

Supplementary table 3 - Variants submitted by multiple centers and with different pathogenicity (PV, pathogenic variant; LPV, likely pathogenic variant; VUS, variant of uncertain significance; LBV, likely benign variant).							
One line represents one variant in one patient. The same submission centre might have multiple interpretations for the same variant as evidences accumulate over time and variant interpretation in one individual might not be the same as interpretation on a second individual later in time.							
Center	Gene	Variant	Disorder name (compulsory)	Clinical features (optional)	Submitted pathogenicity	ClinVar Interpretation (access date 08/04/2021)	Gene-specific database
Grupo Español de Alteraciones Plaquetarias	ACTN1	R46Q	Macrothrombocytopenia	mean platelet volume >13 fl, platelet count <100 from routine blood test. Platelet count <50,000/mm ³ . Bleeding Assessment Tool (BAT)	LPV	3 submissions: LPV, PV, PV (accession VCV000051196.1)	-
Grupo Español de Alteraciones Plaquetarias			Macrothrombocytopenia		LPV		
NIHR Biosource - Cambridge University, UK			Macrothrombocytopenia		PV		
Center for Molecular and Vascular Biology, L			Macrothrombocytopenia		PV		
Center for Molecular and Vascular Biology, L			Macrothrombocytopenia		PV		
NIHR Biosource - Cambridge University, UK	ANKRD26	c.-126T>C	Autosomal dominant thrombocytopenia 2		LPV	4 submissions: PV, LPV, PV, PV (accession VCV000615436.1)	-
NIHR Biosource - Cambridge University, UK			Autosomal dominant thrombocytopenia 2		PV		
Institute of Experimental Biomedicine, Univ			Thrombocytopenia		PV		
Grupo Español de Alteraciones Plaquetarias	ANKRD26	c.-128G>A	Thrombocytopenia	Misdiagnosis immune thrombocytopenia	LPV	1 submission: PV (accession VCV000801119.1)	-
NIHR Biosource - Cambridge University, UK			Autosomal dominant thrombocytopenia 2		PV		
Northern Blood Research Centre, Sydney, AU			Autosomal dominant thrombocytopenia 2	Thrombocytopenia, menorrhagia, post-surgical bleeding. Three generations affected. Grandmother died of leukemia	PV		
Northern Blood Research Centre, Sydney, AU			Autosomal dominant thrombocytopenia 2	Thrombocytopenia, menorrhagia, post-surgical bleeding. Three generations affected. Grandmother died of leukemia	PV		
NIHR Biosource - Cambridge University, UK	F7	G156S	Factor VII deficiency		LPV	Absent	https://r17-00.eanao.org/reports/5/cases/3/homozygous/2/heterozygous . Pathogenicity: to be confirmed.
Katharine Dormandy Haemophilia and Thro			Factor VII deficiency		PV		
NIHR Biosource - Cambridge University, UK	GP1BB	L16P	Mild macrothrombocytopenia		PV	2 submissions: LPV, LPV (accession VCV000615638.1)	-
Center for Molecular and Vascular Biology, L			Mild macrothrombocytopenia	Autosomal dominant thrombocytopenia / aggregation defect with ADP / bleeding requiring transfusion after delivery	PV		
Center for Molecular and Vascular Biology, L			Mild macrothrombocytopenia		LPV		
Center for Molecular and Vascular Biology, L			Bernard-Soulier syndrome	Macrothrombocytopenia	LPV		
Center for Molecular and Vascular Biology, L			Bernard-Soulier syndrome	Macrothrombocytopenia	LPV		
NIHR Biosource - Cambridge University, UK	ITGA2B	R1026W	Platelet-type bleeding disorder 16		LPV	6 submissions: PV, PV, PV, PV, PV (2 associated with Glanzmann thrombastenia, 4 associated with platelet disorder type 16 that includes thrombocytopenia). ClinGen has curated this variant for Glanzmann thrombastenia as VUS (but not yet curated this variant for platelet disorder type 16) (accession	Not present in the Glanzmann thrombastenia database https://glanzmann.mcw.edu
NIHR Biosource - Cambridge University, UK			Platelet-type bleeding disorder 16		PV		
NIHR Biosource - Cambridge University, UK			Platelet-type bleeding disorder 16		PV		
Northern Blood Research Centre, Sydney, AU			Glanzmann 's thrombastenia AD	Bleeding history without family history. Mild thrombocytopenia previously diagnosed as immune thrombocytopenia. Bleeding Assessment Tool (BAT)	PV		
Grupo Español de Alteraciones Plaquetarias	MYH9	E1841K	May-Hegglin and other MYH9 disorders	Macrothrombocytopenia, Döhle bodies	LPV	4 submissions: PV, PV, PV, PV (accession RCV000015119.28)	-
Northern Blood Research Centre, Sydney, AU			May-Hegglin and other MYH9 disorders	Severe macrothrombocytopenia, Döhle body-like inclusions	PV		
Institute of Experimental Biomedicine, Univ	MYH9	S96L	Thrombocytopenia		VUS	5 submissions: PV, PV, PV, PV, LPV (accession RCV000015138.25)	-
Grupo Español de Alteraciones Plaquetarias			May-Hegglin and other MYH9 disorders	Macrothrombocytopenia and Döhle bodies, deafness and mild renal disease	PV		
Northern Blood Research Centre, Sydney, AU			May-Hegglin and other MYH9 disorders	Macrothrombocytopenia	PV		
Institute of Experimental Biomedicine, Univ			Thrombocytopenia May-Hegglin and other MYH9 disorders		PV		
NIHR Biosource - Cambridge University, UK	MYH9	W828R	May-Hegglin and other MYH9 disorders		LPV	Absent	-
Northern Blood Research Centre, Sydney, AU			MYH9-related disorders	Familial macrothrombocytopenia with Döhle-like bodies by immunofluorescence	PV		
Center for Molecular and Vascular Biology, L	PROC	F362I	Protein C deficiency	Pulmonary embolism, low protein C	VUS	Absent	-
NIHR Biosource - Cambridge University, UK			Protein C deficiency		LPV		
Center for Molecular and Vascular Biology, L	PROC	P369L	Protein C deficiency	Deep vein thrombosis, pulmonary embolism, low prote	VUS	Absent	-
NIHR Biosource - Cambridge University, UK			Protein C deficiency		LPV		
NIHR Biosource - Cambridge University, UK	PROS1	S501P	Protein S deficiency	Deep vein thrombosis	VUS	6 submissions: PV, VUS, VUS, VUS, LBV (accession VCV000013316.6)	-
Center for Molecular and Vascular Biology, L			Protein S deficiency	Pulmonary embolism	VUS		
Center for Molecular and Vascular Biology, L			Protein S deficiency	Deep vein thrombosis	VUS		
Center for Molecular and Vascular Biology, L			Protein S deficiency		VUS		
Center for Molecular and Vascular Biology, L			Protein S deficiency	Deep vein thrombosis, Low protein S	LPV		
Center for Molecular and Vascular Biology, L			Protein S deficiency	familial thrombosis	LPV		

Center for Molecular and Vascular Biology, L			Protein S deficiency	Deep vein thrombosis, Low protein S	LPV		
Center for Molecular and Vascular Biology, L			Protein S deficiency	Deep vein thrombosis and arterial thrombosis	LPV		
Center for Molecular and Vascular Biology, L			Protein S deficiency	Low protein S	LPV		
NIHR Bioresource - Cambridge University, UK	TUBB1	G109E	Macrothrombocytopenia, beta-tubulin 1 related		VUS	3 submissions: LPV, VUS, VUS (accession RCV000851597.1)	-
Center for Molecular and Vascular Biology, L			Macrothrombocytopenia, beta-tubulin 1 related	Macrothrombocytopenia	VUS		
Northern Blood Research Centre, Sydney, Australia			Macrothrombocytopenia	Macrothrombocytopenia without bleeding; family history	VUS		
Katharine Dormandy Haemophilia and Thrombosis Centre, London, UK			Thrombocytopenia		VUS		
Grupo Español de Alteraciones Plaquetarias			Macrothrombocytopenia, autosomal dominant, TUBB1-related	Macrothrombocytopenia; No significant bleeding	LPV		
Center for Molecular and Vascular Biology, L	VWF	N2546Y	von Willebrand disease	Low VWF antigen and activity / mild thrombocytopenia	VUS	1 submission: no class (accession RCV000086894.1)	www.vwf.group.shef.ac.uk. Homozygous patient VWDtype3
NIHR Bioresource - Cambridge University, UK			von Willebrand disease		LPV		
NIHR Bioresource - Cambridge University, UK	VWF	R1308C	von Willebrand disease		LPV	6 submissions: PV, PV, PV, PV, LPV, no evidence (accession RCV000000313.3)	www.vwf.group.shef.ac.uk. Patient VWDtype2B
University of Perugia, Department of Medicine			von Willebrand disease		PV		
Center for Molecular and Vascular Biology, L	VWF	R1399C	von Willebrand disease	von Willebrand disease type 2	VUS	5 submissions: LPV, LPV, LPV, VUS, no evidence (accession VCV000100337.7)	www.vwf.group.shef.ac.uk. Homozygous patient and heterozygous patient
NIHR Bioresource - Cambridge University, UK			von Willebrand disease		LPV		
Center for Molecular and Vascular Biology, L	VWF	Y1584C	von Willebrand disease	Miscarriage / low VWF antigen and activity	VUS	9 submissions: no evidence, risk factor, VUS, VUS, VUS, LPV, LPV, LPV, PV (accession VCV000015349.2)	www.vwf.group.shef.ac.uk. VWDtype1 and reported as polymorphism
Center for Molecular and Vascular Biology, L			von Willebrand disease	von Willebrand disease type1	VUS		
Center for Molecular and Vascular Biology, L			von Willebrand disease	low VWF levels / Platelet aggregation defect / neonatal in	VUS		
Katharine Dormandy Haemophilia and Thrombosis Centre, London, UK			von Willebrand disease		VUS		
NIHR Bioresource - Cambridge University, UK			von Willebrand disease		LPV		
NIHR Bioresource - Cambridge University, UK			von Willebrand disease		PV		