

Bleeding Disorders	Gene Symbol
Ehlers-Danlos syndrome, classic type	COL1A1; COL5A1; COL5A2
Ehlers-Danlos syndrome, musculocontractural type	CHST14
Ehlers-Danlos syndrome, vascular type	COL3A1
Juvenile polyposis	SMAD4
Telangiectasia, hereditary hemorrhagic syndrome	SMAD4
Telangiectasia, hereditary hemorrhagic, type 1	ENG
Telangiectasia, hereditary hemorrhagic, type 2	ACVRL1
Coagulation Disorders	Gene Symbol
Alpha 2 antiplasmin deficiency	SERPINF2
Angioedema, hereditary, type III & Factor XII deficiency	F12
Combined factor V and VIII deficiency	LMAN1; MCFD2
Factor V deficiency	F5
Factor VII deficiency	F7
Factor X deficiency	F10
Factor XI deficiency	F11
Factor XIII deficiency	F13A1; F13B
Fibrinogen deficiency	FGA; FGB; FGG
Haemophilia A	F8
Haemophilia B	F9
Kininogen deficiency	KNG1
Multiple coagulation factor deficiency type 2	VKORC1
Multiple coagulation factor deficiency type 3	GGCX
Plasminogen activator Inhibitor 1 deficiency	SERPINE1
Prothrombin deficiency	F2
von Willebrand disease	VWF
Platelet Disorders	Gene Symbol
ADP receptor defect	P2RY12
ARC syndrome	VIPAS39; VPS33B
Autosomal dominant thrombocytopenia 2	ANKRD26
Autosomal dominant thrombocytopenia 4	CYCS
Autosomal dominant macrothrombocytopenia	GP1BA; GP1BB
Bernard-Soulier syndrome	GP1BA; GP1BB; GP9
Bleeding diathesis due to glycoprotein VI deficiency	GP6
Bleeding disorder, platelet-type, 20	SLFN14
Congenital amegakaryocytic thrombocytopenia (CAMT)	MPL
Deficiency of phospholipase A2, group IV A	PLA2G4A
Dense granule abnormality	NBEA
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Familial hemophagocytic lymphohistiocytosis type 5	STXBP2
Familial platelet disorder with predisposition to AML	RUNX1
Ghosal syndrome	TBXAS1
Glanzmann thrombasthenia	ITGA2B; ITGB3

Gray platelet syndrome	NBEAL2
Gray platelet-like syndrome	GFI1B
Hermansky-Pudlak syndrome	AP3B1; AP3D1; BLOC1S3; BLOC1S6; DTNBP1; HPS1; HPS3; HPS4; HPS5; HPS6
Leukocyte integrin adhesion deficiency, type III	FERMT3
Macrothrombocytopenia	ACTN1; FLNA; GP1BB
Macrothrombocytopenia and sensorineural hearing loss	DIAPH1
Macrothrombocytopenia, Beta-tubulin 1 related	TUBB1
May-Hegglin and other MYH9 disorders	MYH9
Myopathy associated with thrombocytopenia	GNE
Noonan Syndrome	PTPN11
Paris-Trousseau thrombocytopenia and Jacobsen syndrome	FLI1
Platelet type von Willebrand disease	GP1BA
Platelet-type bleeding disorder 18	RASGRP2
Quebec platelet disorder	PLAU
Radioulnar synostosis with amegakaryocytic thrombocytopenia 1	HOXA11
Radioulnar synostosis with amegakaryocytic thrombocytopenia 2	MECOM
Roifman Syndrome	RNU4ATAC
Scott syndrome	ANO6
Sitosterolemia & Macrothrombocytopenia	ABCG5; ABCG8
Storage Pool Disorder	GFI1B
Stormorken syndrome	STIM1
Thrombocytopenia & Immune Deficiency	ARPC1B
Thrombocytopenia 3	FYB1
Thrombocytopenia and erythrokeraderma	KDSR
Thrombocytopenia and susceptibility to cancer	ETV6
Thrombocytopenia and thrombocythemia 1	THPO
Thrombocytopenia-absent radius syndrome (TAR)	RBM8A
Thrombocytopenia, anemia and myelofibrosis	MPIG6B
Thromboxane A2 receptor defect	TBXA2R
Wiskott-Aldrich syndrome	WAS
X-linked thrombocytopenia with dyserythropoiesis	GATA1
Thrombotic Disorders	Gene Symbol
Antithrombin deficiency	SERPINC1
Familial thrombotic thrombocytopenic purpura	ADAMTS13
Heparin cofactor 2 deficiency	SERPIND1
Histidine-rich glycoprotein deficiency	HRG
Plasminogen deficiency	PLG
Protein C deficiency	PROC
Protein S deficiency	PROS1
Thrombomodulin deficiency	THBD
Tissue plasminogen activator deficiency	PLAT