Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification	Segregation
MD-0012	STGD	ABCA4	NM_000350	25;42	c.[3758C>T;5882G>A]	p.[Thr1253Met; Gly1961Glu]	27	c.3943C>T	p.(Gln1315*)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0014	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0015	STGD	ABCA4	NM_000350	17	c.[2588G>C;3163C>T]	p.[Gly863Ala,Gly863del;Arg1055Trp]	19	c.2888del	p.(Gly963Alafs*14)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0017	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	46	c.6320G>C	p.(Arg2107Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0022	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	45	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0031	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	35	c.4926C>G	p.(Ser1642Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0038	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	33	c.4739del	p.(Leu1580*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0039	STGD	ABCA4	NM_000350	6	c.699_768+341del	p.(Gln234Phefs*5)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0040	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	27	c.3943C>T	p.(Gln1315*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0047	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0051	STGD	ABCA4	NM_000350	22	c.3292C>T	p.(Arg1098Cys)	35	c.4855T>C	p.(Phe1619Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0057	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0060	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0061	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0062	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0064	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0065	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	IVS7	c.859-506G>C	p.[Phe287Thrfs*32,=]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0066	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0072	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	22	c.3287C>T	p.(Ser1096Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0076	STGD	ABCA4	NM_000350	6	c.768G>T	p.(Val256Val)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0078	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	48	c.6559C>T	p.(Gln2187*)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0079	CCRD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	19	c.2888del	p.(Gly963Alafs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0081	STGD	ABCA4	NM_000350	29	c.4297G>A	p.(Val1433Ile)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0082	STGD	ABCA4	NM_000350	23	c.3364G>A	p.(Glu1122Lys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0084	STGD	ABCA4	NM_000350	47	c.6410G>A	p.(Cys2137Tyr)	47	c.6410G>A	p.(Cys2137Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0086	CCRD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	19	c.2888del	p.(Gly963Alafs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0088	STGD	ABCA4	NM_000350	28	c.4139C>T	p.(Pro1380Leu)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0090	STGD	ABCA4	NM_000350	IVS22	c.3329-2A>T	p.?	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0094	OtherMD	ABCA4	NM_000350	14	c.1964T>G	p.(Phe655Cys)	16	c.2481del	p.(Thr829Argfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0096	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	28	c.[4222T>C;4918C>T]	p.[Trp1408Arg;Arg1640Trp]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0110	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0111	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0116	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	30	c.4469G>A	p.(Cys1490Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0119	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0125	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	40	c.5630_5644dup	p.(Lys1877_Ala1881dup)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0126	CCRD	ABCA4	NM_000350	43	c.5929G>A	p.(Gly1977Ser)	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0128	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0133	CCRD	ABCA4	NM_000350	1	c.32T>C	p.(Leu11Pro)	19	c.2888del	p.(Gly963Alafs*14)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0135	STGD	ABCA4	NM_000350	8	c.1029dup	p.(Asn344*)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0137	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0138	STGD	ABCA4	NM_000350	17	c.2588G>C	p.(Gly863Ala)	30	c.4537dup	p.(Gln1513Profs*42)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0139	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0146	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	44	c.6140T>A	p.(Ile2047Asn)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0153	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	42	c.5881G>A	p.(Gly1961Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0155	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0158	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	30	c.4537del	p.(Gln1513Argfs*13)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0162	STGD	ABCA4	NM_000350	IVS6-Ex6	c.699_768+341del	p.(Gln234Phefs*5)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0163	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	36	c.5172G>T	p.(Trp1724Cys)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0164	STGD	ABCA4	NM_000350	6	c.700C>T	p.(Gln234*)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0165	STGD	ABCA4	NM_000350	22;40	c.[3210_3211dup;5603A>T]	p.[Ser1071Cysfs*14;Asn1868Ile]	40	c.5603A>T	p.(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0166	STGD	ABCA4	NM_000350	IVS6-Ex6	c.699_768+341del	p.(Gln234Phefs*5)	46	c.6320G>A	p.(Arg2107His)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0167	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	22	c.3281C>G	p.(Pro1094Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0168	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0170	STGD	ABCA4	NM_000350	9	c.1222C>T	p.(Arg408*)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0173	STGD	ABCA4	NM_000350	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0174	CCRD	ABCA4	NM_000350	25	c.4918C>T	p.(Arg1640Trp)	IVS44	c.6147+2T>A	p.?	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0176	CCRD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	45	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0178	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	46	c.6320G>C	p.(Arg2107Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0181	STGD	ABCA4	NM_000350	22	c.3323G>A	p.(Arg1108His)	IVS38	c.5460+5G>A	p.(Trp1772Argfs*9)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0183	STGD	ABCA4	NM_000350	43	c.5929G>A	p.(Gly1977Ser)	44	c.6079C>T	p.(Leu2027Phe)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0187	STGD	ABCA4	NM_000350	28	c.4139C>T	p.(Pro1380Leu)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0190	CCRD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	38	c.5395A>G	p.(Asn1799Asp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0191	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0194	STGD	ABCA4	NM_000350	19	c.2791G>A	p.(Val931Met)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0196	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	40	c.5644A>G	p.(Met1882Val)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0198	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0200	STGD	ABCA4	NM_000350	14	c.2041C>T	p.(Arg681*)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0203	STGD	ABCA4	NM_000350	35	c.4918C>T	p.(Arg1640Trp)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0206	CCRD	ABCA4	NM_000350	35;36	c.[4926C>G;5044_5058del]	p.[Ser1642Arg;Val1682_Val1686del]	35;36	c.[4926C>G;5044_5058del]	p.[Ser1642Arg;Val1682_Val1686del]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0207	STGD	ABCA4	NM_000350	30	c.4537dup	p.(Gln1513Profs*42)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0213	CCRD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	13	c.1804C>T	p.(Arg602Trp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0215	STGD	ABCA4	NM_000350	36	c.5044_5058del	p.(Val1682_Val1686del)	47	c.6449G>A	p.(Cys2150Tyr)	AR	AR	Stargardt disease/cone-rod dystrophy		NA

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification	Segregation
MD-0216	STGD	ABCA4	NM_000350	31	c.4577C>T	p.(Thr1526Met)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0218	STGD	ABCA4	NM_000350	19	c.2894A>G	p.(Asn965Ser)	19	c.2894A>G	p.(Asn965Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0225	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	48	c.6559C>T	p.(Gln2187*)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0227	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0238	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0240	STGD	ABCA4	NM_000350	15	c.2285C>A	p.(Ala762Glu)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0242	STGD	ABCA4	NM_000350	12	c.1715G>C	p.(Arg572Pro)	37	c.5242G>A	p.(Gly1748Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0244	STGD	ABCA4	NM_000350	14	c.1957C>T	p.(Arg653Cys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0245	STGD	ABCA4	NM_000350	IVS38	c.4253+5G>A	p.(Ile1377Hisfs*3)	33	c.4672G>A	p.(Gly1558Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0247	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	47	c.6410G>A	p.(Cys2137Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0249	STGD	ABCA4	NM_000350	36	c.5044_5058del	p.(Val1682_Val1686del)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0252	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23;48	c.[3386G>T;6718A>G]	p.[Arg1129Leu;Thr2240Ala]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0254	STGD	ABCA4	NM_000350	5	c.454C>T	p.(Arg152*)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0260	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0262	STGD	ABCA4	NM_000350	13	c.1819G>A	p.(Gly607Arg)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0264	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	47	c.6449G>A	p.(Cys2150Tyr)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0266	CCRD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	45	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0267	STGD	ABCA4	NM_000350	19	c.2791G>A	p.(Val931Met)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0270	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0277	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0279	STGD	ABCA4	NM_000350	15	c.2300T>A	p.(Val767Asp)	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0280	STGD	ABCA4	NM_000350	12	c.1648G>A	p.(Gly550Arg)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0281	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	36	c.5044_5058del	p.(Val1682_Val1686del)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0283	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0284	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0286	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	IVS36	c.5196+1056A>G	p.(Met1733Valfs*2)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0287	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0288	STGD	ABCA4	NM_000350	8	c.982G>T	p.(Glu328*)	22	c.3322C>T	p.(Arg1108Cys)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0290	CCRD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	35	c.4919G>A	p.(Arg1640Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0291	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0293	OtherMD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	38	c.5395A>G	p.(Asn1799Asp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0295	CCRD	ABCA4	NM_000350	43	c.5917del	p.(Val1973*)	43	c.5917del	p.(Val1973*)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0298	STGD	ABCA4	NM_000350	8	c.982G>T	p.(Glu328*)	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0299	CCRD	ABCA4	NM_000350	1	c.52C>T	p.(Arg18Trp)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0300	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	39	c.5549T>C	p.(Leu1850Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0301	STGD	ABCA4	NM_000350	13	c.1792G>A	p.(Val598Met)	43	c.5914G>A	p.(Gly1972Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0302	CCRD	ABCA4	NM_000350	12	c.[1622T>C;3113C>T]	p.[Leu541Pro;Ala1038Val]	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0305	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	22	c.3323G>A	p.(Arg1108His)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0307	STGD	ABCA4	NM_000350	28	c.4200C>A	p.(Tyr1400*)	IVS35	c.5018+2T>C	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0308	STGD	ABCA4	NM_000350	12	c.1592A>G	p.(Glu531Gly)	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0317	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0318	STGD	ABCA4	NM_000350	27	c.4069G>A	p.(Ala1357Thr)	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0323	STGD	ABCA4	NM_000350	27	c.3899G>A	p.(Arg1300Gln)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0324	STGD	ABCA4	NM_000350	1	c.3G>A	p.(Met1Ile)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0326	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	43	c.5929G>A	p.(Gly1977Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0329	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0331	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0334	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0335	STGD	ABCA4	NM_000350	27	c.3943C>T	p.(Gln1315*)	IVS36	c.5196+1137G>A	p.[Met1733Glufs*78,=]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0336	CCRD	ABCA4	NM_000350	44	c.6088C>T	p.(Arg2030*)	44	c.6088C>T	p.(Arg2030*)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0340	STGD	ABCA4	NM_000350	6	c.671del	p.(Thr224Argfs*17)	12	c.1633A>T	p.(Asn545Tyr)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0341	STGD	ABCA4	NM_000350	30	c.4519G>A	p.(Gly1507Arg)	38	c.5377G>A	p.(Val1793Met)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0342	STGD	ABCA4	NM_000350	9	c.1222C>T	p.(Arg408*)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0345	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0349	STGD	ABCA4	NM_000350	20	c.2966T>C	p.(Val989Ala)	27	c.3988G>T	p.(Glu1330*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0353	STGD	ABCA4	NM_000350	15	c.2285C>A	p.(Ala762Glu)	45	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0354	STGD	ABCA4	NM_000350	8	c.1025_1038del	p.(Asp342Glyfs*6)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0359	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	38	c.5395A>G	p.(Asn1799Asp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0360	STGD	ABCA4	NM_000350	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0364	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	28	c.4139C>T	p.(Pro1380Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0370	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0373	STGD	ABCA4	NM_000350	16	c.2401G>A	p.(Ala801Thr)	23	c.3364G>A	p.(Glu1122Lys)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0388	STGD	ABCA4	NM_000350	45	c.6230G>A	p.(Arg2077Gln)	47	c.6449G>A	p.(Cys2150Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0390	CCRD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0392	CCRD	ABCA4	NM_000350	46	c.6320G>C	p.(Arg2107Pro)	46	c.6320G>C	p.(Arg2107Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0394	STGD	ABCA4	NM_000350	5	c.454C>T	p.(Arg152*)	14	c.2023G>A	p.(Val675Ile)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0395	STGD	ABCA4	NM_000350	8	c.950del	p.(Gly317Alafs*57)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0400	STGD	ABCA4	NM_000350	38	c.5451G>T	p.(Glu1817Asp)	38	c.5451G>T	p.(Glu1817Asp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0401	STGD	ABCA4	NM_000350	19	c.2791G>A	p.(Val931Met)	11Del	c.1357_1554del	p.(Asp453_Glu518del)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0402	STGD	ABCA4	NM_000350	12;21	c.[1622T>C;3113C>T]	p.[Leu541Pro; Ala1038Val]	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0408	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	47	c.6449G>A	p.(Cys2150Tyr)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0410	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0412	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	28	c.4139C>T	p.(Pro1380Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-0414	STGD	ABCA4	NM_000350	21	c.3113C>T	p.(Ala1038Val)	35	c.4918C>T	p.(Arg1640Trp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0416	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0420	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	48	c.6563T>C	p.(Phe2188Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0423	STGD	ABCA4	NM_000350	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0427	STGD	ABCA4	NM_000350	13	c.1832T>C	p.(Leu611Pro)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0428	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0431	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0432	STGD	ABCA4	NM_000350	5;22	c.[560G>A;3210_3211dup]	p.[Arg187His;Ser1071Cysfs*14]	14	c.2041C>T	p.(Arg681*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0433	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0435	STGD	ABCA4	NM_000350	12	c.1609C>T	p.(Arg537Cys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0437	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0439	STGD	ABCA4	NM_000350	8	c.871C>G	p.(Pro291Ala)	23	c.3364G>A	p.(Glu1122Lys)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0445	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0450	STGD	ABCA4	NM_000350	8	c.1025_1038del	p.(Asp342Glyfs*6)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0451	CCRD	ABCA4	NM_000350	IVS15	c.2382+5G>C	p.[=,His721_Val794del]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0452	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	45	c.6229C>T	p.(Arg2077Trp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0456	STGD	ABCA4	NM_000350	13;29	c.[1819G>A;4283C>T]	p.[Gly607Arg;Thr1428Met]	41	c.5761G>A	p.(Val1921Met)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0460	STGD	ABCA4	NM_000350	IVS36	c.5196+1137G>A	p.[Met1733Glufs*78,=]	Ex6-IVS6	c.699_768+341del	p.(Gln234Phefs*5)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0463	STGD	ABCA4	NM_000350	8	c.1035T>G	p.(Tyr345*)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0464	STGD	ABCA4	NM_000350	17	c.2588G>C	p.[Gly863Ala,Gly863del]	IVS44	c.6147+2T>A	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0465	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0466	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0467	CCRD	ABCA4	NM_000350	12	c.1622T>C	p.(Leu541Pro)	43	c.5917del	p.(Val1973*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0474	STGD	ABCA4	NM_000350	12	c.1622T>C	p.(Leu541Pro)	28	c.4234C>T	p.(Gln1412*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0479	STGD	ABCA4	NM_000350	12;12	c.1751_1753delinsAT	p.(Ile584Asnfs*65)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0481	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	39;42	c.[5512C>G;5882G>A]	p.[His1838Asp;Gly1961Glu]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0482	STGD	ABCA4	NM_000350	14	c.2057T>C	p.(Leu686Ser)	27	c.4069G>A	p.(Ala1357Thr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0486	STGD	ABCA4	NM_000350	17	c.2588G>C	p.[Gly863Ala,Gly863del]	45	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0493	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0494	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0497	STGD	ABCA4	NM_000350	1	c.52C>T	p.(Arg18Trp)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0498	STGD	ABCA4	NM_000350	14	c.2041C>T	p.(Arg681*)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0502	CCRD	ABCA4	NM_000350	5	c.560G>A	p.(Arg178His)	27	c.3871C>T	p.(Gln1291*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0506	STGD	ABCA4	NM_000350	22	c.3292C>T	p.(Arg1098Cys)	35	c.4919G>A	p.(Arg1640Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0509	STGD	ABCA4	NM_000350	17	c.2588G>C	p.[Gly863Ala,Gly863del]	44	c.6118C>T	p.(Arg2040*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0512	STGD	ABCA4	NM_000350	30	c.4537dup	p.(Gln1513Profs*42)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0514	STGD	ABCA4	NM_000350	22	c.3323G>A	p.(Arg1108His)	36	c.5044_5058del	p.(Val1682_Val1686del)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0516	STGD	ABCA4	NM_000350	36	c.5044_5058del	p.(Val1682_Val1686del)	36	c.5044_5058del	p.(Val1682_Val1686del)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0518	STGD	ABCA4	NM_000350	6	c.742_768+29del	p.(Val248_Val256del)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0519	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0521	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0523	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0524	STGD	ABCA4	NM_000350	28;35	c.[4222T>C;4918C>T]	p.[Trp1408Arg;Arg1640Trp]	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0528	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	44	c.6089G>A	p.(Arg2030Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0529	STGD	ABCA4	NM_000350	39	c.5531G>A	p.(Gly1844Asp)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0530	STGD	ABCA4	NM_000350	39	c.5512C>G	p.(His1838Asp)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0534	STGD	ABCA4	NM_000350	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	48	c.6718A>G	p.(Thr2240Ala)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0535	STGD	ABCA4	NM_000350	5	c.457A>T	p.(Ile153Leu)	35	c.4918C>T	p.(Arg1640Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0536	STGD	ABCA4	NM_000350	21	c.3113C>T	p.(Ala1038Val)	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0537	STGD	ABCA4	NM_000350	8	c.1022A>T	p.(Glu341Val)	35	c.4918C>T	p.(Arg1640Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0539	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0544	STGD	ABCA4	NM_000350	22	c.3292C>T	p.(Arg1098Cys)	IVS33	c.4773+1G>T	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0545	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	46	c.6329G>A	p.(Trp2110*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0547	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	39;42	c.[5512C>G;5882G>A]	p.[His1838Asp;Gly1961Glu]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0548	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	5;46	c.6310C>T	p.(Gln2104*)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0553	STGD	ABCA4	NM_000350	6	c.735T>G	p.(Tyr245*)	44	c.6089G>A	p.(Arg2030Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0554	CCRD	ABCA4	NM_000350	22	c.3299T>C	p.(Ile1100Thr)	22	c.3299T>C	p.(Ile1100Thr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0555	STGD	ABCA4	NM_000350	1;44	c.[1A>G;6089G>A]	p.[Met1Val;Arg2030Gln]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0556	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0558	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0559	STGD	ABCA4	NM_000350	35	c.4919G>A	p.(Arg1640Gln)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0560	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	47	c.6410G>A	p.(Cys2137Tyr)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0563	STGD	ABCA4	NM_000350	23	c.3364G>A	p.(Glu1122Lys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0565	CCRD	ABCA4	NM_000350	23	c.3380G>A	p.(Gly1127Glu)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0568	STGD	ABCA4	NM_000350	IVS47	c.6480-1G>T	p.?	40	c.5603A>T	p.(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0571	STGD	ABCA4	NM_000350	23	c.3409A>G	p.(Arg1137Gly)	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0572	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	40	c.5655del	p.(Val1887Trpfs*6)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0577	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0578	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0580	STGD	ABCA4	NM_000350	26	c.3832G>T	p.(Glu1278*)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0581	STGD	ABCA4	NM_000350	IVS32	c.4668-1G>A	p.?	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0582	STGD	ABCA4	NM_000350	34	c.4793C>A	p.(Ala1598Asp)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0583	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	42	c.5858del	p.(Pro1953Glnfs*21)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-0585	STGD	ABCA4	NM_000350	13	c.1766G>A	p.(Trp589*)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0588	STGD	ABCA4	NM_000350	22	c.3323G>A	p.(Arg1108His)	IVS26	c.3862+1G>A	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0589	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	35;36	c.[4926C>G;5044_5058del]	p.[Ser1642Arg;Val1681_Cys1685del]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0590	STGD	ABCA4	NM_000350	27	c.4003_4004del	p.(Pro1335Argfs*86)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0595	STGD	ABCA4	NM_000350	31	c.4576A>G	p.(Thr1526Ala)	IVS44	c.6147+2T>A	p.?	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0597	STGD	ABCA4	NM_000350	12	c.1714C>T	p.(Arg572*)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0599	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0600	STGD	ABCA4	NM_000350	23	c.3364G>A	p.(Glu1122Lys)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0602	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	46	c.6320G>A	p.(Arg2107His)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0604	CCRD	ABCA4	NM_000350	4	c.393del	p.(Leu132Cysfs*22)	19	c.2888del	p.(Gly963Alafs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0605	STGD	ABCA4	NM_000350	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0607	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0611	STGD	ABCA4	NM_000350	3	c.184C>G	p.(Pro62Ala)	13	c.1933G>A	p.(Asp645Asn)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0617	STGD	ABCA4	NM_000350	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	46	c.6320G>A	p.(Arg2107His)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0619	STGD	ABCA4	NM_000350	15	c.2300T>A	p.(Val767Asp)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0621	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS19	c.2919-826T>A	p.(Leu973Phefs*1)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0622	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0625	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0626	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	44	c.6088C>T	p.(Arg2030*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0631	STGD	ABCA4	NM_000350	8	c.982G>T	p.(Glu328*)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0632	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0635	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	35	c.4918C>T	p.(Arg1640Trp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0640	CCRD	ABCA4	NM_000350	45	c.6179T>G	p.(Leu2060Arg)	45	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0653	STGD	ABCA4	NM_000350	14	c.2057T>C	p.(Leu686Ser)	40	c.5603A>T	p.(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0658	STGD	ABCA4	NM_000350	45	c.6179T>G	p.(Leu2060Arg)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0663	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	27	c.3898C>T	p.(Arg1300*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0675	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23;48	c.[3386G>T;6718A>G]	p.[Arg1129Leu;Thr2240Ala]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0683	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	47	c.6449G>A	p.(Cys2150Tyr)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0691	CCRD	ABCA4	NM_000350	8	c.871C>G	p.(Pro291Ala)	13	c.1804C>T	p.(Arg602Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0692	STGD	ABCA4	NM_000350	12	c.1751_1753delinsAT	p.(Ile584Asnfs*65)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0694	CCRD	ABCA4	NM_000350	3	c.287del	p.(Asn96Thrfs*19)	23;30	c.[3386G>T;4537dup]	p.[Arg1129Leu;Gln1513Profs*42]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0696	OtherMD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	43	c.5914G>A	p.(Gly1972Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0698	CCRD	ABCA4	NM_000350	13	c.1755del	p.(Lys585Lysfs*63)	30	c.4457C>T	p.(Pro1486Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0701	CCRD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	IVS46	c.6387-1G>A	p.?	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0704	CCRD	ABCA4	NM_000350	16	c.2481del	p.(Thr829Argfs*14)	46	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0714	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0716	STGD	ABCA4	NM_000350	IVS40	c.5714+5G>A	p.(=,Glu1863Leufs*33)	42;45	c.[5843C>T;6179T>G]	p.[Pro1948Leu;Leu2060Arg]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0717	STGD	ABCA4	NM_000350	14	c.2041C>T	p.(Arg681*)	48	c.6718A>G	p.(Thr2240Ala)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0720	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0723	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	47	c.6410G>A	p.(Cys2137Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0724	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	45	c.6221G>A	p.(Gly2074Asp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0731	OtherMD	ABCA4	NM_000350	13	c.1819G>C	p.(Gly607Arg)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0736	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	28	c.4139C>T	p.(Pro1380Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0740	STGD	ABCA4	NM_000350	47	c.6410G>A	p.(Cys2137Tyr)	IVS28	c.4253+43G>A	p.[=, Ile1377Hisfs*3]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0741	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0742	STGD	ABCA4	NM_000350	IVS13	c.1938-514A>G	p.[Phe647Serfs*155,Phe647Serfs*22,=]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0746	STGD	ABCA4	NM_000350	14	c.2041C>T	p.(Arg681*)	14	c.2041C>T	p.(Arg681*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0747	CCRD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	35	c.4849del	p.(Val1617Cysfs*45)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0748	STGD	ABCA4	NM_000350	9	c.1222C>T	p.(Arg408*)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0750	STGD	ABCA4	NM_000350	12	c.1751_1753delinsAT	p.(Ile584Asnfs*65)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0751	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0754	CCRD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0759	STGD	ABCA4	NM_000350	14	c.2041C>T	p.(Arg681*)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0763	STGD	ABCA4	NM_000350	12	c.1667T>G	p.(Met556Arg)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0765	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0768	STGD	ABCA4	NM_000350	20;38	c.[2971G>C;3899G>A]	p.[Gly991Arg;Arg1300Gln]	38	c.5317_5318insA	p.(Ala1773Aspfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0769	STGD	ABCA4	NM_000350	14	c.2023G>A	p.(Val675Ile)	16	c.2488G>T	p.(Glu830*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0770	CCRD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	30	c.4537dup	p.(Gln1513Profs*42)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0771	STGD	ABCA4	NM_000350	15	c.2285C>A	p.(Ala762Glu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0778	STGD	ABCA4	NM_000350	34	c.4793C>A	p.(Ala1598Asp)	34	c.4793C>A	p.(Ala1598Asp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0779	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	IVS28	c.4253+43G>A	p.[=,Ile1377Hisfs*3]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0780	STGD	ABCA4	NM_000350	8	c.982G>T	p.(Glu328*)	39	c.5549T>C	p.(Leu1850Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0781	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	13	c.1804C>T	p.(Arg602Trp)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0783	STGD	ABCA4	NM_000350	IVS30;42	c.[4540-8T>C;5882G>A]	p.[Gln1513insProGln;Gly1961Glu]	8	c.950del	p.(Gly317Alafs*57)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0787	STGD	ABCA4	NM_000350	43	c.5917del	p.(Val1973*)	43	c.5917del	p.(Val1973*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0790	STGD	ABCA4	NM_000350	12	c.1715G>C	p.(Arg572Pro)	35	c.4918C>T	p.(Arg1640Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0791	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0793	STGD	ABCA4	NM_000350	21	c.3113C>T	p.(Ala1038Val)	36	c.5172G>T	p.(Trp1724Cys)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0794	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	27	c.3988G>T	p.(Glu1330*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0797	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0799	STGD	ABCA4	NM_000350	43	c.5929G>A	p.(Gly1977Ser)	IVS28	c.4253+43G>A	p.[=, Ile1377Hisfs*3]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0807	STGD	ABCA4	NM_000350	6;16	c.[634C>T;2546T>C]	p.[Arg212Cys;Val849Ala]	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-0819	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0821	STGD	ABCA4	NM_000350	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0822	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0826	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0829	STGD	ABCA4	NM_000350	14	c.1957C>T	p.(Arg653Cys)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0832	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	47	c.6415C>T	p.(Arg2139Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0834	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0847	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0852	STGD	ABCA4	NM_000350	27	c.3874C>T	p.(Gln1292*)	21	c.3113C>T	p.(Ala1038Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0853	STGD	ABCA4	NM_000350	30	c.4537dup	p.(Gln1513Profs*42)	40	c.5603A>T	p.(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0855	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	30	c.4353-1G>A	p.?	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0856	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	43	c.5917del	p.(Val1973*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0859	STGD	ABCA4	NM_000350	6	c.611C>A	p.(Ala204Asp)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0863	STGD	ABCA4	NM_000350	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0864	STGD	ABCA4	NM_000350	13	c.1868A>G	p.(Gln623Arg)	22	c.3259G>A	p.(Glu1087Lys)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0869	CCRD	ABCA4	NM_000350	13	c.1832T>C	p.(Leu611Pro)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0870	STGD	ABCA4	NM_000350	3	c.223T>G	p.(Cys75Gly)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0877	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0878	OtherMD	ABCA4	NM_000350	8	c.982G>T	p.(Glu328*)	8	c.982G>T	p.(Glu328*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0881	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	44	c.6089G>A	p.(Arg2030Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0882	OtherMD	ABCA4	NM_000350	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	IVS39	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0885	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	46	c.6316C>T	p.(Arg2106Cys)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0886	STGD	ABCA4	NM_000350	35;36	c.[4926C>G;5044_5058del]	p.[Ser1642Arg;Val1682_Val1686del]	46	c.6380C>T	p.(Ser2127Phe)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0894	CCRD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0895	STGD	ABCA4	NM_000350	23	c.3364G>A	p.(Glu1122Lys)	IVS28	c.4253+43G>A	p.[=,Ile1377Hisfs*3]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0906	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3364G>A	p.(Glu1122Lys)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0908	STGD	ABCA4	NM_000350	8	c.1025_1038del	p.(Asp342Glyfs*6)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0911	STGD	ABCA4	NM_000350	13	c.1879G>T	p.(Glu627*)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0918	STGD	ABCA4	NM_000350	44	c.6088C>T	p.(Arg2030*)	45	c.6216T>A	p.(Ser2072Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0925	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0929	CCRD	ABCA4	NM_000350	36	c.5044_5058del	p.(Val1681_Cys1685del)	36	c.5044_5058del	p.(Val1682_Val1686del)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0932	STGD	ABCA4	NM_000350	28;35	c.[4222T>C;4918C>T]	p.[Trp1408Arg;Arg1640Trp]	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0939	STGD	ABCA4	NM_000350	IVS36	c.5196+1137G>A	p.[Met1733Glufs*78,=]	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0941	STGD	ABCA4	NM_000350	19	c.2894A>G	p.(Asn965Ser)	45	c.6272T>A	p.(Leu2091Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0943	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	40	c.5644A>G	p.(Met1882Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0945	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS29	c.4353-1G>A	p.?	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0946	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	29;48	c.[3386G>T;6718A>G]	p.[Arg1129Leu;Thr2240Ala]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0952	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0953	OtherMD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0954	STGD	ABCA4	NM_000350	21	c.3113C>T	p.(Ala1038Val)	IVS21	c.3050+5G>A	p.(Leu973_His1017delinsPhe)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0955	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly693Alafs*14)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0956	STGD	ABCA4	NM_000350	17;40	c.[2588G>C;5603A>T]	p.[Gly863Ala,Gly863del;Asn1868Ile]	IVS38;40	c.[5461-10T>C;5603A>T]	p.[Thr1821Aspfs*6;Asn1868Ile]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0959	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0960	STGD	ABCA4	NM_000350	23	c.3383A>G	p.(Asp1128Gly)	23	c.3383A>G	p.(Asp1128Gly)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0962	STGD	ABCA4	NM_000350	29	c.4254C>A	p.(Ser1418Arg)	29	c.4254C>A	p.(Ser1418Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0963	STGD	ABCA4	NM_000350	14	c.2023G>A	p.(Val675Ile)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0964	STGD	ABCA4	NM_000350	15	c.2285C>A	p.(Ala762Glu)	34	c.4793C>A	p.(Ala1598Asp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0966	STGD	ABCA4	NM_000350	39	c.5549T>C	p.(Leu1850Pro)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0967	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	IVS34	c.4848+3A>G	p.[Gly1592_Lys1616del,=]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0968	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	27	c.3988G>T	p.(Glu1330*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0979	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	27	c.3943C>T	p.(Gln1315*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0980	STGD	ABCA4	NM_000350	45	c.6179T>G	p.(Leu2060Arg)	48	c.6718A>G	p.(Thr2240Ala)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0981	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0984	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0985	STGD	ABCA4	NM_000350	13	c.1766G>C	p.(Trp589Ser)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0989	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0991	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0993	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-0995	CCRD	ABCA4	NM_000350	11	c.1364T>A	p.(Leu455Gln)	14	c.2099G>A	p.(Trp700*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0997	STGD	ABCA4	NM_000350	23	c.3383A>G	p.(Asp1128Gly)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1000	STGD	ABCA4	NM_000350	16	c.[2483C>T;2481del]	p.[Pro828Leu;Thr829Argfs*14]	40	c.5603A>T	p.(Asn1868Ile)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1001	STGD	ABCA4	NM_000350	44	c.6089G>A	p.(Arg2030Gln)	47	c.6410G>A	p.(Cys2137Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1002	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1007	STGD	ABCA4	NM_000350	23	c.3383A>G	p.(Asp1128Gly)	39	c.5549T>C	p.(Leu1850Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1010	STGD	ABCA4	NM_000350	12	c.1622T>C	p.(Leu541Pro)	17	c.2588G>C	p.[Gly863Ala,Gly863del]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1015	STGD	ABCA4	NM_000350	38	c.5384T>C	p.(Leu1795Ser)	38	c.5384T>C	p.(Leu1795Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1018	OtherMD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1022	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1025	STGD	ABCA4	NM_000350	35	c.4918C>T	p.(Arg1640Trp)	45	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1034	STGD	ABCA4	NM_000350	23	c.3383A>G	p.(Asp1128Gly)	23	c.3383A>G	p.(Asp1128Gly)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1036	STGD	ABCA4	NM_000350	22;44	c.[3322C>T;6071A>G]	p.[Arg1108His;Asp2024Gly]	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1041	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	36	c.5196+1G>A	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1042	STGD	ABCA4	NM_000350	23	c.3383A>G	p.(Asp1128Gly)	IVS41	c.5836-1G>C	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-1045	STGD	ABCA4	NM_000350	13	c.1819G>C	p.(Gly607Arg)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1046	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS33	c.4773+1del	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1047	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1048	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1049	STGD	ABCA4	NM_000350	4	c.428C>T	p.(Pro143Leu)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1050	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1059	STGD	ABCA4	NM_000350	15	c.2285C>A	p.(Ala762Glu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1063	CCRD	ABCA4	NM_000350	35;36	c.[4926C>G;5044_5058del]	p.[Ser1642Arg; Val1681_Cys1685del]	35;36	c.[4926C>G;5044_5058del]	p.[Ser1642Arg; Val1681_Cys1685del]	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1064	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS30	c.4539+2064C>T	p.[=, Arg1514Leufs*36]#	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1067	STGD	ABCA4	NM_000350	45	c.6229C>T	p.(Arg2077Trp)	45	c.6229C>T	p.(Arg2077Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1070	STGD	ABCA4	NM_000350	13	c.1927G>A	p.(Ala643Met)	44	c.6089G>A	p.(Arg2030Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1071	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1072	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1075	STGD	ABCA4	NM_000350	9	c.1222C>T	p.(Arg408*)	IVS28;40	c.[4253+43G>A;5603A>T]	p.[=,Ile1377Hisfs*3](;)(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1077	STGD	ABCA4	NM_000350	8	c.1025_1038del	p.(Asp342Glyfs*6)	22	c.3292C>T	p.(Arg1098Cys)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1079	OtherMD	ABCA4	NM_000350	19	c.2791G>A	p.(Val931Met)	38	c.5318C>T	p.(Ala1773Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1086	OtherMD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1087	STGD	ABCA4	NM_000350	IVS34	c.4848+3A>G	p.[Gly1592_Lys1616del,=]	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1100	STGD	ABCA4	NM_000350	3	c.286A>G	p.(Asn96Asp)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1101	STGD	ABCA4	NM_000350	17	c.2588G>C	p.(Gly863Ala, Gly863del)	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1102	STGD	ABCA4	NM_000350	23;48	c.[3386G>T;6718A>G]	p.[Arg1129Leu;Thr2240Ala]	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1106	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	40	c.5644A>G	p.(Met1882Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1107	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	22	c.3277G>A	p.(Asp1093Asn)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1110	STGD	ABCA4	NM_000350	29	c.4322G>A	p.(Gly1441Asp)	48	c.6718A>G	p.(Thr2240Ala)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1114	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	30	c.4469G>A	p.(Cys1490Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1116	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1119	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1120	STGD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	40	c.5603A>T	p.(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1121	STGD	ABCA4	NM_000350	ISV40	c.5714+1G>A	p.?	48	c.6718A>G	p.(Thr2240Ala)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1123	CCRD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1130	STGD	ABCA4	NM_000350	39	c.5549T>C	p.(Leu1850Pro)	39	c.5549T>C	p.(Leu1850Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1137	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1138	CCRD	ABCA4	NM_000350	4	c.378G>A	p.(Trp126*)	6	c.634C>T	p.(Arg212Cys)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1139	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	48	c.6559C>T	p.(Gln2187*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1145	OtherMD	ABCA4	NM_000350	9	c.1222C>T	p.(Arg408*)	40	c.5603A>T	p.(Asn1868Ile)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1146	STGD	ABCA4	NM_000350	14	c.2023G>A	p.(Val675Ile)	47	c.6410G>A	p.(Cys2137Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1147	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	28	c.4139C>T	p.(Pro1380Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1156	STGD	ABCA4	NM_000350	3	c.287del	p.(Asn96Thrfs*19)	12	c.1667T>G	p.(Met556Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1157	CCRD	ABCA4	NM_000350	8	c.982G>T	p.(Glu328*)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1163	OtherMD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	IVS38;40	c.[5461-10T>C;5603A>T]	p.[Thr1821Aspfs*6,Thr1821Valfs*13;Asn1868Ile]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1164	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	30	c.4457C>T	p.(Pro1486Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1184	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1193	STGD	ABCA4	NM_000350	22	c.3292C>T	p.(Arg1098Cys)	40	c.5630_5644dup	p.(Lys1877_Ala1881dup)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1202	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1205	STGD	ABCA4	NM_000350	41	c.5819T>C	p.(Leu1940Pro)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1207	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	30	c.4436G>A	p.(Trp1479*)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1208	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	47	c.6437G>T	p.(Gly2146Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1211	STGD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1212	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	38	c.5318C>T	p.(Ala1773Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1215	STGD	ABCA4	NM_000350	13	c.1819G>C	p.(Gly607Arg)	40	c.5603A>T	p.(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1216	STGD	ABCA4	NM_000350	13	c.1766G>A	p.(Trp589*)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1219	STGD	ABCA4	NM_000350	8	c.874A>C	p.(Ser292Arg)	22	c.3251T>C	p.(Ile1084Thr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1220	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	47	c.6449G>A	p.(Cys2150Tyr)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1223	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	35	c.4849del	p.(Val1617Cysfs*45)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1228	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	39	c.5549T>C	p.(Leu1850Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1230	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	38	c.5318C>T	p.(Ala1773Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1233	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1235	STGD	ABCA4	NM_000350	27	c.3988G>T	p.(Glu1330*)	IVS38	c.5461-1G>T	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1236	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	48	c.6718A>G	p.(Thr2240Ala)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1238	STGD	ABCA4	NM_000350	22	c.3277G>A	p.(Asp1093Asn)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1239	STGD	ABCA4	NM_000350	22	c.3259G>A	p.(Glu108Lys)	36	c.5044_5058del	p.(Val1682_Val1686del)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1240	STGD	ABCA4	NM_000350	1	c.32T>C	p.(Leu11Pro)	30	c.4457C>T	p.(Pro1486Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1243	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	27	c.3943C>T	p.(Gln1315*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1246	STGD	ABCA4	NM_000350	35	c.4918C>T	p.(Arg1640Trp)	46	c.6329G>A	p.(Trp2110*)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1249	OtherMD	ABCA4	NM_000350	8	c.871C>G	p.(Pro291Ala)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1251	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1255	STGD	ABCA4	NM_000350	35	c.4918C>T	p.(Arg1640Trp)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1256	STGD	ABCA4	NM_000350	IVS14	c.2161-8G>A	p.(His721_Val794del)	17	c.2613G>A	p.(Trp871*)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1257	STGD	ABCA4	NM_000350	3	c.214G>A	p.(Gly72Arg)	43	c.5929G>A	p.(Gly1977Ser)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1258	OtherMD	ABCA4	NM_000350	22	c.3259G>A	p.(Glu1087Lys)	22	c.3259G>A	p.(Glu1087Lys)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1264	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	27	c.4070C>T	p.(Ala1357Val)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1271	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1272	STGD	ABCA4	NM_000350	IVS6	c.768+2T>G	p.?	43	c.5899T>G	p.(Cys1967Gly)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-1273	STGD	ABCA4	NM_000350	23;48	c.[3386G>T;6718A>G]	p.[Arg1129Leu;Thr2240Ala]	23;48	c.[3386G>T;6718A>G]	p.[Arg1129Leu;Thr2240Ala]	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1274	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1279	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS38;40	c.[5461-10T>C;5603A>T]	p.[Thr1821Aspfs*6,Thr1821Valfs*13;Asn1868Ile]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1298	STGD	ABCA4	NM_000350	35	c.4919G>A	p.(Arg1640Gln)	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs*33]	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1300	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS38	c.5461-10T>C	p.(Thr1821Aspfs*6)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1302	STGD	ABCA4	NM_000350	30	c.4457C>T	p.(Pro1486Leu)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1339	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1343	STGD	ABCA4	NM_000350	38	c.5318C>T	p.(Ala1773Val)	38	c.5318C>T	p.(Ala1773Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1348	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1349	STGD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1350	STGD	ABCA4	NM_000350	17	c.2588G>C	p.[Gly863Ala,Gly863del]	19	c.2888del	p.(Gly963Alafs*14)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1353	OtherMD	ABCA4	NM_000350	12	c.1609C>T	p.(Arg537Cys)	43	c.5981G>A	p.(Gly1994Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1356	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1359	STGD	ABCA4	NM_000350	12;21	c.[1622T>C;3113C>T]	p.[Leu541Pro;Ala1038Val]	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1375	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1376	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	38	c.5318C>T	p.(Ala1773Val)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1377	OtherMD	ABCA4	NM_000350	13	c.1853G>T	p.(Gly618Val)	39	c.5531_5557dup	p.(Gly1844_Gln1852dup)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1381	OtherMD	ABCA4	NM_000350	IVS28	c.4253+43G>A	p.[=, Ile1377Hisfs*3]	35	c.4919G>A	p.(Arg1640Gln)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-0033	CCRD	ABCA4	NM_000350	13	c.1848del	p.(Glu616Aspfs*33)	13	c.1848del	p.(Glu616Aspfs*33)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-0193	STGD	ABCA4	NM_000350	45	c.6179T>G	p.(Leu2060Arg)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-0266	CCRD	ABCA4	NM_000350	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	46	c.6179T>G	p.(Leu2060Arg)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-0267	CCRD	ABCA4	NM_000350	36	c.5044_5058del	p.(Val1682_Val1686del)	36	c.5044_5058del	p.(Val1682_Val1686del)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-0280	STGD	ABCA4	NM_000350	38	c.5413A>G	p.(Asn1805Asp)	38	c.5413A>G	p.(Asn1805Asp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-0298	CCRD	ABCA4	NM_000350	8	c.950del	p.(Gly317Alafs*57)	33	c.4720G>T	p.(Glu1574*)	AR	AR	Stargardt disease/cone-rod dystrophy		yes
RP-0532	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	38	c.5318C>T	p.(Ala1773Val)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-0674	CCRD	ABCA4	NM_000350	3	c.287del	p.(Asn96Thrfs*19)	30	c.4537dup	p.(Gln1513Profs*42)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-0714	CCRD	ABCA4	NM_000350	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	IVS28	c.4253+4C>T	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		yes
RP-0741	CCRD	ABCA4	NM_000350	43	c.5917del	p.(Val1973*)	43	c.5917del	p.(Val1973*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-0998	CCRD	ABCA4	NM_000350	14	c.2041C>T	p.(Arg681*)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-1102	CCRD	ABCA4	NM_000350	15	c.2285C>A	p.(Ala762Glu)	15	c.2285C>A	p.(Ala762Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-1112	CCRD	ABCA4	NM_000350	1	c.1A>G	p.(Met1Val)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1126	CCRD	ABCA4	NM_000350	IVS26	c.3862+1G>A	p.?	46	c.6329G>A	p.(Trp2110*)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1367	CCRD	ABCA4	NM_000350	IVS26	c.3862+1G>A	p.?	IVS26	c.3862+1G>A	p.?	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1455	CCRD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	48	c.6688del	p.(Leu2230Serfs*17)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-1481	STGD	ABCA4	NM_000350	21	c.3113C>T	p.(Ala1038Val)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1539	CCRD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	30	c.4417C>A	p.(Leu1437Met)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1543	CCRD	ABCA4	NM_000350	28	c.4234C>T	p.(Gln1412*)	43	c.5917del	p.(Val1973*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1578	STGD	ABCA4	NM_000350	22	c.3287C>T	p.(Ser1096Leu)	34	c.4793C>A	p.(Ala1598Asp)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1603	CCRD	ABCA4	NM_000350	16	c.2568C>A	p.(Tyr856*)	16	c.2568C>A	p.(Tyr856*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1669	CCRD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1680	CCRD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	36	c.5044_5058del	p.(Val1682_Val1686del)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1715	CCRD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	13	c.1804C>T	p.(Arg602Trp)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-1742	STGD	ABCA4	NM_000350	20	c.2980A>G	p.(Ile994Val)	38	c.5383T>G	p.(Leu1795Val)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1769	STGD	ABCA4	NM_000350	5	c.560G>A	p.(Arg187His)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1819	CCRD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-1844	CCRD	ABCA4	NM_000350	30	c.4537dup	p.(Gln1513Profs*42)	30	c.4537dup	p.(Gln1513Profs*42)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2028	STGD	ABCA4	NM_000350	15	c.2300T>A	p.(Val767Asp)	IVS41	c.5836-1G>C	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2104	STGD	ABCA4	NM_000350	13	c.1879G>T	p.(Glu627*)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2133	CCRD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	36	c.5044_5058del	p.(Val1682_Val1686del)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2143	CCRD	ABCA4	NM_000350	9	c.1222C>T	p.(Arg408*)	IVS38	c.5461-1G>T	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2198	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2210	CCRD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	21	c.3056C>T	p.(Thr1019Met)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2252	CCRD	ABCA4	NM_000350	19	c.2878G>A	p.(Ala960Thr)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2343	CCRD	ABCA4	NM_000350	6	c.613T>G	p.(Cys205Gly)	41	c.5819T>C	p.(Leu1940Pro)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2386	CCRD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	13	c.1804C>T	p.(Arg602Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2419	STGD	ABCA4	NM_000350	11	c.1364T>A	p.(Leu455Gln)	34	c.4793C>A	p.(Ala1598Asp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2492	STGD	ABCA4	NM_000350	22	c.3210_3211dup	p.(Ser1071Cysfs*14)	33	c.4672G>A	p.(Gly1558Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2500	STGD	ABCA4	NM_000350	6	c.671del	p.(Thr224Argfs*17)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2520	CCRD	ABCA4	NM_000350	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2531	STGD	ABCA4	NM_000350	IVS6-Ex6	c.699_768+341del	p.(Gln234Phefs*5)	IVS6-Ex6	c.699_768+341del	p.(Gln234Phefs*5)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2668	CCRD	ABCA4	NM_000350	IVS6-Ex6	c.699_768+341del	p.(Gln234Phefs*5)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2680	CCRD	ABCA4	NM_000350	19	c.2888del	p.(Gly963Alafs*14)	30	c.4457C>T	p.(Pro1486Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-2711	STGD	ABCA4	NM_000350	IVS20	c.3050+5G>A	p.(Leu973_His1017delinsPhe)#	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2867	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS28	c.4253+5G>A	p.(Ile1377Hisfs*3)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2948	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-2956	CCRD	ABCA4	NM_000350	28	c.4253+5G>A	p.(Ile1377Hisfs*3)	28	c.4253+5G>A	p.(Ile1377Hisfs*3)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-3080	STGD	ABCA4	NM_000350	45	c.6179T>G	p.(Leu2060Arg)	48	c.6647C>T	p.(Ala2216Val)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
RP-3093	CCRD	ABCA4	NM_000350	33	c.4773+1G>T	p.?	33	c.4773+1G>T	p.?	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-3118	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	25	c.4918C>T	p.(Arg1640Trp)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
RP-3136	STGD	ABCA4	NM_000350	23	c.3420C>G	p.(Cys1140Trp)	23	c.3386G>T	p.(Arg1129Leu)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1391	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	41	c.5819T>C	p.(Leu1940Pro)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1396	STGD	ABCA4	NM_000350	13	c.1766G>A	p.(Trp589*)	22	c.3322C>T	p.(Arg1108Cys)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1403	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	27	c.3943C>T	p.(Gln1315*)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-1405	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	30	c.4457C>T	p.(Pro1486Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1407	STGD	ABCA4	NM_000350	27	c.3874C>T	p.(Gln1292*)	47	c.6449G>A	p.(Cys2150Tyr)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1411	STGD	ABCA4	NM_000350	22	c.3323G>A	p.(Arg1108His)	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1415	OtherMD	ABCA4	NM_000350	IVS30	c.4539+2064C>T	p.[=,Arg1514Leufs*36]	36	c.5044_5058del	p.(Val1682_Val1686del)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1416	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	48	c.6718A>G	p.(Thr2240Ala)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1419	STGD	ABCA4	NM_000350	44	c.6112C>T	p.(Arg2038Trp)	44	c.6112C>T	p.(Arg2038Trp)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1430	OtherMD	ABCA4	NM_000350	47	c.6449G>A	p.(Cys2150Tyr)	48	**c.6512T>G**	**p.(Ile2171Ser) novel**	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1434	OtherMD	ABCA4	NM_000350	13	c.1804C>T	p.(Arg602Trp)	42	c.5882G>A	p.(Gly1961Glu)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1436	OtherMD	ABCA4	NM_000350	6	c.634C>T	p.(Arg212Cys)	23	c.3386G>T	p.(Arg1129Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1438	STGD	ABCA4	NM_000350	12	c.1622T>C	p.(Leu541Pro)	21	c.3113C>T	p.(Ala1038Val)	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1442	OtherMD	ABCA4	NM_000350	21	c.3113C>T	p.(Ala1038Val)	35	c.4919G>A	p.(Arg1640Gln)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1443	STGD	ABCA4	NM_000350	21	c.3056C>T	p.(Thr1019Met)	22	c.3323G>A	p.(Arg1108His)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1457	STGD	ABCA4	NM_000350	44	c.6088C>T	p.(Arg2030*)	47	**c.6475T>C**	**p.(Ser2159Pro) novel**	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1464	STGD	ABCA4	NM_000350	13	c.1766G>A	p.(Trp589*)	22	c.3292C>T	p.(Arg1098Cys)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1468	CCRD	ABCA4	NM_000350	IVS6-Ex6	c.699_768+341del	p.(Gln234Phefs*5)	IVS6-Ex6	c.699_768+341del	p.(Gln234Phefs*5)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1470	OtherMD	ABCA4	NM_000350	IVS38	**c.5461-6T>C**	**p.? novel**	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1471	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	45	c.6179T>G	p.(Leu2060Arg)[†]	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1473	STGD	ABCA4	NM_000350	3	c.287del	p.(Asn96Thrfs*19)	40	c.5714+5G>A	p.[=,Glu1863Leufs 33]	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1479	OtherMD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	43	c.5929G>A	p.(Gly1977Ser)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1485	OtherMD	ABCA4	NM_000350	20	c.2966T>C	p.(Val989Ala)	46	**c.6306C>G**	**p.(Asp2102Glu) novel**	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1489	STGD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	IVS4	c.4253+4C>T	p.(Ile1377Hisfs*3)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1494	STGD	ABCA4	NM_000350	25	c.3813G>C	p.(Glu1271Asp)[†]	48	c.6718A>G	p.(Thr2240Ala)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1502	STGD	ABCA4	NM_000350	IVS40	c.5714+5G>A	p.[=,Glu1863Leufs 33]	20	c.3004C>T	p.(Arg1002Trp)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1510	STGD	ABCA4	NM_000350	14	c.2041C>T	p.(Arg681*)	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1517	CCRD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1519	STGD	ABCA4	NM_000350	42	c.5882G>A	p.(Gly1961Glu)	45	c.6179T>G	p.(Leu2060Arg)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1524	OtherMD	ABCA4	NM_000350	8	c.1025_1038del	p.(Asp342Glyfs*6)	42	c.5882G>A	p.(Gly1961Glu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1535	OtherMD	ABCA4	NM_000350	16	c.2568C>A	p.(Tyr856*)	IVS33	c.4773+1G>T	p.?	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1536	STGD	ABCA4	NM_000350	28	c.4200C>A	(p.Tyr1400*)	44	c.6089G>A	p.(Arg2030Gln)	sporadic	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1538	STGD	ABCA4	NM_000350	23	c.[3386G>T;4947del]	p.[Arg1129Leu;Glu1650Argfs*12]	40	c.5603A>T	p.(Asn1868Ile)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1540	CCRD	ABCA4	NM_000350	23	c.3386G>T	p.(Arg1129Leu)	44	c.6118C>T	p.(Arg2040*)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1543	STGD	ABCA4	NM_000350	22	c.3292C>T	p.(Arg1098Cys)	33	**c.4673G>A**	**p.(Gly1558Glu) novel**	unknown	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1544	STGD	ABCA4	NM_000350	23;48	c.[3386G>T;6718A>G]	p.[Arg1129Leu;Thr2240Ala]	6	c.700C>T	p.(Gln234*)	AR	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1546	STGD	ABCA4	NM_000350	8	c.982G>T	p.(Glu328*)	30	c.4457C>T	p.(Pro1486Leu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-1551	STGD	ABCA4	NM_000350	22;46	c.[3322C>T;6320G>A]	p.[Arg1108Cys;Arg2107His]	23	c.3386G>T	p.(Arg1129Leu)	sporadic	AR	Stargardt disease/cone-rod dystrophy		Yes
MD-1555	OtherMD	ABCA4	NM_000350	IVS41	c.5836-1G>C	p.?	42	c.5882G>A	p.(Gly1961Glu)	AR	AR	Stargardt disease/cone-rod dystrophy		NA
MD-0159	CCRD	ADAM9	NM_003816	14	c.1420C>T	p.(Arg474*)	14	**c.1420C>T**	**p.(Arg474*) novel**	sporadic	AR	Cone-rod dystrophy		Yes
MD-0224	CCRD	BBS1	NM_024649	12	c.1169T>G	p.(Met390Arg)	12	c.1169T>G	p.(Met390Arg)	sporadic	AR	Cone-rod dystrophy		NA
MD-0352	OtherMD	BBS1	NM_024649	16	c.1645G>T	p.(Glu549*)	13	**c.1205T>C**	**p.(Leu402Pro) novel**	AR	AR	Cone-rod dystrophy		Yes
MD-0365	OtherMD	BBS1	NM_024649	12	c.1169T>G	p.(Met390Arg)	12	c.1169T>G	p.(Met390Arg)	sporadic	AR	Cone-rod dystrophy		Yes
MD-1060	OtherMD	BBS1	NM_024649	12	c.1169T>G	p.(Met390Arg)	12	c.1169T>G	p.(Met390Arg)	sporadic	AR	Cone-rod dystrophy		NA
MD-0134	CCRD	BEST1	NM_004183	4	c.388C>A	p.(Arg130Ser)	4	c.388C>A	p.(Arg130Ser)	AR	AR	Best disease		NA
MD-0154	OtherMD	BEST1	NM_004183	4	c.388C>A	p.(Arg130Ser)	9	c.1100+1G>A	p.?	AR	AR	Best disease		NA
MD-0314	OtherMD	BEST1	NM_004183	5	c.536_538del	p.(Asn179del)				sporadic	AD	Best disease	Segregation studies unavailable	NA
MD-0375	OtherMD	BEST1	NM_004183	2	c.11C>T	p.(Thr4Iso)				sporadic	AD	Best disease	Segregation studies unavailable	NA
MD-0459	OtherMD	BEST1	NM_004183	4	c.388C>A	p.(Arg130Ser)	4	c.388C>A	p.(Arg130Ser)	sporadic	AR	Best disease		Yes
MD-0490	CCRD	BEST1	NM_004183	4	c.400C>G	p.(Leu134Val)	6	c.638A>G	p.(Glu213Gly)	sporadic	AR	Best disease		NA
MD-0525	OtherMD	BEST1	NM_004183	8	c.911A>T	p.(Asp304Val)				sporadic	AD	Best disease	Segregation studies unavailable	NA
MD-0570	OtherMD	BEST1	NM_004183	6	c.638A>G	p.(Glu213Gly)	6	c.638A>G	p.(Glu213Gly)	sporadic	AR	Best disease		NA
MD-0610	CCRD	BEST1	NM_004183	9	c.974T>C	p.(Met325Thr)	9	c.974T>C	p.(Met325Thr)	sporadic	AR	Best disease		NA
MD-0685	OtherMD	BEST1	NM_004183	10	c.1550C>G	p.(Ser517*)	10	c.1550C>G	p.(Ser517*)	sporadic	AR	Best disease		Yes
MD-0749	STGD	BEST1	NM_004183	8	**c.884T>A**	**p.(Ile295Asn) novel**				sporadic	AD	Best disease	Segregation studies unavailable	NA
MD-1092	OtherMD	BEST1	NM_004183	5	c.596G>A	p.(Gly199Asp) novel	9	c.979del	p.(Gln327Argfs*42)	sporadic	AR	Best disease		Yes
MD-1363	OtherMD	BEST1	NM_004183	4	c.388C>A	p.(Arg130Ser)	5	c.521_522delTG	p.(Leu174Glnfs*57)	sporadic	AR	Best disease		NA
MD-1369	OtherMD	BEST1	NM_004183	7	c.728C>T	p.(Ala243Val)				sporadic	AD	Best disease	Segregation studies unavailable	NA
MD-1402	OtherMD	BEST1	NM_004183	2	c.44G>A	p.(Gly15Asp)				unknown	AD	Best disease	Segregation studies unavailable	NA
MD-1437	OtherMD	BEST1	NM_004183	3	c.388C>A	(p.Arg130Ser)	3	**c.466C>T**	**p.(His156Tyr) novel**	AR	AR	Best disease		NA
MD-0453	CCRD	CDHR1	NM_033100	15	c.1720C>G	p.(Pro574Ala)	15	c.1720C>G	p.(Pro574Ala)	sporadic	AR	Cone-rod dystrophy		Yes
MD-0930	OtherMD	CDHR1	NM_033100	9	c.838C>T	p.(Arg280*)	9	c.838C>T	p.(Arg280*)	sporadic	AR	Cone-rod dystrophy		NA
MD-0034	CCRD	CERKL	NM_001030311	5	**c.769C>T**	**p.(Arg257*) novel**	5	c.769C>T	p.(Arg257*)	sporadic	AR	Cone-rod dystrophy/Retinitis pigmentosa		Yes
MD-0470	CCRD	CERKL	NM_001030311	2	c.356G>A	p.(Gly119Asp)	5	c.769C>T	p.(Arg257*)	sporadic	AR	Cone-rod dystrophy/Retinitis pigmentosa		Yes
MD-0662	OtherMD	CERKL	NM_001030311	6	c.847C>T	p.(Arg283*)	6	c.847C>T	p.(Arg283*)	sporadic	AR	Cone-rod dystrophy/Retinitis pigmentosa		NA
MD-0707	CCRD	CERKL	NM_001030311	2	c.356G>A	p.(Gly119Asp)	2	c.356G>A	p.(Gly119Asp)	sporadic	AR	Cone-rod dystrophy/Retinitis pigmentosa		NA
MD-0827	CCRD	CERKL	NM_001030311	6	c.847C>T	p.(Arg283*)	14	c.1641_1642del	p. Tyr548Trpfs*17	sporadic	AR	Cone-rod dystrophy/Retinitis pigmentosa		NA
MD-0850	STGD	CERKL	NM_001030311	6	c.847C>T	p.(Arg283*)	6	c.847C>T	p.(Arg283*)	sporadic	AR	Cone-rod dystrophy/Retinitis pigmentosa		NA
MD-0890	CCRD	CERKL	NM_001030311	6	c.847C>T	p.(Arg283*)	6	c.847C>T	p.(Arg283*)	sporadic	AR	Cone-rod dystrophy/Retinitis pigmentosa		Yes
MD-0951	OtherMD	CERKL	NM_001030311	6	c.847C>T	p.(Arg283*)	6	c.847C>T	p.(Arg283*)	unknown	AR	Cone-rod dystrophy/Retinitis pigmentosa		NA
MD-1297	OtherMD	CERKL	NM_001030311	6	c.847C>T	p.(Arg283*)	6	c.847C>T	p.(Arg283*)	AR	AR	Cone-rod dystrophy/Retinitis pigmentosa		NA
MD-1465	OtherMD	CERKL	NM_001030311	2	**c.317G>A**	**p.(Arg106His) novel**	2	c.317G>A	p.(Arg106His)	AR	AR	Cone-rod dystrophy/Retinitis pigmentosa		Yes

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-1488	CCRD	CERKL	NM_001030311	2	c.316C>T	p.(Arg106Cys)	2	c.316C>T	p.(Arg106Cys)	AR	AR	Cone-rod dystrophy/Retinitis pigmentosa		NA
MD-0455	STGD	CHM	NM_000390	8	**c.1053del**	**p.(Ile352*) novel**				sporadic	XL	Choroideremia	Segregation studies unavailable. Affected males in family	NA
MD-0426	CCRD	CNGA3	NM_001298	10	c.1279C>T	p.(Arg427Cys)	14	c.1706G>A	p.(Arg569His)	sporadic	AR	Cone-rod dystrophy		NA
MD-0526	CCRD	CNGA3	NM_001298	8	c.829C>T	p.(Arg277Cys)	8	c.829C>T	p.(Arg277Cys)	AR	AR	Cone-rod dystrophy		Yes
MD-0994	CCRD	CNGA3	NM_001298	8	**c.1691G>C**	**p.(Arg564Thr) novel**	8	c.1691G>C	p.(Arg564Thr)	AR	AR	Cone-rod dystrophy		NA
MD-1030	OtherMD	CNGA3	NM_001298	8	c.1011C>A	p.(Tyr337*)	8	c.1642G>A	p.(Gly548Arg)	AR	AR	Cone-rod dystrophy		NA
MD-1117	OtherMD	CNGA3	NM_001298	8	c.778G>A	p.(Asp260Asn)	8	c.940_942del	p.(Ile314del)	AR	AR	Cone-rod dystrophy		NA
MD-1252	CCRD	CNGA3	NM_001298	8	c.811C>G	p.(Pro271Ala)	8	c.1201T>C	p.(Ser401Pro)	sporadic	AR	Cone-rod dystrophy		Yes
MD-1525	OtherMD	CNGA3	NM_001298	8	**c.1320del**	**p.(Trp440Cysfs*25) novel**	8	c.1320del	p.(Trp440Cysfs*25) novel	unknown	AR	Cone-rod dystrophy		NA
MD-1104	CCRD	CNGB1	NM_001297	IVS6	**c.412+1G>T**	**p.? novel**	IVS6	c.412+1G>T	p.?	AR	AR	Retinitis pigmentosa		NA
MD-0193	CCRD	CNGB3	NM_019098	10	c.1148del	p.(Thr383Ilefs*13)	10	c.1148del	p.(Thr383Ilefs*13)	sporadic	AR	Cone dystrophy		NA
MD-0397	CCRD	CNGB3	NM_019098	12	c.1432C>T	p.(Arg478*)	12	c.1432C>T	p.(Arg478*)	sporadic	AR	Cone dystrophy		NA
MD-0938	CCRD	CNGB3	NM_019098	5	c.503C>T	p.(Thr168Met)	5	c.503C>T	p.(Thr168Met)	sporadic	AR	Cone dystrophy		Yes
MD-0982	CCRD	CNGB3	NM_019098	7	c.886_896delinsT	p.(Thr296Tyrfs*9)	10	**c.1157T>A**	**p.(Val386Glu) novel**	sporadic	AR	Cone dystrophy		NA
MD-1023	CCRD	CNGB3	NM_019098	IVS6	c.852+1G>C	p.?	10	c.1148del	p.(Thr383Ilefs*13)	sporadic	AR	Cone dystrophy		NA
MD-1066	OtherMD	CNGB3	NM_019098	10	c.1148del	p.(Thr383Ilefs*13)	12	c.1432C>T	p.(Arg478*)	sporadic	AR	Cone dystrophy		NA
MD-1221	OtherMD	CNGB3	NM_019098	1	c.2T>C	p.Met1?	10	c.1148del	p.(Thr383Ilefs*13)	AR	AR	Cone dystrophy		NA
MD-1227	STGD	CNGB3	NM_019098	10	**c.1098_1101dup**	**p.(Ala368*) novel**	11	c.1208G>A	p.(Arg403Gln)	sporadic	AR	Cone dystrophy		Yes
MD-1492	CCRD	CNGB3	NM_019098	10	c.1148del	p.(Thr383Ilefs*13)	10	c.1148del	p.(Thr383Ilefs*13)	AR	AR	Cone dystrophy		NA
MD-1513	OtherMD	CNGB3	NM_019098	10	c.1148del	p.(Thr383Ilefs*13)	18	**c.2181_2184del**	**p.(Glu729Metfs*99) novel**	AR	AR	Cone dystrophy		NA
MD-1554	OtherMD	CNGB3	NM_019098	10	c.1148del	p.(Thr383Ilefs*13)	18	c.2181_2184del	p.(Glu729Metfs*99) novel	unknown	AR	Cone dystrophy		NA
MD-0211	STGD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	9	c.2843G>A	p.(Cys948Tyr)	sporadic	AR	Cone-rod dystrophy		NA
MD-0303	OtherMD	CRB1	NM_201253	9	c.2843G>A	p.(Cys948Tyr)	9	c.2843G>A	p.(Cys948Tyr)	sporadic	AR	Leber congenital amaurosis		Yes
MD-0351	OtherMD	CRB1	NM_201253	9	c.2843G>A	p.(Cys948Tyr)	9	c.3157A>G	p.(Met1053Val)	sporadic	AR	Cone-rod dystrophy		Yes
MD-0361	OtherMD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	9	**c.3055_3059dup**	**p.(Met1020Ilefs*4) novel**	sporadic	AR	Cone-rod dystrophy		NA
MD-0385	CCRD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	9	c.3299T>C	p.(Ile1100Thr)	sporadic	AR	Cone-rod dystrophy		NA
MD-0517	CCRD	CRB1	NM_201253	2	c.613_619del	p.(Ile205Aspfs*13)	6	c.1690G>T	p.(Asp564Tyr)	sporadic	AR	Leber congenital amaurosis		Yes
MD-0643	CCRD	CRB1	NM_201253	7	c.2234C>T	p.(Thr745Met)	9	c.2843T>A	p.(Cys948Tyr)	sporadic	AR	Leber congenital amaurosis		Yes
MD-0652	OtherMD	CRB1	NM_201253	6	c.1910C>T	p.(Pro637Leu)	6	c.1910C>T	p.(Pro637Leu)	AR	AR	Cone-rod dystrophy		NA
MD-0670	OtherMD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	7	c.2843G>A	p.(Cys948Tyr)	sporadic	AR	Cone-rod dystrophy		Yes
MD-0766	CCRD	CRB1	NM_201253	7	c.2290C>T	p.(Arg764Cys)	7	c.2290C>T	p.(Arg764Cys)	sporadic	AR	Cone-rod dystrophy		NA
MD-0824	OtherMD	CRB1	NM_201253	2	**c.635G>T**	**p.(Cys212Phe) novel**	11	**c.3946_3949del**	**p.(Leu1316Thrfs*24) novel**	AR	AR	Cone-rod dystrophy		NA
MD-0860	OtherMD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	9	c.3307G>A	p.(Gly1103Arg)	AR	AR	Cone-rod dystrophy		NA
MD-0876	CCRD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	6	c.1360G>C	p.(Gly454Arg)	AR	AR	Cone-rod dystrophy		NA
MD-0907	STGD	CRB1	NM_201253	2	c.481G>A	p.(Ala161Thr)	2	c.481G>A	p.(Ala161Thr)	sporadic	AR	Cone-rod dystrophy		NA
MD-1247	OtherMD	CRB1	NM_201253	6	c.1910C>T	p.(Pro637Leu)	6	c.1910C>T	p.(Pro637Leu)	sporadic	AR	Cone-rod dystrophy		Yes
MD-1253	OtherMD	CRB1	NM_201253	3	c.653-1G>T	p.?	8	c.2688T>A	p.(Cys986*)	unknown	AR	Cone-rod dystrophy		NA
MD-1266	OtherMD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	7	c.2843G>A	p.(Cys948Tyr)	sporadic	AR	Cone-rod dystrophy		NA
MD-1283	OtherMD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	7	c.2843G>A	p.(Cys948Tyr)	sporadic	AR	Cone-rod dystrophy		Yes
MD-1491	OtherMD	CRB1	NM_201253	2	c.498_506del	p.(Ile167_Gly169del)	7	c.2291G>A	p.(Arg764His)	AR	AR	Cone-rod dystrophy		NA
MD-0142	CCRD	CRX	NM_000554	3	c.121C>T	p.(Arg41Trp)				AR	AD	Cone-rod dystrophy	Inherited variant	Yes
MD-0258	STGD	CRX	NM_000554	3	c.121C>T	p.(Arg41Trp)				sporadic	AD	Cone-rod dystrophy	Incomplete penetrance	Yes
MD-0893	OtherMD	CRX	NM_000554	3	c.121C>T	p.(Arg41Trp)				sporadic	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-1289	OtherMD	CRX	NM_000554	3	c.121C>T	p.(Arg41Trp)				sporadic	AD	Cone-rod dystrophy	Inherited variant	Yes
MD-0269	STGD	EYS	NM_001142800	IVS29	c.6079-2A>G	p.?	IVS29	c.6079-2A>G	p.?	sporadic	AR	Retinitis pigmentosa		NA
MD-1435	CCRD	GNAT2	NM_005272	7	c.811_813del	p.(Lys271del)	7	c.811_813del	p.(Lys271del)	sporadic	AR	Cone dystrophy		NA
MD-1069	OtherMD	GUCY2D	NM_000180	13	c.2512C>T	p.(Arg838Cys)				AR	AD	Cone-rod dystrophy	Inherited variant, daugther affected, AD pattern confirmed	Yes
MD-1131	CCRD	GUCY2D	NM_000180	13	c.2513G>A	p.(Arg838His)				sporadic	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-0151	CCRD	KCNV2	NM_133497	1	c.778A>T	p.(Lys260*)	1	c.778A>T	p.(Lys260*)	AR	AR	Cone-rod dystrophy		NA
MD-0616	CCRD	KCNV2	NM_133497	1	**c.625G>T**	**p.(Glu209*) novel**	deletion entire gene			sporadic	AR	Cone-rod dystrophy		Yes
MD-0775	STGD	OPA1	NM_015560	9	c.1861C>T	p.(Gln621*)				sporadic	AD	Optic atrophy	Segregation studies unavailable	NA
MD-0073	OtherMD	PDE6C	NM_006204	4	**c.833C>T**	**p.(Ser278Phe) novel**	4	c.833C>T	p.(Ser278Phe)	sporadic	AR	Cone dystrophy		NA
MD-0117	OtherMD	PDE6C	NM_006204	4	c.833C>T	p.(Ser278Phe)	4	c.833C>T	p.(Ser278Phe)	sporadic	AR	Cone dystrophy		NA
MD-1020	OtherMD	PDE6C	NM_006204	1	c.311G>A	p.(Arg104Gln)	1	c.311G>A	p.(Arg104Gln)	AR	AR	Cone dystrophy		NA
MD-1504	CCRD	PDE6C	NM_006204	IVS1	**c.480+1G>A**	**p.? novel**	IVS1	c.480+1G>A	p.?	AR	AR	Cone dystrophy		NA
MD-1514	CCRD	PDE6C	NM_006204	13	**c.1651A>G**	**p.(Thr551Ala) novel**	13	c.1651A>G	p.(Thr551Ala)	AR	AR	Cone dystrophy		NA
MD-0403	STGD	PLA2G5	NM_000929	5	**c.309C>A**	**p.(Cys103*) novel**	5	c.309C>A	p.(Cys103*)	AR	AR	Fundus albipunctatus like		NA
MD-1458	STGD	PLA2G5	NM_000929	5	c.309C>A	p.(Cys103*)	5	c.309C>A	p.(Cys103*)	sporadic	AR	Fundus albipunctatus like		Yes
MD-0100	CCRD	PROM1	NM_006017	IVS5	c.630+1G>A	p.?	IVS5	c.630+1G>A	p.?	sporadic	AR	Cone-rod dystrophy		Yes
MD-0383	STGD	PROM1	NM_006017	12	c.1354dup	p.(Tyr452Leufs*13)	12	c.1354dup	p.(Tyr452Leufs*13)	sporadic	AR	Cone-rod dystrophy		NA
MD-0649	CCRD	PROM1	NM_006017	IVS5	c.630+1G>A	p.?	IVS5	c.630+1G>A	p.?	sporadic	AR	Cone-rod dystrophy		NA
MD-0654	CCRD	PROM1	NM_006017	12	c.1414del	p.(Arg472Glufs*18)	12	c.1414del	p.(Arg472Glufs*18)	sporadic	AR	Cone-rod dystrophy		NA
MD-0682	OtherMD	PROM1	NM_006017	12	c.1354dup	p.(Tyr452Leufs*13)	12	c.1354dup	p.(Tyr452Leufs*13)	sporadic	AR	Cone-rod dystrophy		NA
MD-0762	CCRD	PROM1	NM_006017	IVS17	c.1984-1G>T	p.?	IVS17	c.1984-1G>T	p.?	sporadic	AR	Cone-rod dystrophy		Yes
MD-0803	CCRD	PROM1	NM_006017	12	c.1354dup	p.(Tyr452Leufs*13)	12	c.1354dup	p.(Tyr452Leufs*13)	sporadic	AR	Cone-rod dystrophy		Yes
MD-0838	CCRD	PROM1	NM_006017	12	c.1354dup	p.(Tyr452Leufs*13)	12	c.1354dup	p.(Tyr452Leufs*13)	sporadic	AR	Cone-rod dystrophy		Yes
MD-0873	CCRD	PROM1	NM_006017	IVS17	c.1984-1G>T	p.?	IVS17	c.1984-1G>T	p.?	AR	AR	Cone-rod dystrophy		Yes
MD-1037	OtherMD	PROM1	NM_006017	12	c.1354dup	p.(Tyr452Leufs*13)	12	**c.1363G>C**	**p.(Gly455Arg) novel**	AR	AR	Cone-rod dystrophy		Yes

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked

Table S2. Description of genetics and clinical findings in 677 characterized *a priori* ar/sMD families. Novel variants are highlighted in gray.

Family	Suspected diagnosis	Gene	RefSeq_NM	Allele1_exon	Allele1_cDNA	Allele1_protein	Allele2_exon	Allele2_cDNA	Allele2_protein	Inheritance a priori	Final inheritance	Clinical reclassification	Genetic reclassification comments	Segregation
MD-1487	OtherMD	PROM1	NM_006017	17	**c.1912-1G>T**	**p.? novel**	deletion exons 9-12			AR	AR	Cone-rod dystrophy		Yes
RP-0855	STGD	PROM1	NM_006017	12	c.1354dup	p.(Tyr452Leufs*13)	12	c.1354dup	p.(Tyr452Leufs*13)	AR	AR	Cone-rod dystrophy		Yes
RP-1852	OtherMD	PROM1	NM_006017	12	c.1435G>A	p.(Gly479Arg)	12	c.1435G>A	p.(Gly479Arg)	AR	AR	Cone-rod dystrophy		NA
MD-0236	STGD	PRPH2	NM_000322	1	c.499G>A	p.(Gly167Ser)				AR	AD	Cone-rod dystrophy	Inherited variant	Yes
MD-0381	OtherMD	PRPH2	NM_000322	2	c.584G>T	p.(Arg195Leu)				AR	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-0489	STGD	PRPH2	NM_000322	1	c.493T>C	p.(Cys165Arg)				AR	AD	Cone-rod dystrophy	Incomplete penetrance	Yes
MD-0511	STGD	PRPH2	NM_000322	1	**c.52C>T**	**p.(Gln18*) novel**				unknown	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-0972	OtherMD	PRPH2	NM_000322	2	**c.734_737dup**	**p.(Trp246Cysfs*56) novel**				AR	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-1345	OtherMD	PRPH2	NM_000322	2	c.628C>T	p.(Pro210Ser)				sporadic	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-1389	STGD	PRPH2	NM_000322	2	c.584G>T	p.(Arg195Leu)				sporadic	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-1395	OtherMD	PRPH2	NM_000322	2	c.584G>T	p.(Arg195Leu)				AR	AD	Cone-rod dystrophy	Cousin affected	NA
MD-1449	CCRD	PRPH2	NM_000322	2	**c.649_650ins17**	**p.(Ser217Ilefs*45) novel**				sporadic	AD	Cone-rod dystrophy	Segregation studies unavailable	NA
MD-0312	CCRD	RAB28	NM_004249	IVS2	c.172+1G>C	p.?	IVS2	c.172+1G>C	p.?	sporadic	AR	Cone-rod dystrophy		Yes
MD-0448	CCRD	RAB28	NM_004249	7	c.651T>G	p.(Cys217Trp)	7	c.651T>G	p.(Cys217Trp)	sporadic	AR	Cone-rod dystrophy		Yes
MD-0491	OtherMD	RDH12	NM_152443	8	c.701G>A	p.(Arg234His)	8	**c.795C>A**	**p.(Ser265Arg) novel**	sporadic	AR	Cone-rod dystrophy		Yes
MD-0639	CCRD	RDH12	NM_152443	8	c.701G>A	p.(Arg234His)	8	c.784dup	p.(Ala262Glyfs*11)	AR	AR	Cone-rod dystrophy		NA
MD-0970	CCRD	RDH12	NM_152443	5	c.278T>C	p.(Leu93Pro)	5	c.278T>C	p.(Leu93Pro)	sporadic	AR	Cone-rod dystrophy		Yes
MD-1078	OtherMD	RDH12	NM_152443	5	c.278T>C	p.(Leu93Pro)	5	c.278T>C	p.(Leu93Pro)	sporadic	AR	Cone-rod dystrophy		NA
MD-1413	CCRD	RDH12	NM_152443	5	c.278T>C	p.(Leu93Pro)	7	c.505C>T	p.(Arg169Trp)	AR	AR	Cone-rod dystrophy		Yes
MD-1378	CCRD	RHO	NM_000539	4	**c.826T>G**	**p.(Phe276Val) novel**				sporadic	AD	Retinitis pigmentosa	Segregation studies unavailable	NA
MD-0620	OtherMD	RLBP1	NM_000326	6	c.504_508del	p.(Ser168Argfs*5)	6	c.504_508del	p.(Ser168Argfs*5)	sporadic	AR	Retinitis pigmentosa		Yes
MD-0693	STGD	RP2	NM_006915	2	c.409_411del	p.(Ile137del)				sporadic	XL	Retinitis pigmentosa	Segregation studies unavailable	NA
MD-0507	CCRD	RPGR	NM_001034853	15	c.2405_2406del	p.(Glu802Glyfs*32)				sporadic	XL	Retinitis pigmentosa	Segregation studies unavailable	NA
MD-0983	CCRD	RPGR	NM_001034853	15	**c.3382C>T**	**p.(Arg1128*) novel**				unknown	XL	Retinitis pigmentosa	Segregation studies unavailable	NA
MD-1062	OtherMD	RPGR	NM_001034853	8	c.872del	p.(Asn291Ilefs*7)				sporadic	XL	Retinitis pigmentosa	Segregation studies unavailable	NA
MD-0327	CCRD	RPGRIP1	NM_020366	IVS17	**c.2895+1G>T**	**p.? novel**	IVS17	c.2895+1G>T	p.?	sporadic	AR	Leber congenital amaurosis		Yes
MD-0671	STGD	RS1	NM_000330	5	c.410T>C	p.(Leu137Pro)				AR	XL	Retinoschisis	Affected males in the family. Inherited variant	Yes
MD-1398	OtherMD	RS1	NM_000330	3	**c.150G>T**	**p.(Trp50Cys) novel**				sporadic	XL	Retinoschisis	Inherited variant	Yes
MD-1344	CCRD	TRPM1	NM_002420	6	c.428-1G>C	p.?	23	c.2782C>T	p.(Arg928Trp)	sporadic	AR	Congenital stationary night blindness		NA
MD-0179	CCRD	TTLL5	NM_015072	5	**c.211C>T**	**p.(Arg71*) novel**	21	**c.2029C>T**	**p.(Arg677*) novel**	AR	AR	Cone dystrophy		NA
MD-0884	CCRD	USH2A	NM_206933	44	c.8693A>C	p.(Tyr2898Ser)	63	c.13531G>A	p.(Ala4511Thr)	sporadic	AR	Retinitis pigmentosa		Yes

Abbreviations: AR: autosomal recessive; AD: autosomal dominant; CCRD, cone and cone-rod dystrophies; IVS, intron; NA, not available; STGD, Stargardt disease;otherMD, other maculopathies; XL: X-linked