**ABCA4 Mutations** | **Reported Pathogenicity** | **Subjects with Mutation**
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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\(^{39}\) | 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\(^{10, 74, 75}\) | KS_10150

c.3259 G>A; p.1087 Glu>Lys | Reported mutation\(^{16}\) | 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 \(^{78}\) | 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\(^{49, 54, 70, 75}\) | 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\(^{16}\) | JC_10222

c.6449 G>A; p.2150 Cys>Tyr | Reported mutation\(^{46}\) | 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.