

PT-VWD Registry

Reported families	investigated individuals	affected individuals	sex	ethnic origin	provisional diagnosis	RIPA	VWF HMWM	GP1BA mutation	Reference
1	14	7	1 F, 6 M	Caucas.	PT-VWD	increased	absent	Gly 233 Val	Miller et al., 1991
1	4	3	2 F, 1 M	Puerto Rican	PT-VWD	increased	absent	Met 239 Val	*Russell and Roth 1993
									*Family described originally by Weiss et al., 1983)
2	2	4	2 F, 2 M	Japan	PT-VWD	increased	absent	Met 239 Val	Takahashi et al., 1995
1	2	2	2 M	Japan	?PT-VWD	increased		Gly 233 Ser	Matsubara et al., 2003
1	5	3	3 F	UK	2B	increased	absent	27 bp deletion	Othman et al., 2005
5	15	9	9F	UK	2B	increased	absent	Gly 233 Val	Enayat's et al., 2006
+3	+4	+4	3F& 1M	UK	3 for 2B & 1 for type 2VWD	increased	absent	Gly 233 Val	Unpublished
1	8	8	?	UK	2B	increased	?	Gly 233 Val	Whalley and Perry 2006
1	2	2	2 F	France	2B/ ITP	increased	absent	Gly 233 Ser	Nurden's et al ., 2007
2	3	3	3 F	Australia/ New Zealand	? PT-VWD	increased/ simplified RIPA confirmed	absent	Gly 233 Ser	Favaloro et al., 2007