Supplementary Table S1: Demographic, genetic and functional data of included subjects.

No.	Sex	Age [Years]	Onset [Years]	BCVA [LogMAR] OD	BCVA [LogMAR] OS	*ABCA4*-gene mutations* Allele 1	*ABCA4*-gene mutations* Allele 2
Group 1							
1	F	12	10	0.9	0.9	E49 c.6746C>A/p.(Ala2249Asp)	E49 c.6746C>A/p.(Ala2249Asp)
2	F	53	20	1.0	0.9	E42 c.5882G>A/p.(Gly1961Glu)	I36 c.5916+2T>C
3	F	17	17	1.0	0.3	E12 c.1622T>C/p.(Leu541Pro) E21 c.3113C>T/p.(Ala1038Val)	E17 c.2588G>C/p.(Gly863Ala) E40 c.5603A>T/p.(Asn1868Ile)
4	F	31	19	0.9	1.0	E3 c.206G>A/p.(Trp69*)	E42 c.5882G>A/p.(Gly1961Glu)
5	M	21	7	0.9	0.9	E6 c.656G>C/p.(Arg219Thr) E17 c.2588G>C/p.(Gly863Ala)	E10 c.1253T>C/p.(Phe418Ser) E40 c.5603A>T/p.(Asn1868Ile)
6	F	42	18	0.8	1.0	E6 c.768G>T/p.(Val256Val)	E42 c.5882G>A/p.(Gly1961Glu)
7	F	37	20	0.5	1.0	E42 c.5882G>A/p.(Gly1961Glu)	I36 c.5916+2T>C
8	F	34	31	0.7	0.1	E28 c.3195G>A/p.(Glu1399Lys) E44 c.6079C>T/p.(Leu2027Phe)	E43 c.5929G>A/p.(Gly1977Ser)
9	M	39	14	0.9	0.9	E12 c.1584C>A/p.(Tyr528*)	E42c.5882G>A/p.(Gly1961Glu)
10	M	54	42	1.1	0.2	E35 c.4918C>T/p.(Arg1640Trp)	E40 c.5693G>A/p.(Arg1898His)
11	F	15	6	0.5	0.3	E10 c.1309delC/p.(Gln437Argfs*12)	E42 c.5882G>A/p.(Gly1961Glu)
12	F	34	27	0.9	0.7	E15 c.2300T>A/p.(Val767Asp)	E42 c.5882G>A/p.(Gly1961Glu)
13	M	57	47	1.5	1.5	E27 c.3871C>T/p.(Glu1291*)	E42 c.5882G>A/p.(Gly1961Glu)
14	F	30	27	1.5	1.5	E42 c.5882G>A/p.(Gly1961Glu)	E43 c.5917delG/p.(Val1973*)
15	F	14	6	0.6	0.5	E3 c.194G>A/p.(Gly65Glu)	E17 c.2588G>C/p.(Gly963Ala)
16	F	36	30	0.9	0.7	E39 c.5512C>G/p.(His1838Asp) E42 c.5882G>A/p.(Gly1961Glu)	E42 c.5882G>A/p.(Gly1961Glu)
17	F	23	17	0.8	0.7	E10 c.1309delC/p.(Glu437Argfs*12)	E42 c.5882G>A/p.(Gly1961Glu)
18	F	10	6	0.3	0.3	E12 c.1622T>C/p.(Leu541Pro) E21 c.3113C>T/p.(Ala1038Val)	E13 Del.
19	F	36	33	0.8	0.8	E21 c.3113C>T/p.(Ala1038Val)	E28 c.4234C>T/p.(Gln1412*)
20	F	48	27	1.0	0.7	E39 c.5512C>G/p.(His1838Asp)	E42 c.5882G>A/p.(Gly1961Glu)
21	F	48	12	0.9	0.9	E39 c.5512C>G/p.(His1838Asp)	E42 c.5882G>A/p.(Gly1961Glu)
Group 2							
22	M	47	20	0.0	0.0	E12 c.1622T>C/p.(Leu541PRo) E21 c.3113C>T/p.(Ala1038Val)	E44 c.6089G>A/p.(Arg2030Gln)
23	M	27	25	0.4	0.2	E12 c.1719G>A/p.(Met573Ile)	
24	F	60	49	0.2	0.1	E36 c.5189G>A/p.(Trp1730*)	E40 c.5603A>T/p.(Asn1868Ile)
25	F	39	33	0.0	0.1	E8 c.1009T>C/p.(Phe337Leu) I13 c.1937+1G>A (IVS13+1G>A)	E40 c.5603A>T/p.(Asn1868Ile)
26	F	77	39	1.0	0.2	E3 c.1822T>A/p.(Phe608Ile) E40 c.5682G>C/p.(Leu1894Leu)	E40 c.5603A>T/p.(Asn1868Ile) I33 c.4773+48C>T
27	F	39	18	1.1	1.2	E12 c.1622T>C/p.(Leu541Pro) E21c.3113C>T/p.(Ala1038Val)	E17 c.2588G>C/p.(Gly863Ala) E40 c.5603A>T/p.(Asn1868Ile)

No.	Sex	Age [Years]	Onset [Years]	BCVA [LogMAR] OD	BCVA [LogMAR] OS	*ABCA4*-gene mutations* Allele 1	*ABCA4*-gene mutations* Allele 2
28	M	17	8	1.1	1.0	E12 c.1622T>C/p.(Leu541Pro) E21 c.3113C>T/p.(Ala1038Val)	E22 c.3212C>T/p.(Ser1071Leu)
29	F	61	14	1.0	1.0	E27 c.3871C>T/p.(Gln1291*)	E42 c.5882G>A/p.(Gly1961Glu)
30	M	50	49	0.0	0.6	E30 c.4527dupC/p.(Gln1513Prof*42) E31 c.4611G>A/p.(Thr1536Thr)	E40 c.5603A>T/p.(Asn1868Ile)
31	F	20	6	1.3	1.0	E43 c.5917delG/p.(Val1973*)	I40 c.5714+5G>A
32	M	61	49	0.1	0.1	E1 c.52C>T/p.(Arg18Trp) E17 c.1588G>A/p.(Gly863Ala) E40 c.5603A>T/p.(ASn1868Ile)	E12 c.1715G>A/p.(Arg572Gln) E19 c.2828G>A/p.(Arg943Gln)
33	F	63	49	0.4	0.1	E23 c.3468C>G/p.(Tyr1156*)	E36 c.5059A>T/p.(Ile1687Phe)
34	F	61	30	1.5	1.0	E23 c.3468C>G/p.(Tyr1156*)	E36 c.5059A>T/p.(Ile1687Phe)
35	F	41	30	0.8	0.8	E1 c.45G>A/p.(Trp15*)	E16 c.2401G>A/p.(Ala801Thr)
36	F	44	20	0.9	0.9	E28 c.4234C>T/p.(Gln1412*)	E42 c.5882G>A/p.(Gly1961Glu)
37	M	9	7	0.6	0.7	E12 c.1622T>C/p.(Leu541Pro) E21 c.3113C>T/p.(Ala1038Val)	
38	F	46	25	0.6	0.7	E16 c.2401G>A/p.(Ala801Thr)	E28 c.4234C>T/p.(Gln1412*)
Group 3							
39	F	39	30	1.2	0.9	E49 c.6746C>A/p.(Ala2249Asp)	E49 c.6746C>A/p.(Ala2249Asp)
40	M	29	6	1.2	1.4	E30 c.4462T>C/p.(Cys1488Arg)	E30 c.4462T>C/p.(Cys1488Arg)
41	F	18	9	1.1	1.0	E39 c.5549T>C/p.(Leu1850Pro)	E39 c.5509C>A/p.(Pro1837Thr)
42	F	28	9	1.2	1.1	E39 c.5549T>C/p.(Leu1850Pro)	E39 c.5509C>A/p.(Pro1837Thr)
43	F	39	12	1.0	0.8	E36 c.5172G>A/p.(Trp1724*)	E36 c.5172G>A/p.(Trp1724*)
44	F	50	44	0.1	0.1	E29 c.4347G>T/p.(Trp1449Cys)	

* = Nomenclature of mutations followed standards of the Human Genome Variation Society (HGVS)
No. = number, F = female, M = male, BCVA = best corrected visual acuity, LogMAR = logarithm of the minimum angle of resolution, OD = right eye, OS = left eye