

Reported Families	Reported Individuals	Gender	Nucleotide variation	AA variation (new nomenclature)	AA variation (old nomenclature)	Country	References
1	1	1M	G>T 3805	Trp 246 Leu	Trp230Leu	Argentina	Woods <i>et al.</i> [17]
1	7	1F, 6M	G>T 746	Gly 249 Val	Gly233Val	USA	Miller <i>et al.</i> [12]
1	2	2M	G>T 746	Gly 249 Val	Gly233Val	Japan	Matsubara <i>et al.</i> [16]
5	9	9F	G>T 746	Gly 249 Val	Gly233Val	UK	Enayat <i>et al.</i> [19]
1	8	7F, 1M	G>T 746	Gly 249 Val	Gly233Val	UK	Whalley and Perry [31]
3	3	2F, 1M	G>T 746	Gly 249 Val	Gly233Val	UK	Hamilton <i>et al.</i> [27]
2	2	2F	G>T 746	Gly 249 Val	Gly233Val	Canada	Hamilton <i>et al.</i> [27]
1	1	1F	G>T 746	Gly 249 Val	Gly233Val	Ireland	Submitted by J.O'Donnell, 2011
1	1	1F	G>T 746	Gly 249 Val	Gly233Val	Canada	Submitted by N.Rydz, 2014
1	2	2F	G>A 746	Gly 249 Ser	Gly233Ser	France	Nurden <i>et al.</i> [34]
2	3	3F	G>A 746	Gly 249 Ser	Gly233Ser	Australia New Zealand	Favaloro <i>et al.</i> [35]
1	2	1F, 1M	G>T 793	Asp 251 Tyr	Asp 235 Tyr	Iran	Enayat <i>et al.</i> [32]
1	3	2F, 1M	A>G 763	Met 255 Val	Met 239 Val	Puerto Rico	Russell and Roth [15] Weiss <i>et al.</i> [8]
2	4	2F, 2M	A>G 763	Met 255 Val	Met 239 Val	Japan	Takahashi <i>et al.</i> [33]
1	1	1F	A>G 763	Met 255 Val	Met 239 Val	Italy	Giannini <i>et al.</i> [23]
2	3	2M, 1F	A>G 763	Met 255 Val	Met 239 Val	USA	Submitted by S.Acharya, 2014
1	3	3F	1306 del 27	436-444 del 9	420_428 del	UK	Othman <i>et al.</i> [18]