Glu)	c.4139C>T (p.Pro1380Leu)	
41	c.5882G>A (p.Gly1961Glu)	c.4793C>A (p.Ala1598Asp)	
42	c.5882G>A (p.Gly1961Glu)	c.5018+2T>C (p.?)	
43	c.5882G>A (p.Gly1961Glu)	c.3994_4017dup (p.Gln1332_Cys1339dup)	c.4577C>T (p.Thr1526Met)
44	c.5882G>A (p.Gly1961Glu)	c.3531C>A (p.Cys1177*)	
45	c.5882G>A (p.Gly1961Glu)	c.2099G>A (p.Trp700*)	
46	c.5882G>A (p.Gly1961Glu)	c.4734_4739delinsCC (p.Phe1579Glnfs*8)	
47	c.5882G>A (p.Gly1961Glu)	c.61C>T (p.Gln21*)	
48	c.5882G>A (p.Gly1961Glu)	c.2519T>G (p.Met840Arg)	

49	c.5882G>A (p.Gly1961Glu)	c.1245C>A (p.Asn415Lys)	
50	c.5882G>A (p.Gly1961Glu)	c.3056C>T (p.T1hr019Met)	
51	c.5882G>A (p.Gly1961Glu)	c.3531C>A (p.Cys1177*)	
52	c.5882G>A (p.Gly1961Glu)	c.3233G>A (p.Gly1078Glu)	
53	c.2908A>C (p.Thr970Pro)	c.3043T>A (p.Phe1015Ile)	c.61C>T (p.Gln21*)
54	c.3584_3585insGT (p.Thr1196*)	c.5381C>A (p.Ala1794Asp)	
55	c.4793C>A (p.Ala1598Asp)	c.4793C>A (p.Ala1598Asp)	
56	c.3531C>A (p.Cys1177*)	c.4793C>A (p.Ala1598Asp)	
57	c.5882G>A (p.Gly1961Glu)	c.3113C>T (p.Ala1038Val)	c.1622T>C (p.Leu541Pro)
58	c.1622T>C (p.Leu541Pro)	c.3113C>T (p.Ala1038Val)	c.2105T>C (p.Leu702Pro)
59	c.5882G>A (p.Gly1961Glu)	c.885del (p.Leu296Cysfs*4)	
60	c.2300T>A (p.Val767Asp)	c.6750del (p.Lys2250Asnfs*60)	
61	c.5714+5G>A (p.?)	c.6750del (p.Lys2250Asnfs*60)	
62	c.5381C>A (p.Ala1794Asp)	c.6072_6074del (p.Asp2024del)	
63	c.5882G>A (p.Gly1961Glu)	c.5961_5964del (p.Asp1988Profs*3)	
64	c.5917del (p.Val1973*)	c.5917del (p.Val1973*)	
65	c.5714+5G>A (p.?)	c.5917del (p.Val1973*)	
66	c.5018+2T>C (p.?)	c.5898+5del (p.?)	
67	c.2300T>A (p.Val767Asp)	c.3584_3585insGT (p.Thr1196*)	
68	c.327dup (p.Gln110Serfs*51)	c.327dup (p.Gln110Serfs*51)	
69	c.5882G>A (p.Gly1961Glu)	c.2099G>A (p.Trp700*)	
70	c.5882G>A (p.Gly1961Glu)	c.3933G>A (p.Gly978Asp)	
71	c.5882G>A (p.Gly1961Glu)	c.6282+1G>C (p.?)	
72	c.3323G>A (p.Arg1108His)	c.4297G>A (p.Val1433Ile)	c.5761G>A (p.Vla1921Met)
73	c.1622T>C (p.Leu541Pro)	c.3113C>T (p.Ala1038Val)	
74	c.5917del (p.Val1973*)	c.5917del (p.Val1973*)	
75	c.1622T>C (p.Leu541Pro)	c.3113C>T (p.Ala1038Val)	
76	c.5714+5G>A (p.?)	c.5917del (p.Val1973*)	
77	c.5882G>A (p.Gly1961Glu)	c.3261A>C (p.Glu1087Asp)	
78	c.667A>C (p.Lys223Gln)	c.3212C>T (p.Ser1071Leu)	c.3607G>A (p.Gly1203Arg)
79	c.634C>T (p.Arg212Cys)	c.3806T>C (p.Leu1269Pro)	
80	c.1714C>T (p.Arg572*)	c.4417C>A (p.Leu1473Met)	
81	c.247_250dup (p.Ser84Thrfs*16)	c.1666A>G (p.Met556Val)	
82	c.5882G>A (p.Gly1961Glu)	c.5425_5426del (p.Ile1809Tyrfs*8)	
83	c.5882G>A (p.Gly1961Glu)	6282+1G>C (p.?)	
84	c.2461T>A (p.Trp821Arg)	c.6229C>T (p.Arg2077Trp)	
85	c.5882G>A (p.Gly1961Glu)	c.6416G>T (p.Arg2139Leu)	
86	c.634C>T (p.Arg212Cys)	c.5087G>A (p.Ser1696Asn)	