

Reported families	Affected individuals	Gender	Country	Provisional diagnosis	RIPA	VWF HMWM	Nucleotide change	Amino acid change	Reference
1	7	1F, 6M	USA	PT-VWD	increased	absent	G>T 746	Gly 249 Val	Miller et al, 1991
1	3	2F, 1M	Puerto Rico	PT-VWD	increased	absent	A>G 763	Met 255 Val	Russell and Roth 1993 Weiss et al, 1983
2	4	2F, 2M	Japan	PT-VWD	increased	absent	A>G 763	Met 249 Val	Takahashi et al, 1995
1	2	2M	Japan	?PT-VWD	increased		G>T 746	Gly 249 Ser	Matsubara et al, 2003
1	3	3F	UK	2B	increased	absent	1306 del 27	436-444 del 9	Othman et al, 2005
5	9	9F	UK	2B	increased	absent	G>T 746	Gly 249 Val	Enayat et al, 2006
1	8	7F, 1M	UK	PT-VWD	increased	absent	G>T 746	Gly 249 Val	Whalley and Perry, 2006
1	2	2F	France	2B/I TP	increased	absent	G>A 746	Gly 249 Ser	Nurden et al, 2007
2	3	3F	Australia/ New Zealand	?PT-VWD	Increased/pos. simplified RIPA	absent	G>A 746	Gly 249 Ser	Favaloro et al, 2007
1	1	1F	Italy	2B	Increased/pos. flow cytometry	NA	A>G 763	Met 255 Val	Giannini et al, 2010
3	3	2F, 1M	UK	2B	increased	absent	G>T 746	Gly 249 Val	Hamilton et al, 2011
2	2	2F	Canada	1 2B, 1 PT-VWD	increased	absent	G>T 746	Gly 249 Val	Hamilton et al, 2011
1	2	1F, 1M	Iran	2B	increased	absent	G>T 793	Asp 251 Tyr	Enayat et al, 2012
1	1	1F	Ireland UK	2B	increased	absent	G>T 746	Gly 249 Val	O'Donnell, NCC 2011, Unpublished
1	1	1F	Canada	2B	increased	Normal	G>T 746	Gly 249 Val	Rydz 2014, unpublished
1	1	1M	Argentina		increased	Intermediate /large M: reduced , small M: increased	G>T 3805	Trp 246 Leu	Woods et al, 2014
1	1	1M	USA	2B	increased	Qualitative defect	A>G 763	Met 255 Val	Acharya 2014, unpublished