Genetic Characteristics of 234 Italian Patients with Macular and Cone/Cone-Rod Dystrophy.

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Table S1. Genes included in custom NGS targeted sequencing panels.

NON-SYNDROMIC IRDs		CONE DYSTROPHY		MACULAR DYSTROPHY			STARGARDT DISEASE	
PANEL NAME (PERIOD OF USE)		PANEL NAME (PERIOD OF USE)	PANEL NAME (PERIOD OF USE)	PANEL NAME (PERIOD OF USE)	PANEL NAME (PERIOD OF USE)	PANEL NAME (PERIOD OF USE)	PANEL NAME (PERIOD OF USE)	PANEL NAME (PERIOD OF USE)
OCULAR (2017-2019)		OSS_NEW (2014-2018)	OCULAR (2017-2019)	OSS_NEW MD_Rev 0 (2014-2017)	OSS_NEW MD_Rev 1 (2017-2018)	OCULAR (2017-2019)	OSS_NEW (2014-2018)	OCULAR (2017-2019)
ABCA4	*LCA5*	*CNGA3*	*ABCA4*	*ABCA4*	*ABCA4*	*ABCA4*	*ABCA4*	*ABCA4*
ABCB6	*LRAT*	*RAB28*	*ADAM9*	*BEST1*	*BEST1*	*BEST1*	*ELOVL4*	*CNGB3*
ADAM9	*LRIT3*	*GUCY2D*	*AIPL1*	*EFEMP1*	*C8orf37*	*C8orf37*	*PROM1*	*ELOVL4*
AGBL5	*MAK*	*PROM1*	*C8orf37*	*ELOVL4*	*CRX*	*CDH3*	*PRPH2*	*PROM1*
AHI1	*MERTK*	*GUCA1A*	*CACNA1F*	*IMPG1*	*EFEMP1*	*CFH*		*PRPH2*
AIPL1	*NEK2*	*CERKL*	*CACNA2D4*	*IMPG2*	*ELOVL4*	*CFI*		
ARHGEF18	*NMNAT1*	*CACNA1F*	*CDHR1*	*PROM1*	*GUCA1A*	*CRX*		
ARL2BP	*NR2E3*	*SEMA4A*	*CEP78*	*PRPH2*	*IMPG1*	*DRAM2*		
ARL6	*NRL*	*CRX*	*CFAP410*	*RP1L1*	*IMPG2*	*EFEMP1*		
ATF6	*NYX*	*AIPL1*	*CNGA3*	*RPGR*	*PROM1*	*ELOVL4*		
BBS2	*OFD1*	*RPGRIP1*	*CRX*	*TIMP3*	*PRPH2*	*GUCA1A*		
BEST1	*OPA1*	*ABCA4*	*DRAM2*		*RAX2*	*IMPG1*		
C1QTNF5	*OPA3*	*PITPNM3*	*GUCA1A*		*RIMS1*	*IMPG2*		
C8orf37	*OTX2*	*PRPH2*	*GUCY2D*		*RLBP1*	*PRDM13*		
CA4	*PCARE*	*CNGB3*	*IFT81*		*RP1L1*	*PROM1*		
CABP4	*PDE6A*	*ADAM9*	*KCNV2*		*RPGR*	*PRPH2*		
CACNA1F	*PDE6B*	*RPGR*	*PDE6C*		*RPGR*	*RAX2*		
CACNA2D4	*PDE6C*	*CDHR1*	*PITPNM3*		*SEMA4A*	*RIMS1*		
CDH3	*PDE6G*	*RIMS1*	*POC1B*		*TIMP3*	*RLBP1*		
CDHR1	*PDE6H*	*RAX2*	*PROM1*		*TTC8*	*RP1L1*		
CEP290	*PITPNM3*		*PRPH2*			*RPGR*		
CEP78	*POC1B*		*RAB28*			*RPGR*		
CERKL	*POMGNT1*		*RAX2*			*SEMA4A*		
CFAP410	*PRCD*		*RIMS1*			*TIMP3*		
CFH	*PROM1*		*RPGR*			*TTC8*		
CFI	*PRPF3*		*RPGR*					
CHM	*PRPF31*		*RPGRIP1*					
CLN3	*PRPF4*		*SEMA4A*					
CLRN1	*PRPF6*		*TTLL5*					
CNGA1	*PRPF8*		*UNC119*					
CNGA3	*PRPH2*							
CNGB1	*RAB28*							
CNGB3	*RAX2*							
CRB1	*RBP3*							
CRX	*RD3*							
CTNNA1	*RDH12*							
DHDDS	*RDH5*							
DNM1L	*REEP6*							
DRAM2	*RGR*							
EFEMP1	*RHO*							
ELOVL4	*RIMS1*							
EYS	*RLBP1*							
FAM161A	*ROM1*							
FLVCR1	*RP1*							
FSCN2	*RP1L1*							
GDF6	*RP2*							
GNAT1	*RP9*							
GNAT2	*RPE65*							
GNB3	*RPGR*							
GPR179	*RPGRIP1*							
GRK1	*RS1*							
GRM6	*SAG*							
GUCA1A	*SEMA4A*							
GUCA1B	*SLC24A1*							
GUCY2D	*SLC7A14*							
HGSNAT	*SNRNP200*							
IDH3A	*SPATA7*							
IDH3B	*TIMP3*							
IFT140	*TMEM126A*							
IFT172	*TOPORS*							
IFT81	*TRPM1*							
IMPDH1	*TTC8*							
IMPG1	*TTLL5*							
IMPG2	*TUBB4B*							
IQCB1	*TULP1*							
KCNJ13	*UNC119*							
KCNV2	*USH2A*							
KIZ	*ZNF408*							
KLHL7	*ZNF513*							

Table S2. Demographic and genetic characteristics of MD probands.

ID	Sex	Age	ONSET	Gene	RefSeq	Exon/ intron	Nucleotide change	Amino acid change	Allele state	dbSNP rs	VARSOME	Transmission	REF
MD-1	F	31	25	*ABCA4*	NM_000350	ex34	c.4793C>A	p.(Ala1598Asp)	HET	rs61750155	P	AR	1
				ABCA4		ex40	c.5603A>T	p.(Asn1868Ile)	HOM	rs1801466	VUS/LP		2
MD-2	F	33	24	*ABCA4*	NM_000350	ex33	c.4710del	p.(Ile1571Serfs*10)	HET	rs281865381	LP	AR	3
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-3	F	21	8	*ABCA4*	NM_000350	ex43	c.5917del	p.(Val1973*)	HOM	rs61751389	P	AR	5
MD-4	F	46	17	*ABCA4*	NM_000350	ex47	c.6445C>T	p.(Arg2149*)	HET	rs61750654	P	AR	6
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-5	F	74	25	*ABCA4*	NM_000350	ex30	c.4354G>T	p.(Glu1452*)	HET	rs886044742	P	AR	7
				ABCA4		ex46	c.6316C>T	p.(Arg2106Cys)	HET	rs61750648	P		4
MD-6	F	16	10	*ABCA4*	NM_000350	ex34	c.4775G>A	p.(Gly1592Asp)	HET	rs1425552175	P	AR	8
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
				ABCA4		ex3	c.288C>A	p.(Asn96Lys)	HET	rs886039297	LP		9
MD-7	M	34	23	*ABCA4*	NM_000350	ex43	c.5917del	p.(Val1973*)	HOM	rs61751389	P	AR	5
MD-8	F	45	NA	*ABCA4*	NM_000350	ex6	*c.667A>C*	*p.(Lys223Gln)*	HET	rs147619585	VUS/LP	AR	9
				ABCA4		ex24	*c.3607G>A*	*p.(Gly1203Arg)*	HET	rs1064793011	P		9
						ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-9	M	20	NA	*ABCA4*	NM_000350	ex15	c.2300T>A	p.(Val767Asp)	HET	rs61751395	P	AR	10
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-10	M	43	20	*ABCA4*	NM_000350	ex1	c.61C>T	p.(Gln21*)	HET	rs770272033	P	AR	11
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-11	M	32	12	*ABCA4*	NM_000350	ex22	c.3295T>C	p.(Ser1099Pro)	HET	rs61750119	LP	AR	12
				ABCA4		ex22	c.3259G>A	p.(Glu1087Lys)	HET	rs61751398	P		4
MD-12	M	25	15	*ABCA4*	NM_000350	ex16	c.2461T>A	p.(Trp821Arg)	HET	rs61749433	P	AR	6
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
				ABCA4		ex16	c.2549A>G	p.(Tyr850Cys)	HET	rs143797418	LP		13
MD-13	M	30	6	*ABCA4*	NM_000350	ex6	c.634C>T	p.(Arg212Cys)	HET	rs61750200	P	AR	14
				ABCA4		ex38	c.5318C>T	p.(Ala1773Val)	HET	rs760549861	P		15
				ABCA4		ex5	c.514G>A	p.(Gly172Ser)	HET	rs61748532	VUS/LP		3
MD-14	M	50	37	*ABCA4*	NM_000350	int35	c.5018+2T>C	P.(?)	HET	rs61750562	P	AR	16
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-15	F	30	13	*ABCA4*	NM_000350	ex33	c.4734_4739delinsCC	p.(Phe1579Glnfs*8)	HET	NA	P	AR	NEW
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-16	F	22	15	*ABCA4*	NM_000350	ex12	c.1622T>C	p.(Leu541Pro)	HET	rs61751392	P	AR	17
				ABCA4		ex21	c.3113C>T	p.(Ala1038Val)	HET	rs61751374	P		4
						ex40	c.5692C>T	p.(Arg1898Cys)	HET	rs201357151	VUS/LP		18
MD-17	M	56	23	*ABCA4*	NM_000350	ex28	c.4217del	p.(His1406Profs*29)	HET	NA	P	AR	NEW
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-18	F	51	38	*ABCA4*	NM_000350	ex34	c.4793C>A	p.(Ala1598Asp)	HET	rs61750155	P	AR	1
				ABCA4		ex48	c.6721C>G	p.(Leu2241Val)	HET	rs61748521	LP		5
MD-19	M	64	NA	*ABCA4*	NM_000350	ex20	c.2971G>C	p. (Gly991Arg)	HET	rs61750129	P	AR	3
				ABCA4		ex22	c.3259G>A	p.(Glu1087Lys)	HET	rs61751398	P		4
MD-20	F	17	NA	*ABCA4*	NM_000350	ex19	c.2888G>T	p.(Gly963Val)	HET	NA	LP	AR	19
				ABCA4		ex47	c.6449G>A	p.(Cys2150Tyr)	HET	rs61751384	P		20

MD-21	M	53	20	*ABCA4*	NM_000350	ex9	c.1222C>T	p.(Arg408*)	HET	rs61748550	P	AR	21
				ABCA4		ex33	c.4710del	p.(Ile1571Serfs*10)	HET	rs281865381	LP		3
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-22	F	52	18	*ABCA4*	NM_000350	ex30	c.4462T>C	p.(Cys1488Arg)	HET	rs61750146	P	AR	6
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
				ABCA4		ex9	c.1140T>A	p.(Asn380Lys)	HET	rs61748549	LP		22
MD-23	F	19	9	*ABCA4*	NM_000350	ex3	c.288C>A	p.(Asn96Lys)	HOM	rs886039297	LP	AR	9
MD-24	F	20	16	*ABCA4*	NM_000350	ex38	c.5384T>G	p.(Leu1795*)	HET	NA	P	AR	NEW
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-25	M	29	1	*ABCA4*	NM_000350	ex21	c.3167A>T	p.(Asn1056Ile)	HET	NA	LP	AR	NEW
				ABCA4		ex22	c.3289A>G	p.(Arg1097Gly)	HET	NA	LP		NEW
MD-26	F	32	NA	*ABCA4*	NM_000350	ex22	c.3289A>T	p.(Arg1097*)	HET	rs886044731	P	AR	23
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-27	M	53	13	*ABCA4*	NM_000350	ex22	c.3210_3211dupGT	p.(Ser1071Cysfs*14)	HET	rs387906385	P	AR	4
				ABCA4		ex17	c.2640G>C	p.(Trp880Cys)	HET	NA	VUS/LP		24
MD-28	M	23	15	*ABCA4*	NM_000350	ex13	c.1819G>A	p.(Gly607Arg)	HET	rs61749412	LP	AR	5
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-29	F	14	10	*ABCA4*	NM_000350	ex1	c.61C>T	p.(Gln21*)	HET	rs770272033	P	AR	11
				ABCA4		ex19	c.2774G>A	p.(Trp925*)	HET	NA	P		25
MD-30	M	24	13	*ABCA4*	NM_000350	int30	c.4539+1G>T	p.(?)	HET	rs61751388	P	AR	26
				ABCA4		ex36	c.5087G>A	p.(Ser1696Asn)	HET	rs61750564	P		6
MD-31	M	45	NA	*ABCA4*	NM_000350	ex13	*c.1819G>A*	*p.(Gly607Arg)*	HET	rs61749412	LP	AR	5
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
				ABCA4		ex5	*c.466A>G*	*p.(Ile156Val)*	HET	rs62646863	LP		17
MD-32	F	42	35	*ABCA4*	NM_000350	ex44	c.6088C>T	p.(Arg2030*)	HET	rs61751383	P	AR	6
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-33	M	21	NA	*ABCA4*	NM_000350	ex19	c.2768del	p.(Pro923Glnfs*9)	HET	NA	LP	AR	27
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-34	M	45	10	*ABCA4*	NM_000350	ex4	c.428C>T	p.(Pro143Leu)	HET	rs62646860	P	AR	3
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-35	F	29	NA	*ABCA4*	NM_000350	int13	c.1937+1G>A	p.(?)	HET	rs61752401	P	AR	5
				ABCA4		int40	c.5714+5G>A	p.(Glu1863Leufs*33)	HET	rs61751407	P		28
MD-36	M	33	19	*ABCA4*	NM_000350	ex43	c.5908C>T	p.(Leu1970Phe)	HET	rs28938473	P	AR	17
				ABCA4		ex44	c.6089G>A	p.(Arg2030Gln)	HET	rs61750641	P		6
				ABCA4		ex27	c.4085G>T	p.(Arg1362Ile)	HET	NA	VUS/LP		NEW
MD-37	M	37	11	*ABCA4*	NM_000350	ex22	c.3289A>T	p.(Arg1097*)	HET	rs886044731	P	AR	23
				ABCA4		ex14	c.1957C>T	p.(Arg653Cys)	HET	rs61749420	P		9
MD-38	M	13	NA	*ABCA4*	NM_000350	int36	c.5196+1G>A	p.(?)	HET	rs61751377	P	AR	4
				ABCA4		ex35	c.4919G>A	p.(Arg1640Gln)	HET	rs61751403	P		10
MD-39	M	35	14	*ABCA4*	NM_000350	ex35	c.4956T>G	p.(Tyr1652*)	HET	rs61750561	LP	AR	12
				ABCA4		ex47	c.6446G>T	p.(Arg2149Leu)	HET	rs61750655	LP		12
MD-40	F	37	9	*ABCA4*	NM_000350	ex13	c.1844T>C	p.(Val615Ala)	HET	NA	LP	AR	29
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
MD-41	F	87	12	*ABCA4*	NM_000350	ex45	c.6184_6188del	p.(Val2062Argfs*33)	HET	NA	P	AR	NEW
				ABCA4		ex34	c.4793C>A	p.(Ala1598Asp)	HET	rs61750155	P		1
MD-42	F	48	24	*ABCA4*	NM_000350	ex43	c.5910_5912dup	p.(Leu1971dup)	HET	NA	LP	AR	NEW
				ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4

MD-43	F	64	52	*ABCA4* *ABCA4* *ABCA4*	NM_000350	ex19 ex42 ex16	c.2875A>G c.5882G>A c.2549A>G	p.(Thr959Ala) p.(Gly1961Glu) p.(Tyr850Cys)	HET HET HET	rs368846708 rs1800553 rs143797418	LP P LP	AR	NEW 4 13
MD-44	F	18	12	*ABCA4* *ABCA4*	NM_000350	ex43 ex42	c.5917del c.5882G>A	p.(Val1973*) p.(Gly1961Glu)	HET HET	rs61751389 rs1800553	P P	AR	5 4
MD-45	F	36	24	*ABCA4* *ABCA4*	NM_000350	ex43 ex42	c.5959_5964delinsTG c.5882G>A	p.(Gly1987*) p.(Gly1961Glu)	HET HET	NA rs1800553	P P	AR	NEW 4
MD-46	M	62	20	*ABCA4* *ABCA4*	NM_000350	ex19 ex42	c.2780C>T c.5882G>A	p.(Pro927Leu) p.(Gly1961Glu)	HET HET	NA rs1800553	VUS/LP P	AR	NEW 4
MD-47	F	11	6	*ABCA4* *ABCA4*	NM_000350	ex43 ex22	c.5917del c.3259G>A	p.(Val1973*) p.(Glu1087Lys)	HET HET	rs61751389 rs61751398	P P	AR	5 4
MD-48	M	57	12	*ABCA4* *ABCA4*	NM_000350	ex27 ex42	c.3999_4000insACCCCAGA GCCAGAGTGCCAGCCT c.5882G>A	p.(Pro1333_Pro1334insThrPro GluProGluCysGlnPro) p.(Gly1961Glu)	HET HET	NA rs1800553	LP P	AR	NEW 4
MD-49	M	23	18	*ABCA4* *ABCA4*	NM_000350	intron6 ex42	c.571-2A>T c.5882G>A	p.(?) p.(Gly1961Glu)	HET HET	rs61748534 rs1800553	P P	AR	30 4
MD-50	M	58	53	*ABCA4* *ABCA4*	NM_000350	ex14 ex48	c.2099G>A c.6721C>G	p.(Trp700*) p.(Leu2241Val)	HET HET	rs61749425 rs61748521	P LP	AR	12 5
MD-51	M	47	16	*ABCA4* *ABCA4*	NM_000350	ex45 ex42	c.6220G>A c.5882G>A	p.(Gly2074Ser) p.(Gly1961Glu)	HET HET	NA rs1800553	LP P	AR	27 4
MD-52	M	36	32	*ABCA4* *ABCA4*	NM_000350	int45 ex42	c.6282+1G>C c.5882G>A	p.(?) p.(Gly1961Glu)	HET HET	rs770453727 rs1800553	P P	AR	27 4
MD-53	M	60	53	*ABCA4* *ABCA4*	NM_000350	ex12 ex42	c.1757A>G c.5882G>A	p.(Asp586Gly) p.(Gly1961Glu)	HET HET	rs1553192682 rs1800553	LP P	AR	31 4
MD-54	F	34	19	*ABCA4* *ABCA4*	NM_000350	ex43 ex44	c.5942C>G c.6089G>A	p.(Thr1981Arg) p.(Arg2030Gln)	HET HET	rs752147871 rs61750641	P P	AR	7 6
MD-55	F	14	11	*ABCA4* *ABCA4*	NM_000350	intron6 ex42	c.571-2A>T c.5882G>A	p.(?) p.(Gly1961Glu)	HET HET	rs61748534 rs1800553	P P	AR	30 4
MD-56	F	16	12	*ABCA4* *ABCA4*	NM_000350	ex44 int40	c.6088C>T c.5714+5G>A	p.(Arg2030*) p.(Glu1863Leufs*33)	HET HET	rs61751383 rs61751407	P P	AR	6 28
MD-57	F	46	35	*ABCA4* *ABCA4*	NM_000350	ex19 ex42	c.2860T>G c.5882G>A	p.(Tyr954Asp) p.(Gly1961Glu)	HET HET	rs61749447 rs1800553	LP P	AR	12 4
MD-58	F	39	17	*ABCA4* *ABCA4*	NM_000350	ex35 ex34	c.4956T>G c.4793C>A	p.(Tyr1652*) p.(Ala1598Asp)	HET HET	rs61750561 rs61750155	LP P	AR	12 1
MD-59	M	37	6	*ABCA4* *ABCA4*	NM_000350	ex21 ex42	c.3064G>A c.5882G>A	p.(Glu1022Lys) p.(Gly1961Glu)	HET HET	rs61749459 rs1800553	P P	AR	12 4
MD-60	F	75	55	*ABCA4* *ABCA4*	NM_000350	ex3 ex42	c.247_250dup c.5882G>A	p.(Ser84Thrfs*16) p.(Gly1961Glu)	HET HET	rs1005271380 rs1800553	P P	AR	10 4
MD-61	M	14	7	*ABCA4* *ABCA4*	NM_000350	ex3 ex28	c.247_250dup c.4139C>T	p.(Ser84Thrfs*16) p.(Pro1380Leu)	HET HET	rs1005271380 rs61750130	P P	AR	10 6
MD-62	F	8	5	*BEST1*	NM_004183	ex7	c.718_720dup	p.(Val240dup)	HET	NA	LP	AD	NEW
MD-63	M	65	62	*BEST1*	NM_004183	ex4	c.436G>A	p.(Ala146Thr)	HET	rs1237501081	LP	AD	32
MD-64	M	44	NA	*BEST1*	NM_004183	ex4	c.278G>C	p.(Trp93Ser)	HET	NA	P	AD	33
MD-65	M	44	9	*BEST1*	NM_004183	ex6	c.652C>A	p.(Arg218Ser)	HET	rs281865238	P	AD	34

MD-66	F	15	8	*BEST1*	NM_004183	ex5	c.544T>C	p.(Trp182Arg)	HET	rs1565390925	LP	AD	33
MD-67	F	15	4	*BEST1*	NM_004183	ex2	c.44G>A	p.(Gly15Asp)	HET	rs766379510	LP	AD	35
MD-68	F	15	NA	*BEST1*	NM_004183	ex6	c.652C>T	p.(Arg218Cys)	HET	rs281865238	P	AD	34
MD-69	M	55	46	*CFI*	NM_000204	ex13	c.1573T>C	p.(Ser525Pro)	HET	NA	VUS/LP	AD	NEW
MD-70	F	47	8	*GUCY2D*	NM_000180	ex13	c.2480A>C	p.(Tyr827Ser)	HET	NA	VUS/LP	AD	NEW
MD-71	F	54	45	*PRPH2*	NM_000322	ex2	c.583C>T	p.(Arg195*)	HET	rs1322278463	P	AD	36
MD-72	F	49	38	*PRPH2*	NM_000322	ex2	c.652T>C	p.(Ser218Pro)	HET	rs1582764878	LP	AD	37
MD-73	F	50	44	*PRPH2*	NM_000322	ex2	c.668T>A	p.(Ile223Asn)	HET	NA	P	AD	38
MD-74	M	57	39	*PRPH2*	NM_000322	ex1	c.515G>A	p.(Arg172Gln)	HET	rs61755793	P	AD	39
MD-75	M	56	NA	*PRPH2*	NM_000322	ex1	c.499G>A	p.(Gly167Ser)	HET	rs527236098	P	AD	40
MD-76	M	54	17	*PRPH2*	NM_000322	ex1	c.514C>T	p.(Arg172Trp)	HET	rs61755792	P	AD	39
MD-77	F	53	48	*PRPH2*	NM_000322	ex1	c.499G>A	p.(Gly167Ser)	HET	rs527236098	P	AD	40
MD-78	F	50	41	*PRPH2*	NM_000322	ex1	c.515G>A	p.(Arg172Gln)	HET	rs61755793	P	AD	39
MD-79	M	50	46	*PRPH2*	NM_000322	ex1	c.499G>A	p.(Gly167Ser)	HET	rs527236098	P	AD	40
MD-80	F	45	34	*RP1L1*	NM_178857	ex2	c.563T>C	p.(Leu188Pro)	HET	NA	VUS/LP	AD	NEW

Legend: AD, autosomal dominant; AR, autosomal recessive; HET, heterozygous; HOM, homozygous; F, female; M, male; ex, exon; int, intron; in **bold**, segregation performed; in *italics*, *in cis* variants; NA, not available; UNK, unknown; VUS, variant of unknown significance; LP, likely pathogenic; P, pathogenic.

Table S3. Demographic and genetic characteristics of CD and CRD probands.

ID	Sex	Age	Genetic class	ONSET	Gene	RefSeq	Exon/ intron	Nucleotide change	Amino acid change	Allele state	dbSNP rs	VARSOME	Transmission	REF
CD/CRD-1	F	66	CD	8	*ABCA4*	NM_000350	ex34	c.4793C>A	p.(Ala1598Asp)	HET	rs61750155	P	AR	1
					ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
					ABCA4		ex3	c.288C>A	p.(Asn96Lys)	HET	rs886039297	LP		9
CD/CRD-2	F	44	CRD	NA	*ABCA4*	NM_000350	ex10	c.52C>T	p.(Arg18Trp)	HET	rs121909205	P	AR	14
					ABCA4		ex15	c.2300T>A	p.(Val767Asp)	HET	rs61751395	P		10
CD/CRD-3	F	67	CRD	NA	*ABCA4*	NM_000350	ex12	c.1622T>C	p.(Leu541Pro)	HET	rs61751392	P	AR	17
					ABCA4		ex21	c.3113C>T	p.(Ala1038Val)	HET	rs61751374	P		4
					ABCA4		ex11	c.1522C>T	p.(Arg508Cys)	HET	rs138157885	VUS/LP		41
					ABCA4		ex37	c.5261A>G	p.(Tyr1754Cys)	HET	NA	VUS/LP		27
CD/CRD-4	M	65	CRD	15	*ABCA4*	NM_000350	ex34	c.4793C>A	p.(Ala1598Asp)	HET	rs61750155	P	AR	1
					ABCA4		ex31	c.4577C>T	p.(Thr1526Met)	HET	rs61750152	P		17
CD/CRD-5	F	25	CD	17	*ABCA4*	NM_000350	ex43	c.5961_5964del	p.(Asp1988Profs*3)	HET	rs61750640	P	AR	12
					ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
					ABCA4		ex43	c.5959G>T	p.(Gly1987Trp)	HET	NA	LP		NEW
CD/CRD-6	F	38	CD	11	*ABCA4*	NM_000350	ex29	c.4346G>A	p.(Trp1449*)	HET	rs61750143	P	AR	6
					ABCA4		ex25	c.3806T>C	p.(Leu1269Pro)	HET	NA	VUS/LP		27
CD/CRD-7	M	67	CRD	16	*ABCA4*	NM_000350	ex14	c.1957C>T	p.(Arg653Cys)	HET	rs61749420	P	AR	9
					ABCA4		ex37	c.5285C>A	p.(Ala1762Asp)	HET	rs121909206	LP		9
CD/CRD-8	F	8	CD	5	*ABCA4*	NM_000350	int35	c.5018+2T>C	p.(?)	HET	rs61750562	P	AR	16
					ABCA4		ex47	c.6449G>A	p.(Cys2150Tyr)	HET	rs61751384	P		20
CD/CRD-9	M	66	CD	50	*ABCA4*	NM_000350	ex1-5	c.1-?_571-?del	p.(?)	HET	NA	P	AR	NEW
					ABCA4		ex38	c.5329A>T	p.(Met1777Leu)	HET	rs375184282	VUS/LP		27
CD/CRD-10	M	16	CD	5	*ABCA4*	NM_000350	ex12	*c.1622T>C*	*p.(Leu541Pro)*	HET	rs61751392	P	AR	17
					ABCA4		int9	c.1239+1G>C	p.(?)	HET	rs765707028	P		
					ABCA4		ex21	*c.3113C>T*	*p.(Ala1038Val)*	HET	rs61751374	P		4
CD/CRD-11	F	42	CD	21	*ABCA4*	NM_000350	int5	c.571-1G>T	p.(?)	HET	rs61748533	P	AR	12
					ABCA4		ex36	c.5087G>A	p.(Ser1696Asn)	HET	rs61750564	P		6
CD/CRD-12	F	16	CRD	13	*ABCA4*	NM_000350	int36	c.5196+1G>A	p.(?)	HET	rs61751377	P	AR	4
					ABCA4		*ex25*	c.3813G>C	p.(Glu1271Asp)	HET	NA	P		
CD/CRD-13	F	53	CRD	20	*ABCA4*	NM_000350	ex4	c.428C>T	p.(Pro143Leu)	HET	rs62646860	P	AR	3
					ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
CD/CRD-14	F	52	CD	5	*ABCA4*	NM_000350	ex4	c.428C>T	p.(Pro143Leu)	HOM	rs62646860	P	AR	3
CD/CRD-15	M	28	CRD	6	*ABCA4*	NM_000350	ex43	c.5917del	p.(Val1973*)	HET	rs61751389	P	AR	5
					ABCA4		ex22	c.3259G>A	p.(Glu1087Lys)	HET	rs61751398	P		4
CD/CRD-16	M	40	CD	12	*ABCA4*	NM_000350	int35	c.5018+2T>C	p.(?)	HET	rs61750562	P	AR	16
					ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
					ABCA4		ex16	c.2549A>G	p.(Tyr850Cys)	HET	rs143797418	LP		13
CD/CRD-17	M	24	CD	11	*ABCA4*	NM_000350	ex8	c.1085_1086del	p.(Tyr362*)	HET	NA	P	AR	NEW
					ABCA4		ex17	c.2624T>C	p.(Leu875Pro)	HET	NA	VUS/LP		NEW
CD/CRD-18	F	63	CRD	9	*ABCA4*	NM_000350	ex47	c.6445C>T	p.(Arg2149*)	HET	rs61750654	P	AR	6
					ABCA4		ex23	c.(3192-90_3418)_(3418_3562)dup	p.(?)	HET	NA	P		NEW
CD/CRD-19	F	52	CRD	30	*ABCA4*	NM_000350	int13	c.1937+1G>A	p.(?)	HET	rs61752401	P	AR	5
					ABCA4		ex42	c.5882G>A	p.(Gly1961Glu)	HET	rs1800553	P		4
CD/CRD-20	M	59	CD	12	*ABCA4*	NM_000350	ex43	c.5959_5964delinsTG	p.(Gly1987*)	HET	NA	P	AR	NEW
					ABCA4		int40	c.5714+5G>A	p.(Glu1863Leufs*33)	HET	rs61751407	P		28
CD/CRD-21	M	9	CRD	1	*ADAM9*	NM_003816	ex8	c.725T>G	p.(Leu242Arg)	HOM	NA	VUS/LP	AR	NEW
CD/CRD-22	M	29	CRD	1	*BEST1*	NM_004183	ex4	c.318dup	p.(Met107Hisfs*125)	HET	NA	P	AD	NEW
CD/CRD-23	M	47	CRD	19	*CACNA1F*	NM_005183	int23	c.2874-1G>C	p.(?)	HEM	NA	P	XL	NEW
CD/CRD-24	M	22	CRD	3	*CACNA1F*	NM_005183	ex23	c.2804_2806del	p.(Phe935del)	HEM	rs782068089	VUS/LP	XL	NEW
CD/CRD-25	F	48	CRD	44	*CDHR1*	NM_033100	ex8	c.783G>A	p.(Pro261=)	HET	rs147346345	VUS/LP	AR	42
					CDHR1		ex13	c.1373T>A	p.(Val458Asp)	HET	rs760942217	VUS/LP		43
CD/CRD-26	M	52	CRD	28	*CDHR1*	NM_033100	ex17	c.2522_2528del	p.(Ile841Serfs*119)	HET	rs794727197	P	AR	44
					CDHR1		ex8	c.783G>A	p.(Pro261=)	HET	rs147346345	VUS/LP		42

CD/CRD-27	M	70	CRD	62	*CFH*	NM_000186	ex16	c.2440C>T	p.(Pro814Ser)	HET	NA	VUS/LP	AD	NEW
CD/CRD-28	F	39	CD	1	*CNGA3*	NM_001298	ex7	c.572G>A	p.(Cys191Tyr)	HET	rs761554853	P	AR	45
					CNGA3		ex8	c.848G>A	p.(Arg283Gln)	HET	rs104893614	P		45
CD/CRD-29	F	15	CD	NA	*CNGA3*	NM_001298	ex8	c.1279C>T	p.(Arg427Cys)	HET	rs141386891	P	AR	46
					CNGA3		ex8	c.940_942del	p.(Ile314del)	HET	rs777878533	P		46
CD/CRD-30	F	39	CD	7	*CNGB3*	NM_019098	ex10	c.1148del	p.(Thr383Ilefs*13)	HOM	rs397515360	P	AR	47
CD/CRD-31	F	44	CD	6	*CNGB3*	NM_019098	ex2	c.143del	p.(Gly48Valfs*35)	HOM	NA	LP	AR	NEW
CD/CRD-32	F	29	CRD	6	*CRB1*	NM_201253	ex7	c.2149G>T	p.(Gly717Cys)	HET	NA	LP	AD	NEW
CD/CRD-33	F	60	CD	52	*CRX*	NM_000554	ex3	c.166G>A	p.(Ala56Thr)	HET	rs61748437	LP	AD	48
CD/CRD-34	M	68	CRD	56	*CRX*	NM_000554	ex4	c.329del	p.(Gly110Alafs*77)	HET	rs761108522	P	AD	NEW
CD/CRD-35	F	56	CRD	20	GUCA1A	NM_000409	ex4	c.312_313delinsGC	p.(Asn104_Gly105delinsLysArg)	HET	NA	VUS/LP	AD	NEW
CD/CRD-36	M	51	CRD	18	*GUCY2D*	NM_000180	ex13	c.2512C>T	p.(Arg838Cys)	HET	rs61750172	P	AD	49
CD/CRD-37	M	43	CD	25	*GUCY2D*	NM_000180	ex13	c.2512 C>T	p.(Arg838Cys)	HOM	rs61750172	P	AR	49
CD/CRD-38	F	12	CD	5	*GUCY2D*	NM_000180	ex13	c.2512C>T	p.(Arg838Cys)	HET	rs61750172	P	AD	49
CD/CRD-39	M	77	CD	65	*GUCY2D*	NM_000180	ex2	c.286T>C	p.(Phe96Leu)	HET	NA	VUS/LP	AD	NEW
CD/CRD-40	M	26	CD	3	*GUCY2D*	NM_000180	ex13	c.2546C>G	p.(Thr849Arg)	HET	NA	LP	AD	NEW
CD/CRD-41	F	47	CRD	35	*IMPG2*	NM_016247	ex2	c.283G>C	p.(Glu95Gln)	HET	rs1198094357	VUS/LP	AD	NEW
CD/CRD-42	F	39	CD	14	*KCNV2*	NM_133497	ex2	c.1381G>T	p.(Gly461*)	HOM	rs149648640	P	AR	50
CD/CRD-43	F	30	CD	3	*KCNV2*	NM_133497	ex2	c.1427T>G	p.(Leu476Arg)	HOM	rs796658305	LP	AR	NEW
CD/CRD-44	M	40	CD	31	*PDE6C*	NM_006204	int20	c.2367+1_2367+5del	p.(?)	HET	rs796051871	P	AR	NEW
					PDE6C		ex17	c.2087C>T	p.(Thr696Met)	HET	rs41290222	VUS/LP		NEW
CD/CRD-45	M	57	CRD	25	*POC1B*	NM_172240	ex4	c.317G>C	p.(Arg106Pro)	HET	rs76216585	LP	AR	51
					POC1B		ex6	c.587C>T	p.(Pro196Leu)	HET	NA	VUS/LP		NEW
CD/CRD-46	M	68	CD	30	*PRPH2*	NM_000322	ex2	c.621C>A	p.(Asp207Glu)	HET	NA	LP	AD	NEW
CD/CRD-47	F	52	CD	40	*PRPH2*	NM_000322	ex1	c.514C>T	p.(Arg172Trp)	HET	rs61755792	P	AD	39
CD/CRD-48	F	63	CD	10	*PRPH2*	NM_000322	ex1	c.568A>G	p.(Lys190Glu)	HET	NA	VUS/LP	AD	NEW
CD/CRD-49	F	64	CD	7	*RAB28*	NM_001017979	ex4	c.321G>A	p.(Trp107*)	HET	NA	P	AD	52
CD/CRD-50	M	62	CRD	16	*RPGR*	NM_001034853	ex15	c.3178_3179del	p.(Glu1060Argfs*18)	HEM	rs771214648	P	XL	53
CD/CRD-51	M	46	CD	25	*RPGR*	NM_001034853	ex15	c.3317dup	p.(Ser1107Valfs*4)	HEM	rs886041376	P	XL	54
CD/CRD-52	M	37	CRD	24	*RPGR*	NM_001034853	ex15	c.2236_2237del	p.(Glu746Argfs*23)	HEM	rs1555961852	P	XL	55
CD/CRD-53	M	24	CRD	12	*TTLL5*	NM_015072	ex13	c.1060G>A	p.(Val354Met)	HOM	rs781509883	VUS/LP	AR	NEW
CD/CRD-54	M	71	CRD	60	*TTLL5*	NM_015072	ex17	c.1442G>C	p.(Gly481Ala)	HOM	rs771482604	VUS/LP	AR	NEW
CD/CRD-55	M	41	CRD	34	*TTLL5*	NM_015072	ex10	c.800T>C	p.(Leu267Pro)	HOM	NA	VUS/LP	AR	NEW
CD/CRD-56	F	7	CRD	1	*TULP1*	NM_003322	ex10	c.901C>T	p.(Gln301*)	HET	rs201070350	P	AR	56
					TULP1		ex8	c.822G>T	p.(Lys274Asn)	HET	NA	P		NEW

Legend: AD, autosomal dominant; AR, autosomal recessive; XL, X-linked; HET, heterozygous; HOM, homozygous; HEM, hemizygous; F, female; M, male; ex, exon; int, intron; in **bold**, segregation performed; in *italics*, *in cis* variants; NA, not available; UNK, unknown; VUS, variant of unknown significance; LP, likely pathogenic; P, pathogenic.

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