

Coagulation Factor Disorders	Genes
Alpha 2 anti-plasmin deficiency	SERPINF2
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
Multiple coagulation factor deficiency type 3	GGCX
Multiple coagulation factor deficiency type 2	VKORC1
Plasminogen activator Inhibitor 1 deficiency	SERPINE1
Prothrombin deficiency	F2
von Willebrand disease type 2 and 3	VWF
Platelet Disorders	Genes
ADP receptor defect	P2RY12
Amegakaryocytic thrombocytopenia with radioulnar synostosis	HOXA11
ARC syndrome (Arthrogryposis, renal dysfunction, and cholestasis 1)	VIPAS39
ARC syndrome (Arthrogryposis, renal dysfunction, and cholestasis 2)	VPS33B
Autosomal dominant thrombocytopenia 2	ANKRD26
Autosomal dominant thrombocytopenia 4	CYCS
Bernard-Soulier syndrome	GP1BA; GP1BB; GP9
Bleeding diathesis due to glycoprotein VI deficiency	GP6
Chediak-Higashi syndrome (CHS)	LYST
Congenital amegakaryocytic thrombocytopenia (CAMT)	MPL
Cyclic thrombocytopenia and thrombocythemia 1	THPO
Deficiency of phospholipase A2, group IV A	PLA2G4A
Dense granule abnormalities	NBEA
Familial hemophagocytic lymphohistiocytosis type 5	STXBP2

Familial platelet disorder with predisposition to acute myelogenous leukemia	RUNX1
Ghosal syndrome	TBXAS1
Glanzmann thrombasthenia	ITGA2B; ITGB3
Gray platelet syndrome	NBEAL2
Gray platelet-like syndrome (GPS)	GFI1B
Hermansky-Pudlak syndrome	AP3B1; BLOC1S3; BLOC1S6; DTNBP1; HPS1; HPS3; HPS4; HPS5; HPS6
Leukocyte integrin adhesion deficiency, type III	FERMT3
Macrothrombocytopenia	ACTN1; FLNA
May-Hegglin and other MYH9 disorders	MYH9
Myopathy associated with thrombocytopenia	GNE
Paris-Trousseau thrombocytopenia and Jacobson syndrome	FLI1
Platelet-type von Willebrand disease	GP1BA
Platelet-type bleeding disorder 18	RASGRP2
Quebec platelet disorder	PLAU
Scott syndrome	ANO6
Stormorken syndrome	STIM1; ORAI1
Thrombocytopenia-absent radius syndrome (TAR)	RBM8A
Thrombocytopenia, red cell macrocytosis and predisposition to lymphoblastic leukaemia	ETV6
Thromboxane A2 receptor defect	TBXA2R
Wiskott-Aldrich syndrome	WAS
X-linked thrombocytopenia with dyserythropoiesis	GATA1
Thrombotic Disorders	Genes
Anti-thrombin deficiency	SERPINC1
Heparin co-factor 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