

PT-VWD Registry

Reported families	investigated individuals	Affected individuals	Sex	Country	Provisional diagnosis	RIPA	VWF HMWM	GP1BA mutation	Reference
1	14	7	1 F, 6 M	USA	PT-VWD	increased	absent	G 233 V	Miller et al., 1991
1	4	3	2 F, 1 M	Puerto Rican	PT-VWD	Increased	absent	M 239 V	Russell and Roth 1993 Weiss et al., 1983
2	2	4	2 F, 2 M	Japan	PT-VWD	increased	absent	M 239 V	Takahashi et al., 1995
1	2	2	2 M	Japan	?PT-VWD	increased		G 233 S	Matsubara et al., 2003
1	5	3	3 F	UK	2B	increased	absent	27 bp deletion	Othman et al., 2005
5	15	9	9 F	UK	2B	increased	absent	G 233 V	Enayat's et al., 2006
1	8	8	7 F, 1 M	UK	PT-VWD	increased	absent	G 233 V	Whalley and Perry 2006
1	2	2	2 F	France	2B/ITP	increased	absent	G 233 S	Nurden's et al., 2007
2	3	3	3 F	Australia/ New Zealand	? PT-VWD	increased/ pos. simplified RIPA	absent	G 233 S	Favaloro et al., 2007
1	6	6	1 F	Italy	2B	Increased/pos. flow cytometry	NA	M 239 V	Giannini et al., 2010
3	3	3	2 F, 1 M	UK	2B	Increased	absent	G 233 V	Hamilton et al., 2011
2	2	2	2 F	Canada	1 2B, 1 PT-VWD	Increased	absent	G 233 V	Hamilton et al., 2011
1	2	2	1 F, 1 M	Iran	2B	increased	absent	D 235 Y	Enayat, ICHCC 2011, unpublished
1	1	1	1F	Ireland –UK	2B	Increased	absent	G 233 V	O'Donnell, NCC 2011, unpublished