ABCA4 Mutations	Reported Pathogenicity	Subjects with Mutation
c.570+1888 A>G (homozygous)	Likely benign	JC_10296, JC_10461, JC_10469
c.676 C>T; p.226 Arg>Cys	Probably Damaging	KS_10306
c.769-1718_c.769-1717insTAT	Likely benign	JC_10461
c.1240-14 C>T	Not likely pathogenic	TC_1048
c.1622 T>C; p.541 Leu>Pro	Reported mutation[39, 70]	KS_10150
c.1804 C>T; p.602 Arg>Trp	Reported mutation[39, 71, 72]	KS_10108
c.1957 C>T; p.653 Arg>Cys	Reported mutation[73]	KS_10241, KS_10242
c.2041 C>T; p.681 Arg>Stop	Reported mutation[70]	DH_1162
c.2160+1 G>T*	Splice site mutation (Likely damaging due to altered canonical splice site; Human Splice Finder v3.0[27])	KS_1027, TC_1048
c.2588 G>C; p.863 Gly>Ala	Reported mutation[39, 70, 74, 75]	KS_10108
c.2919-927 T>A	VUS (Altered splice enhancer; Human Splice Finder v3.0[27])	KS_10306
c.3113 C>T; p.1038 Ala>Val	Reported mutation[70, 74, 75]	KS_10150
c.3259 G>A; p.1087 Glu>Lys	Reported mutation[76]	DH_10019
c.4469 G>A; p.1490 Cys>Tyr	Probable disease-causing[77]	DH_1158
c.4577 C>T; p.1526 Thr>Met	Reported mutation[39]	JC_10461, JC_10469
c.5018+8 A>G	VUS (No impact on splicing; Human Splice Finder v3.0[27])	KS_10306
c.5603 A>T; p.1868 Asn>Ile	VUS**;** Benign; Non-disease-causing[78]	KS_1027, TC_1048, DH_1158
c.5682 G>C; p.1894 Leu>Leu	Not likely pathogenic; Non-disease-causing	TC_1048, DH_1158
c.5714+5 G>A	Reported mutation[39, 70, 74]	KS_10150
c.5882 G>A; p.1961 Gly>Glu	Reported mutation[39, 54, 70, 75]	KS_10242, JC_10296, JC_10461, JC_10469
c.6079 C>T; p.2027 Leu>Phe	Reported mutation[77]	JC_10222
c.6320 G>A; p.2107 Arg>His	Reported mutation[76]	JC_10222
c.6449 G>A; p.2150 Cys>Tyr	Reported mutation[76]	JC_10296
c.6730-579 T>C	VUS (Altered splice enhancer & silencer; Human Splice Finder v3.0[27])	KS_10306

VUS = Variant of Unknown Significance
*Novel Mutation (novel variant at known mutation site)

Supplementary Table 1. Reported pathogenicity of the complete list of *ABCA4* mutations.