

**Supplemental table 1. Summary of genetic findings in 345 patients with STGD1**

Study ID	Institute	Variant 1		Variant 2		Variant 3		Variant 4		Segregation
		Nucleotide change, amino acid change/effect	Pathogenicity	Nucleotide change, amino acid change/effect	Pathogenicity	Nucleotide change, amino acid change/effect	Pathogenicity	Nucleotide change, amino acid change/effect	Pathogenicity	
11002	JHU	<i>c.1267C&gt;T, p.His423Tyr</i>	LLP	<i>c.3608G&gt;A, p.Gly1203Glu</i>	LLP	<i>c.5460+4A&gt;G, splice site alteration</i>	LP	<i>c.5461-10T&gt;C, splice site alteration</i>	LP	
11004	JHU	<i>c.1957C&gt;T, p.Arg653Cys</i>	LP	<i>c.3602T&gt;G, p.Leu1201Arg</i>	LLP					
11006	JHU	<i>c.5461-10T&gt;C, splice site alteration</i>	LP	<i>c.5882G&gt;A, p.Gly1961Glu</i>	LP					
11007	JHU	<i>c.4363T&gt;C, p.Cys1455Arg</i>	LP	<i>c.5882G&gt;A, p.Gly1961Glu</i>	LP					
11009	JHU	<i>c.4540-2A&gt;G, splice site alteration</i>	LP	<i>c.5882G&gt;A, p.Gly1961Glu</i>	LP					
11010	JHU	<i>c.5461-10T&gt;C, splice site alteration</i>	LP	<i>c.5714+5G&gt;A, splice site alteration</i>	LP					
11015	JHU	<i>c.1A&gt;G, p.Met1Val (splice site alteration)</i>	LP	<i>c.5882G&gt;A, p.Gly1961Glu</i>	LP					
11016	JHU	<i>c.3758C&gt;T, p.Thr1253Met</i>	LLP	<i>c.4139C&gt;T, p.Pro1380Leu</i>	LP	<i>c.5882G&gt;A, p.Gly1961Glu</i>	LP			
11017	JHU	<i>c.2966T&gt;C, p.Val989Ala</i>	LP	<i>c.5461-10T&gt;C, splice site alteration</i>	LP					
11018	JHU	<i>c.2915C&gt;A, p.Thr972Asm</i>	LP	<i>c.5882G&gt;A, p.Gly1961Glu</i>	LP					

11019	JHU	c.2453G>A, p.Gly818Glu	LP	c.4532C>A, p.Pro1511His	LP		
11020	JHU	c.1927G>A, p.Val643Met	LP	c.3259G>A, p.Glu1087Lys	LP		
11024	JHU	c.2546T>C, p.Val849Ala	LLP	c.6320G>A, p.Arg2107His	LP		
11025	JHU	c.3322C>T, p.Arg1108Cys	LP	c.6079C>T, p.Leu2027Phe	LP		
11026	JHU	c.1A>G, p.Met1Val (splice site alteration)	LP	c.5882G>A, p.Gly1961Glu	LP		
11027	JHU	c.2971G>C, p.Gly991Arg	LP	c.5337C>G, p.Tyr1779Ter	LP		
11028	JHU	c.237T>C, p.Asn79Asn	LLP	c.6320G>A, p.Arg2107His	LP	c.6449G>A, p.Cys2150Tyr	LP
11029	JHU	c.16C>T, p.Gln6Ter	LP	c.2588G>C, p.Gly863Ala	LP		
11030	JHU	c.3322C>T, p.Arg1108Cys	LP	c.6079C>T, p.Leu2027Phe	LP		
11031	JHU	c.2608C>T, p.Pro870Ser	LP	c.2966T>C, p.Val989Ala	LP	c.6320G>A, p.Arg2107His	LP
11032	JHU	c.283T>C, p.Ser95Pro	LP	c.283T>C, p.Ser95Pro	LP		
11033	JHU	c.2588G>C, p.Gly863Ala	LP	c.3210_3211insGT, p.Ser1071CysfsTer14	LP		
11034	JHU	c.454C>T, p.Arg152Ter	LP	c.6079C>T, p.Leu2027Phe	LP	c.6148G>C, p.Val2050Leu	LP

12004	GBMC	c.634C>T, p.Arg212Cys	LP	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP
12005	GBMC	c.4234C>T, p.Gln1412Ter	LP	<i>c.5018+5G&gt;A, splice site alteration</i>	LP		
12006	GBMC	c.2966T>C, p.Val989Ala	LP	<i>c.3287C&gt;T, p.Ser1096Leu</i>	LP	<i>c.6522G&gt;A, p.Pro2174Pro</i>	LLP
12008	GBMC	c.2977_2984delGACAT TGA, p.Asp993AsnfsTer27	LP	c.3602T>G, p.Leu1201Arg	LLP	c.6320G>A, p.Arg2107His	LP
12009	GBMC	c.5882G>A, p.Gly1961Glu	LP	c.6005+1G>T, splice site alteration	LP		
12011	GBMC	c.2588G>C, p.Gly863Ala	LP	<i>c.2383-1G&gt;A, splice site alteration</i>	LP		
12013	GBMC	c.859-9T>C, splice site alteration	LP	c.4139C>T, p.Pro1380Leu	LP		
12015	GBMC	c.302+1G>A, splice site alteration	LP	<i>c.4774-30G&gt;A, deep intron</i>	LLP	c.6320G>A, p.Arg2107His	LP
12017	GBMC	c.4139C>T, p.Pro1380Leu	LP	c.5461-10T>C, splice site alteration	LP		
12019	GBMC	<i>c.4231G&gt;A, p.Gly1411Arg</i>	LP	c.6729+5_19delGTTGG CCCTGGGGCA, splice site alteration	LP		
12020	GBMC	c.768G>T, p.Val256Val (splice site alteration)	LP	c.2609C>T, p.Pro870Leu	LP		
12022	GBMC	c.1574T>C, p.Phe525Ser	LP	c.2588G>C, p.Gly863Ala	LP		

12023	GBMC	c.4139C>T, p.Pro1380Leu	LP	c.4594G>A, p.Asp1532Asn	LP		
12025	GBMC	c.1A>G, p.Met1Val (splice site alteration)	LP	c.4139C>T, p.Pro1380Leu	LP		
12028	GBMC	c.3758C>T, p.Thr1253Met	LLP	c.4793C>A, p.Ala1598Asp	LP	c.5882G>A, p.Gly1961Glu	LP
12030	GBMC	c.4139C>T, p.Pro1380Leu	LP	c.5311G>A, p.Gly1771Arg	LP		
12032	GBMC	c.5018+8A>G, possible splice site alteration	LLP	<i>c.5584+14G&gt;A, deep intron</i>	LLP		
12036	GBMC	c.1819G>A, p.Gly607Arg	LP	c.2971G>C, p.Gly991Arg	LP		
12037	GBMC	c.1937+1G>A, splice site alteration	LP	c.3899G>A, p.Arg1300Gln	LLP	c.5056G>A, p.Val1686Met	LP
12038	GBMC	c.3758C>T, p.Thr1253Met	LLP	c.5461-10T>C, splice site alteration	LP	c.5882G>A, p.Gly1961Glu	LP
12039	GBMC	c.1622T>C, p.Leu541Pro	LP	c.3056C>T, p.Thr1019Met	LP	c.3113C>T, p.Ala1038Val	LLP
12040	GBMC	c.634C>T, p.Arg212Cys	LP	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP
12042	GBMC	c.194G>A, p.Gly65Glu	LP	c.5196+1G>A, splice site alteration	LP		
12044	GBMC	c.3322C>T, p.Arg1108Cys	LP	c.5461-10T>C, splice site alteration	LP		
12046	GBMC	c.161G>A, p.Cys54Tyr	LP	c.3113C>T, p.Ala1038Val	LLP		

12048	GBMC	c.2401G>A, p.Ala801Thr	LP	c.5461-10T>C, splice site alteration	LP		
12050	GBMC	c.618C>G, p.Ser206Arg	LLP	c.1927G>A, p.Val643Met	LP	c.2977_2984delGACAT TGA, p.Asp993AsnfsTer27	LP
16002	PENN	c.1022A>G, p.Glu341Gly	LP	c.1822T>A, p.Phe608Ile	LP		
16006	PENN	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5882G>A, p.Gly1961Glu	LP
16007	PENN	c.5461-10T>C, splice site alteration	LP	c.1086T>A, p.Tyr362Ter	LP		
16009	PENN	c.2804T>C, p.Val935Ala	LP	c.5714+5G>A, splice site alteration	LP		
16010	PENN	c.4919G>A, p.Arg1640Gln	LP	c.5882G>A, p.Gly1961Glu	LP		
16011	PENN	c.454C>T, p.Arg152Ter	LP	c.5882G>A, p.Gly1961Glu	LP		
16012	PENN	c.2588G>C, p.Gly863Ala	LP	c.4469G>A, p.Cys1490Tyr	LP		
16013	PENN	c.3364G>A, p.Glu1122Lys	LP	c.5882G>A, p.Gly1961Glu	LP		
16014	PENN	c.4530_4531insC, p.Pro1511ProfsTer44	LP	c.5114G>A, p.Arg1705Gln	LP		
16015	PENN	c.768G>T, p.Val256Val (splice site alteration)	LP	c.5882G>A, p.Gly1961Glu	LP		
16016	PENN	c.658C>T, p.Arg220Cys	LP	c.3051-1G>A, splice site alteration	LP		

16018	PENN	c.2588G>C, p.Gly863Ala	LP	c.5018+2T>C, splice site alteration	LP		
16020	PENN	c.454C>T, p.Arg152Ter	LP	c.5882G>A, p.Gly1961Glu	LP		
16026	PENN	c.2461T>A, p.Trp821Arg	LP	c.2894A>G, p.Asn965Ser	LP		
16030	PENN	c.4139C>T, p.Pro1380Leu	LP	c.5882G>A, p.Gly1961Glu	LP		
16031	PENN	c.4577C>T, p.Thr1526Met	LP	c.6089G>A, p.Arg2030Gln	LP		
16032	PENN	c.768G>T, p.Val256Val (splice site alteration)	LP	c.4577C>T, p.Thr1526Met	LP		
16033	PENN	exon7del, large exonic deletion	LP	c.4457C>T, p.Pro1486Leu	LP		
16036	PENN	c.3386G>T, p.Arg1129Leu	LP	c.5819T>C, p.Leu1940Pro	LP		
16037	PENN	c.2588G>C, p.Gly863Ala	LP	c.3322C>T, p.Arg1108Cys	LP		
16039	PENN	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5773A>G, p.Arg1925Gly	LP
16040	PENN	c.2564G>A, p.Trp855Ter	LP	c.4506C>T, p.Cys1502Cys	LLP		
16041	PENN	c.2005_2006delAT, p.Met669AspfsTer96	LP	c.5882G>A, p.Gly1961Glu	LP		
16042	PENN	c.4222T>C, p.Trp1408Arg	LP	c.5714+5G>A, splice site alteration	LP		

16043	PENN	c.3056C>T, p.Thr1019Met	LP	c.5882G>A, p.Gly1961Glu	LP		
17001	RFSW	c.2588G>C, p.Gly863Ala	LP	<i>c.6181_6184delACTG, p.Thr2061SerfsTer53</i>	LP		
17002	RFSW	c.2588G>C, p.Gly863Ala	LP	c.5084C>A, p.Ala1695Asp	LP		
17003	RFSW	c.3602T>G, p.Leu1201Arg	LLP	c.6764G>T, p.Ser2255Ile	LLP		
17004	RFSW	c.2971G>C, p.Gly991Arg	LP	<i>c.4504T&gt;G, p.Cys1502Gly</i>	LP		
17005	RFSW	c.4222T>C, p.Trp1408Arg	LP	c.4918C>T, p.Arg1640Trp	LP	c.5882G>A, p.Gly1961Glu	LP
17006	RFSW	c.1927G>A, p.Val643Met	LP	c.1937+1G>A, splice site alteration	LP	c.3602T>G, p.Leu1201Arg	LLP
17007	RFSW	c.2588G>C, p.Gly863Ala	LP	c.4577C>T, p.Thr1526Met	LP		
17008	RFSW	c.4577C>T, p.Thr1526Met	LP	c.5196+1137G>A, deep intron	LLP		
17009	RFSW	c.4139C>T, p.Pro1380Leu	LP	c.5882G>A, p.Gly1961Glu	LP		
17011	RFSW	c.1015T>G, p.Trp339Gly	LP	c.2300T>A, p.Val767Asp	LP		
17012	RFSW	c.1715G>A, p.Arg572Gln	LP	c.1919C>T, p.Pro640Leu	LP	c.2588G>C, p.Gly863Ala	LP
17019	RFSW	c.3386G>T, p.Arg1129Leu	LP	c.6221G>T, p.Gly2074Val	LP		

17020	RFSW	c.4919G>A, p.Arg1640Gln	LP	c.5882G>A, p.Gly1961Glu	LP		
17022	RFSW	c.634C>T, p.Arg212Cys	LP	c.5882G>A, p.Gly1961Glu	LP		
17023	RFSW	c.3602T>G, p.Leu1201Arg	LLP	c.5077G>A, p.Val1693Ile	LP	c.6386+2C>G, splice site alteration	LP
17024	RFSW	c.5395A>G, p.Asn1799Asp	LP	c.768G>T, p.Val256Val (splice site alteration)	LP		
17027	RFSW	c.2819C>G, p.Pro940Arg	LLP				
17028	RFSW	c.768G>T, p.Val256Val (splice site alteration)	LP				
17030	RFSW	<i>c.1529T&gt;C, p.Leu510Pro</i>	LP	c.5882G>A, p.Gly1961Glu	LP		
17032	RFSW	c.5882G>A, p.Gly1961Glu	LP	c.6088C>T, p.Arg2030Ter	LP		
17033	RFSW	c.4771G>A, p.Gly591Arg	LP				
17034	RFSW	<i>c.67-1G&gt;C, splice site alteration</i>	LP				
17037	RFSW	c.5196+1G>A, Splicedonor	LP	c.5882G>A, p.Gly1961Glu	LP		
17038	RFSW	<i>c.1610G&gt;A, p.Arg537His</i>	LP	c.4577C>T, p.Thr1526Met	LP		
17039	RFSW	c.32T>C, p.Leu11Pro	LP	c.4469G>A, p.Cys1490Tyr	LP		✓



17040	RFSW	c.768G>T, p.Val256Val (splice site alteration)	LP			
17041	RFSW	c.2588G>C, p.Gly863Ala	LP	c.4139C>T, p.Pro1380Leu	LP	
17042	RFSW	c.4538A>G, p.Gln1513Arg (splice site alteration)	LP	c.6320G>A, p.Arg2107His	LP	
17044	RFSW	c.1648G>A, p.Gly550Arg	LP	c.3385C>T, p.Arg1129Cys	LP	
17045	RFSW	c.3389T>C, p.Ile1130Thr	LP	<i>c.4516G&gt;A, p.Ala1506Thr</i>	LP	c.5882G>A, p.Gly1961Glu
17046	RFSW	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5882G>A, p.Gly1961Glu
17047	RFSW	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5395A>G, p.Asn1799Asp
17048	RFSW	c.3364G>A, p.Glu1122Lys	LP			
18007	MEC	c.2588G>C, p.Gly863Ala	LP	c.4139C>T, p.Pro1380Leu	LP	
18009	MEC	c.3322C>T, p.Arg1108Cys	LP			
18011	MEC	c.3385C>T, p.Arg1129Cys	LP			
18013	MEC	c.2894A>G, p.Asn965Ser	LP	c.4222T>C, p.Trp1408Arg	LP	c.4918C>T, p.Arg1640Trp
18014	MEC	c.4253+4C>T, splice site alteration	LP	c.4463G>A, p.Cys1488Tyr	LP	c.6148G>C, p.Val2050Leu

18016	MEC	<i>c.160+5G&gt;A, splice site alteration</i>	LP	<i>c.3259G&gt;A, p.Glu1087Lys</i>	LP		
18018	MEC	<i>c.2588G&gt;C, p.Gly863Ala</i>	LP	<i>c.5172G&gt;A, p.Trp1724Ter</i>	LP		
18020	MEC	<i>c.1529T&gt;C, p.Leu510Pro</i>	LP				
18021	MEC	<i>c.2300T&gt;A, p.Val767Asp</i>	LP	<i>c.4918C&gt;T, p.Arg1640Trp</i>	LP		
18023	MEC	<i>c.45G&gt;A, p.Trp15Ter</i>	LP	<i>c.2690C&gt;T, p.Thr897Ile</i>	LP		
18024	MEC	<i>c.2915C&gt;A, p.Thr972Asm</i>	LP	<i>c.5461-10T&gt;C, splice site alteration</i>	LP		
18025	MEC	<i>c.4253+4C&gt;T, splice site alteration</i>	LP	<i>c.5318C&gt;T, p.Ala1773Val</i>	LP		
18027	MEC	<i>c.3385C&gt;T, p.Arg1129Cys</i>	LP				
18028	MEC	<i>c.2564G&gt;A, p.Trp855Ter</i>	LP	<i>c.4506C&gt;T, p.Cys1502Cys</i>	LLP	<i>c.4709C&gt;T, p.Pro1570Leu</i>	LLP
18029	MEC	<i>c.3259G&gt;A, p.Glu1087Lys</i>	LP	<i>c.160+5G&gt;A, splice site alteration</i>	LP		
18030	MEC	<i>c.4253+4C&gt;T, splice site alteration</i>	LP	<i>c.5882G&gt;A, p.Gly1961Glu</i>	LP		
18032	MEC	<i>c.3113C&gt;T, p.Ala1038Val</i>	LLP	<i>c.3259G&gt;A, p.Glu1087Lys</i>	LP		
18033	MEC	<i>c.4139C&gt;T, p.Pro1380Leu</i>	LP				

18034	MEC	c.2588G>C, p.Gly863Ala	LP	c.6383A>G, p.His2128Arg	LP		
18035	MEC	c.868C>T, p.Arg290Trp	LP				
18036	MEC	c.5461-10T>C, splice site alteration	LP				
19001	CC	c.5077G>A, p.Val1693Ile	LP	c.834delT, p.Ser278SerfsTer22	LP		
19002	CC	c.466A>G, p.Ile156Val	LLP	c.4469G>A, p.Cys1490Tyr	LP		
19004	CC	c.4222T>C, p.Trp1408Arg	LP	c.4918C>T, p.Arg1640Trp	LP	c.5882G>A, p.Gly1961Glu	LP
19005	CC	c.4537_4538insC, p.Pro1513GlnfsTer42	LP	c.5882G>A, p.Gly1961Glu	LP		
19006	CC	c.3260A>G, p.Glu1087Gly	LP	c.6089G>A, p.Arg2030Gln	LP		
19007	CC	c.2588G>C, p.Gly863Ala	LP	c.3041T>G, p.Leu1014Arg	LP		
19008	CC	c.2300T>A, p.Val767Asp	LP	c.5882G>A, p.Gly1961Glu	LP		
19009	CC	c.2588G>C, p.Gly863Ala	LP	c.6386+2C>G, splice site alteration	LP		
19010	CC	c.3113C>T, p.Ala1038Val	LLP				
19011	CC	c.5114G>T, p.Arg1705Leu	LP	c.6079C>T, p.Leu2027Phe	LP		

19012	CC	c.4919G>A, p.Arg1640Gln	LP	c.5461-10T>C, splice site alteration	LP		
19013	CC	c.1086T>A, p.Tyr362Ter	LP	c.6089G>A, p.Arg2030Gln	LP		
19014	CC	c.5882G>A, p.Gly1961Glu	LP				
19015	CC	<i>c.160+5G&gt;C, splice site alteration</i>	LP	c.5714+5G>A, splice site alteration	LP		
19016	CC	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP		
19019	CC	c.1919C>T, p.Pro640Leu	LP	c.5461-10T>C, splice site alteration	LP		
19020	CC	c.4139C>T, p.Pro1380Leu	LP				
19021	CC	c.4919G>A, p.Arg1640Gln	LP	c.5882G>A, p.Gly1961Glu	LP		
19022	CC	c.1853G>A, p.Gly618Glu	LP				
19023	CC	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5882G>A, p.Gly1961Glu	LP
19024	CC	c.1610G>A, p.Arg537His	LP	c.4577C>T, p.Thr1526Met	LP		
14003	MEH	c.1317G>A, p.Trp439Ter	LP	c.2588G>C, p.Gly863Ala	LP	c.5908C>T, p.Leu1970Phe	LP
14006	MEH	c.180delG, p.Ala60AlafsTer18	LP	c.4328G>A, p.Arg1443His	LP		

14008	MEH	c.4926C>G, p.Ser1642Arg	LP	c.5041_5055delGTGGT TGCCATCTGC, p.Val1681_Cys1685del	LP		
14010	MEH	c.3191-1G>T, splice site alteration	LP	c.4139C>T, p.Pro1380Leu	LP		
14011	MEH	c.5714+5G>A, splice site alteration	LP	c.6209C>G, p.Thr2070Arg	LP		
14012	MEH	c.2942C>T, p.Pro981Leu	LP	c.3210_3211insGT, p.Ser1071CysfsTer14	LP	c.6529G>A, p.Asp2177Asn	LLP
14013	MEH	c.3392_3393delCCinsG, p.Ala1131GlyfsTer17	LP	c.5882G>A, p.Gly1961Glu	LP		
14015	MEH	c.2861A>C, p.Tyr954Ser	LP	c.5461-10T>C, splice site alteration	LP		
14017	MEH	c.466A>G, p.Ile156Val	LLP	c.4363T>C, p.Cys1455Arg	LP	c.5516T>C, p.Phe1839Ser	LP
14019	MEH	c.2588G>C, p.Gly863Ala	LP	c.6148G>C, p.Val2050Leu	LP		
14020	MEH	c.2588G>C, p.Gly863Ala	LP	c.3364G>A, p.Glu1122Lys	LP	c.6088C>T, p.Arg2030Ter	LP
14021	MEH	c.1957C>T, p.Arg653Cys	LP	c.4139C>T, p.Pro1380Leu	LP	c.6088C>T, p.Arg2030Ter	LP
14022	MEH	c.2588G>C, p.Gly863Ala	LP	c.5161_5162delAC, p.Thr1721TfsTer65	LP		
14024	MEH	c.5461-10T>C, splice site alteration	LP	c.6089G>A, p.Arg2030Gln	LP		
14025	MEH	c.1957C>T, p.Arg653Cys	LP	c.6089G>A, p.Arg2030Gln	LP		

14026	MEH	c.1757A>G, p.Asp586Gly	LP	c.5882G>A, p.Gly1961Glu	LP					
14027	MEH	c.4222T>C, p.Trp1408Arg	LP	c.4222T>C, p.Trp1408Arg	LP	c.4918C>T, p.Arg1640Trp	LP	c.4918C>T, p.Arg1640Trp	LP	✓
14028	MEH	c.1726G>C, p.Asp576His	LP	c.4469G>A, p.Cys1490Tyr	LP					
14029	MEH	c.3322C>T, p.Arg1108Cys	LP	c.6079C>T, p.Leu2027Phe	LP					
14031	MEH	c.6089G>A, p.Arg2030Gln	LP	c.6118C>T, p.Arg2040Ter	LP					
14034	MEH	c.4139C>T, p.Pro1380Leu	LP	c.4594G>A, p.Asp1532Asn	LP					
14035	MEH	c.658C>T, p.Arg220Cys	LP	c.2588G>C, p.Gly863Ala	LP					
14036	MEH	c.872C>T, p.Pro291Leu	LP	c.1531C>T, p.Arg511Cys	LP	c.5882G>A, p.Gly1961Glu	LP			
14037	MEH	c.1531C>T, p.Arg511Cys	LP	<i>c.5216_5218dupGCT, p.Ala1739dup</i>	LP	c.5882G>A, p.Gly1961Glu	LP			
14038	MEH	c.5882G>A, p.Gly1961Glu	LP	c.5882G>A, p.Gly1961Glu	LP					✓
14039	MEH	c.1411G>A, p.Glu471Lys	LLP	c.2588G>C, p.Gly863Ala	LP	c.4594G>A, p.Asp1532Asn	LP	c.5693G>A, p.Arg1898His	LLP	
14041	MEH	c.4469G>A, p.Cys1490Tyr	LP	c.5461-10T>C, splice site alteration	LP					
14042	MEH	c.161G>A, p.Cys54Tyr	LP	c.2588G>C, p.Gly863Ala	LP					

14044	MEH	c.3323G>A, p.Arg1108His	LP	c.5461-10T>C, splice site alteration	LP	
14046	MEH	c.1906C>T, p.Gln636Ter	LP	c.2588G>C, p.Gly863Ala	LP	
14047	MEH	c.1922G>C, p.Cys641Ser	LP	c.6079C>T, p.Leu2027Phe	LP	
14049	MEH	c.4552A>C, p.Ser1518Arg	LP	c.5882G>A, p.Gly1961Glu	LP	
14050	MEH	c.3323G>A, p.Arg1108His	LP	c.6079C>T, p.Leu2027Phe	LP	
14051	MEH	c.1760G>A, p.Arg587Lys(splice site alteration)	LP	c.2564G>A, p.Trp855Ter	LP	
14053	MEH	c.5018+2T>C, splice site alteration	LP	c.5882G>A, p.Gly1961Glu	LP	
14054	MEH	c.1906C>T, p.Gln636Ter	LP	c.6817-2A>C, splice site alteration	LP	
14055	MEH	c.5882G>A, p.Gly1961Glu	LP	c.2522A>C, p.Gln841Pro	LLP	
14056	MEH	c.2966T>C, p.Val989Ala	LP	c.6286G>A, p.Glu2096Lys	LP	
14058	MEH	c.5882G>A, p.Gly1961Glu	LP	c.5882G>A, p.Gly1961Glu	LP	✓
14059	MEH	c.3364G>A, p.Glu1122Lys	LP	c.3758C>T, p.Thr1253Met	LLP	c.5882G>A, p.Gly1961Glu LP
14060	MEH	c.21_22insA, p.Gln8ThrfsTer46	LP	c.5461-10T>C, splice site alteration	LP	

14062	MEH	c.4739T>C, Leu1580Ser	LLP	c.6729+5_19delGTTGG CCCTGGGGCA, splice site alteration	LP		
14063	MEH	c.303-2A>G, <i>Spliceacceptor</i>	LP	c.5882G>A, p.Gly1961Glu	LP		
14065	MEH	c.5882G>A, p.Gly1961Glu	LP	c.1906C>T, p.Gln636Ter	LP		✓
14067	MEH	c.4537_4538insC, p.Pro1513GlnfsTer42	LP	c.5578C>T, p.Arg1860Trp	LP		
14069	MEH	c.4469G>A, p.Cys1490Tyr	LP	c.6089G>A, p.Arg2030Gln	LP		
14070	MEH	c.1317G>A, p.Trp439Ter	LP	c.4139C>T, p.Pro1380Leu	LP		
14072	MEH	c.5882G>A, p.Gly1961Glu	LP	c.5882G>A, p.Gly1961Glu	LP		✓
14073	MEH	c.2588G>C, p.Gly863Ala	LP	c.4139C>T, p.Pro1380Leu	LP	c.6088C>T, p.Arg2030Ter	LP
14074	MEH	c.4139C>T, p.Pro1380Leu	LP	c.6079C>T, p.Leu2027Phe	LP		
14075	MEH	c.2453G>A, p.Gly818Glu	LP	c.6817-2A>C, splice site alteration	LP		
14076	MEH	c.4253+4C>T, splice site alteration	LP	c.4253+4C>T, splice site alteration	LP		✓
14077	MEH	c.3292C>T, p.Arg1098Cys	LP	c.5461-10T>C, splice site alteration	LP		
14078	MEH	c.634C>T, p.Arg212Cys	LP	c.5461-10T>C, splice site alteration	LP		



14079	MEH	c.3113C>T, p.Ala1038Val	LLP	c.5882G>A, p.Gly1961Glu	LP
14080	MEH	c.5898+1G>A, splice site alteration	LP	c.4253+43G>A, deep intron	LLP
14082	MEH	c.634C>T, p.Arg212Cys	LP	c.5882G>A, p.Gly1961Glu	LP
14083	MEH	c.5196+1G>A, splice site alteration	LP		
14085	MEH	c.3322C>T, p.Arg1108Cys	LP	c.4577C>T, p.Thr1526Met	LP
14087	MEH	c.4469G>A, p.Cys1490Tyr	LP		
14092	MEH	c.3224C>A, p.Ala1075Asp	LP	c.5882G>A, p.Gly1961Glu	LP
14093	MEH	c.2588G>C, p.Gly863Ala	LP	c.5177C>A, p.Thr1726Asn	LP
14094	MEH	c.655A>T, p.Arg219Ter	LP	c.2588G>C, p.Gly863Ala	LP
14095	MEH	c.2160+2T>G, splice site alteration	LP	c.5196+1137G>A, deep intron	LLP
14096	MEH	c.6729+5_19delGTTGG CCCTGGGGCA, splice site alteration	LP	c.6729+5_19delGTTGG CCCTGGGGCA, splice site alteration	LP
14098	MEH	c.834delT, p.Ser278SerfsTer22	LP	c.768+353T>C, deep intron	LLP
14099	MEH	c.5461-10T>C, splice site alteration	LP		

✓

14100	MEH	c.5882G>A, p.Gly1961Glu	LP	c.1757A>G, p.Asp586Gly	LP	c.4667+45T>C, deep intron	LLP
14101	MEH	c.5714+5G>A, splice site alteration	LP	c.6209C>G, p.Thr2070Arg	LP		
14102	MEH	c.3322C>T, p.Arg1108Cys	LP	c.4469G>A, p.Cys1490Tyr	LP	c.6320G>A, p.Arg2107His	LP
14103	MEH	c.4139C>T, p.Pro1380Leu	LP	c.5882G>A, p.Gly1961Glu	LP		
14105	MEH	c.6729+5_19delGTTGG CCCTGGGGCA, splice site alteration	LP				
14106	MEH	c.3481C>T, p.Arg1161Cys	LP				
14107	MEH	c.2588G>C, p.Gly863Ala	LP	c.4469G>A, p.Cys1490Tyr	LP	c.4685T>C, p.Ile1562Thr	LP
14108	MEH	c.5882G>A, p.Gly1961Glu	LP	c.4793C>A, p.Ala1598Asp	LP		✓
14109	MEH	c.2912C>A, p.Thr971Asn	LP				
14110	MEH	c.859-9T>C, splice site alteration	LP				
14111	MEH	c.6712C>T, p.Gln2238Ter	LP	c.5882G>A, p.Gly1961Glu	LP		
14112	MEH	c.6286G>A, p.Glu2096Lys	LP	c.2894A>G, p.Asn965Ser	LP		
14114	MEH	c.1267C>T, p.His423Tyr	LLP	c.3329G>T, p.Gly1110Val	LP		

14115	MEH	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.3322C>T, p.Arg1108Cys	LP	
14116	MEH	c.2588G>C, p.Gly863Ala	LP	c.3210_3211insGT, p.Ser1071CysfsTer14	LP			
14117	MEH	c.1317G>A, p.Trp439Ter	LP	c.5882G>A, p.Gly1961Glu	LP	c.5908C>T, p.Leu1970Phe	LP	
14118	MEH	c.3364G>A, p.Glu1122Lys	LP	c.5882G>A, p.Gly1961Glu	LP			
14119	MEH	c.2588G>C, p.Gly863Ala	LP	c.6079C>T, p.Leu2027Phe	LP			
13003	INSERM	c.3386G>T, p.Arg1129Leu	LP	c.4139C>T, p.Pro1380Leu	LP			
13007	INSERM	c.2041C>T, p.Arg681Ter	LP	c.2041C>T, p.Arg681Ter	LP			✓
13008	INSERM	c.4577C>T, p.Thr1526Met	LP	c.6319C>T, p.Arg2107Cys	LP			✓
13009	INSERM	c.3386G>T, p.Arg1129Leu	LP	<i>c.5835+5G&gt;A, splice site alteration</i>	LP			
13015	INSERM	c.3899G>A, p.Arg1300Gln	LLP	c.6320G>A, p.Arg2107His	LP			
13019	INSERM	<i>c.2868C&gt;A, p.Asn956Lys</i>	LP	<i>c.5936C&gt;T, p.Thr1979Ile</i>	LP			
13025	INSERM	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.4139C>T, p.Pro1380Leu	LP	✓
13026	INSERM	c.5961_5964delGGAC, p.Gly1987GlyfsTer4	LP	c.5961_5964delGGAC, p.Gly1987GlyfsTer4	LP			✓

13029	INSERM	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP		
13030	INSERM	c.6077T>C, p.Leu2026Pro	LP	c.6077T>C, p.Leu2026Pro	LP		✓
13032	INSERM	c.6079C>T, p.Leu2027Phe	LP	c.6693delC, p.Ile2231MetfsTer16	LP		
13033	INSERM	c.3322C>T, p.Arg1108Cys	LP	c.5714+5G>A, splice site alteration	LP		
13034	INSERM	c.4537_4538insC, p.Pro1513GlnfsTer42	LP	c.5882G>A, p.Gly1961Glu	LP		
13036	INSERM	c.3113C>T, p.Ala1038Val	LLP	c.4139C>T, p.Pro1380Leu	LP	c.5177C>A, p.Thr1726Asn	LP ✓
13037	INSERM	c.5882G>A, p.Gly1961Glu	LP	c.6386+2C>G, splice site alteration	LP		
13039	INSERM	c.4469G>A, p.Cys1490Tyr	LP	c.6079C>T, p.Leu2027Phe	LP		
13040	INSERM	c.4352+1G>T, splice site alteration	LP	c.5882G>A, p.Gly1961Glu	LP		
13041	INSERM	c.4139C>T, p.Pro1380Leu	LP	c.5882G>A, p.Gly1961Glu	LP		✓
13043	INSERM	c.5714+5G>A, splice site alteration	LP	c.6079C>T, p.Leu2027Phe	LP		
13044	INSERM	c.1819G>A, p.Gly607Arg	LP	c.5461-10T>C, splice site alteration	LP		
13046	INSERM	c.634C>T, p.Arg212Cys	LP	c.5882G>A, p.Gly1961Glu	LP		

13047	INSERM	c.4720G>T, p.Glu1574Ter	LP	c.5882G>A, p.Gly1961Glu	LP		
13049	INSERM	c.2564G>A, p.Trp855Ter	LP	c.3386G>T, p.Arg1129Leu	LP		
13050	INSERM	c.4919G>A, p.Arg1640Gln	LP	c.5603A>T, p.Asn1868Ile	LP		
13052	INSERM	c.2560_2561insG, p.Ala854GlyfsTer5	LP	c.5882G>A, p.Gly1961Glu	LP		
13054	INSERM	c.1556G>A, p.Cys519Tyr	LP	c.3292C>T, p.Arg1098Cys	LP		
13056	INSERM	c.423T>A, p.Thr141Thr	LLP	c.6320G>A, p.Arg2107His	LP		
13057	INSERM	c.634C>T, p.Arg212Cys	LP	c.6148G>C, p.Val2050Leu	LP		
13060	INSERM	c.5018+2T>C, splice site alteration	LP	c.6320G>A, p.Arg2107His	LP		
13061	INSERM	c.2948C>T, p.Thr983Ile	LP	c.5882G>A, p.Gly1961Glu	LP		
13063	INSERM	c.5882G>A, p.Gly1961Glu	LP	c.5929G>A, p.Gly1977Ser	LP		
13064	INSERM	c.5585-10T>C, splice site alteration	LP	c.5603A>T, p.Asn1868Ile	LP		
13068	INSERM	c.4537_4538insC, p.Pro1513GlnfsTer42	LP	c.5603A>T, p.Asn1868Ile	LP	c.6282+7G>A, possible splice site alteration	LLP
13069	INSERM	c.3064G>A, p.Glu1022Lys	LP	c.6320G>A, p.Arg2107His	LP		

13070	INSERM	c.2041C>T, p.Arg681Ter	LP	c.2041C>T, p.Arg681Ter	LP		
13072	INSERM	<i>c.1019A&lt;G, p.Tyr340Cys</i>	LP				
13074	INSERM	c.4537_4538insC, p.Pro1513GlnfsTer42	LP	c.5882G>A, p.Gly1961Glu	LP		✓
13075	INSERM	c.872C>T, p.Pro291Leu	LP	c.4222T>C, p.Trp1408Arg	LP		
13076	INSERM	<i>c.1201A&gt;C, p.Thr401Pro</i>	LP	c.2588G>C, p.Gly863Ala	LP		
13079	INSERM	c.885delC, p.Asp295AspfsTer5	LP	c.6089G>A, p.Arg2030Gln	LP		
13080	INSERM	c.5018+2T>C, splice site alteration	LP	c.3292C>T, p.Arg1098Cys	LP		
13081	INSERM	c.4222T>C, p.Trp1408Arg	LP	c.4918C>T, p.Arg1640Trp	LP		
13083	INSERM	c.3210_3211insGT, p.Ser1071CysfsTer14	LP	c.5603A>T, p.Asn1868Ile	LP	c.5603A>T, p.Asn1868Ile	LP
13084	INSERM	c.4139C>T, p.Pro1380Leu	LP	c.6079C>T, p.Leu2027Phe	LP		✓
13087	INSERM	c.4462T>C, p.Cys1488Arg	LP	c.5882G>A, p.Gly1961Glu	LP		
13088	INSERM	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.4577C>T, p.Thr1526Met	LP
13090	INSERM	c.3386G>T, p.Arg1129Leu	LP	c.4139C>T, p.Pro1380Leu	LP		

13091	INSERM	c.5882G>A, p.Gly1961Glu	LP	<i>c.6181_6184delACTG, p.Thr2061SerfsTer53</i>	LP		
13092	INSERM	c.5461-10T>C, splice site alteration	LP	c.5714+5G>A, splice site alteration	LP		
13093	INSERM	c.2588G>C, p.Gly863Ala	LP	c.6445C>T, p.Arg2149Ter	LP		
13094	INSERM	c.5461-10T>C, splice site alteration	LP	c.1819G>A, p.Gly607Arg	LP		
13095	INSERM	<i>c.3311T&gt;C, p.Leu1104Pro</i>	LP	c.6730-3T>C, splice site alteration	LLP	c.6764G>T, p.Ser225Ile	LLP
13096	INSERM	c.868C>T, p.Arg290Trp	LP	c.5882G>A, p.Gly1961Glu	LP		✓
13097	INSERM	c.4139C>T, p.Pro1380Leu	LP	c.6730-3T>C, splice site alteration	LLP	c.6764G>T, p.Ser225Ile	LLP
13098	INSERM	c.286A>G, p.Asn96Asp	LP	c.2813T>C, p.Phe938Ser	LP		
13099	INSERM	c.3386G>T, p.Arg1129Leu	LP	c.5929G>A, p.Gly1977Ser	LP		
13100	INSERM	c.2894A>G, p.Asn965Ser	LP	c.5882G>A, p.Gly1961Glu	LP	<i>c.6250G&gt;A, p.Ala2084Thr</i>	LP
13101	INSERM	c.3322C>T, p.Arg1108Cys	LP	c.5196+1G>A, splice site alteration	LP		
13102	INSERM	c.5693G>A, p.Arg1898His	LLP	c.5882G>A, p.Gly1961Glu	LP		
13103	INSERM	c.2588G>C, p.Gly863Ala	LP	c.3322C>T, p.Arg1108Cys	LP		

13104	INSERM	c.4926C>G, p.Ser1642Arg	LP	c.5041_5055delGTGGT TGCCATCTGC, p.Val1681_Cys1685del	LP		
15002	EKU	c.634C>T, p.Arg212Cys	LP	c.3113C>T, p.Ala1038Val	LLP		
15004	EKU	c.3113C>T, p.Ala1038Val	LLP	<i>c.5936C&gt;T, p.Thr1979Ile</i>	LP		
15006	EKU	c.5461-10T>C, splice site alteration	LP	c.5714+5G>A, splice site alteration	LP		
15008	EKU	c.1648G>A, p.Gly550Arg	LP	c.2588G>C, p.Gly863Ala	LP		
15010	EKU	c.1574T>C, p.Phe525Ser	LP	c.2588G>C, p.Gly863Ala	LP		
15012	EKU	<i>c.161-1G&gt;A, splice site alteration</i>	LP	c.5882G>A, p.Gly1961Glu	LP		✓
15016	EKU	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5882G>A, p.Gly1961Glu	LP
15017	EKU	<i>c.183G&gt;C, p.Met61Ile</i>	LP	c.4653G>A, p.Trp1551Ter	LP		
15018	EKU	c.1622T>C, p.Leu541Pro	LP	c.2588G>C, p.Gly863Ala	LP	c.3113C>T, p.Ala1038Val	LLP
15020	EKU	<i>c.4663_4664delCA, p.Gln1555GlufsTer41</i>	LP	c.5882G>A, p.Gly1961Glu	LP		
15022	EKU	c.571-1G>T, splice site alteration	LP	c.5882G>A, p.Gly1961Glu	LP		✓
15026	EKU	c.3703A>G, p.Asn1235Asp	LLP	<i>c.5936C&gt;T, p.Thr1979Ile</i>	LP		



15028	EKU	c.5137C>A, p.Gln1713Lys	LP	c.5312+1G>A, splice site alteration	LP				
15030	EKU	c.3210_3211insGT, p.Ser1071CysfsTer14	LP	c.6089G>A, p.Arg2030Gln	LP				
15031	EKU	c.1648G>A, p.Gly550Arg	LP	c.5882G>A, p.Gly1961Glu	LP				
15032	EKU	c.5288T>C, p.Leu1763Pro	LP	c.4739T>C, Leu1580Ser	LLP				
15034	EKU	c.2588G>C, p.Gly863Ala	LP	c.5520C>A, p.Cys1840Ter	LP				
15036	EKU	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5584+6T>C, splice site alteration	LP	✓	
15038	EKU	c.2966T>C, p.Val989Ala	LP	c.52C>T, p.Arg18Trp	LP				
15040	EKU	c.3113C>T, p.Ala1038Val	LLP	c.4254-41C>T, deep intron	LLP	c.6005+1G>T, splice site alteration	LP		
15042	EKU	c.1034A>G, p.Tyr345Cys	LP	c.6721C>G, p.Leu2241Val	LP				
15046	EKU	c.5882G>A, p.Gly1961Glu	LP	c.6229C>T, p.Arg2077Trp	LP				
15048	EKU	c.5461-10T>C, splice site alteration	LP					✓	
15052	EKU	c.1654G>A, p.Val552Ile	LLP	c.4254-1G>C, splice site alteration	LP				
15056	EKU	c.455G>A, p.Arg152Gln	LLP	c.3322C>T, p.Arg1108Cys	LP	c.5882G>A, p.Gly1961Glu	LP	c.6320G>A, p.Arg2107His	LP

15060	EKU	c.5461-10T>C, splice site alteration	LP	c.5882G>A, p.Gly1961Glu	LP		
15062	EKU	c.3322C>T, p.Arg1108Cys	LP	c.5882G>A, p.Gly1961Glu	LP		✓
15063	EKU	c.5714+5G>A, splice site alteration	LP	c.3386G>T, p.Arg1129Leu	LP		
15067	EKU	c.5461-10T>C, splice site alteration	LP	c.5882G>A, p.Gly1961Glu	LP		✓
15070	EKU	c.1964T>G, p.Phe655Cys	LP	c.5882G>A, p.Gly1961Glu	LP		✓
15071	EKU	c.5461-10T>C, splice site alteration	LP	c.5882G>A, p.Gly1961Glu	LP		
15074	EKU	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LLP	c.5882G>A, p.Gly1961Glu	LP
15076	EKU	c.2588G>C, p.Gly863Ala	LP	c.3113C>T, p.Ala1038Val	LLP	<i>c.5715-1G&gt;T, splice site alteration</i>	LP
15078	EKU	c.6112C>T, p.Arg2038Trp	LP	c.6112C>T, p.Arg2038Trp	LP		
15080	EKU	c.194G>A, p.Gly65Glu	LP	c.4253+43G>A, deep intron	LLP		
15084	EKU	c.1622T>C, p.Leu541Pro	LP	c.3113C>T, p.Ala1038Val	LP	c.4253+43G>A, deep intron	LLP
15085	EKU	<i>c.183G&gt;C, p.Met61Ile</i>	LP	<i>c.5777A&gt;C, p.Gln1926Pro</i>	LP		✓
15088	EKU	c.5714+5G>A, splice site alteration	LP	c.5714+5G>A, splice site alteration	LP		

15090	EKU	c.571-2A>T, splice site alteration	LP			
15092	EKU	<i>c.4313C&gt;G, p.Pro1438Arg</i>	LP	c.4328G>A, p.Arg1443His		LP
15094	EKU	c.45G>A, p.Trp15Ter	LP	c.5882G>A, p.Gly1961Glu		LP
15095	EKU	c.1819G>A, p.Gly607Arg	LP			
15096	EKU	c.2588G>C, p.Gly863Ala	LP	c.5461-10T>C, splice site alteration	LP	c.656G>C, p.Arg219Thr LP
15097	EKU	c.5882G>A, p.Gly1961Glu	LP			
15098	EKU	c.2041C>T, p.Arg681Ter	LP	<i>c.3482G&gt;A, p.Arg1161His</i>		LP
15102	EKU	c.5461-10T>C, splice site alteration	LP	c.4253+43G>A, deep intron		LLP
15106	EKU	c.5882G>A, p.Gly1961Glu	LP	<i>c.760_768+25delTTCC GTGTGGTAAGGGAG GGGTTTGGCTGCTCG C, p.Phe2541GlnfsTer8</i>		LP
15108	EKU	c.2023G>A, p.Val675Ile	LP	c.3305A>T, p.Asp1102Val	LP	c.6647C>T, p.Ala2216Val LP
15110	EKU	c.6721C>G, p.Leu2241Val	LP	<i>c.3007_3008insC, p.Gln1003ArgfsTer20</i>		LP

JHU = The Wilmer Eye Institute, Johns Hopkins University, Baltimore, MD; GBMC = Greater Baltimore Medical Center, Baltimore, MD; Penn = Sheie Eye Institute, University of Philadelphia, Philadelphia, PA; RFSW = Retina Foundation of the Southwest, Dallas, TX; MEC = Moran Eye Center, Salt Lake City, UT; CC = Cole Eye Institute, Cleveland Clinic, Cleveland, OH; MEH = Moorfields Eye Hospital, London, United Kingdom; INSERM = Université de Paris 06, Institut national de la santé et de la recherche médicale, Paris France; EKU = Eberhard-Karls University Hospital, Tuebingen, Germany. LP = Likely pathogenic; LLP = less likely pathogenic.

Novel variants are shown in *Italic*.

Transcript: ENST00000370225

Co-segregation analysis was unavailable in 16 patients with three pathogenic variants and the chromosomal positional relationship between these variants are unknown (11031, 11034, 17012, 17045, 18014, 14003, 14020, 14021, 14036, 14037, 14073, 14107, 13100, 15056, 15096, 15108).