

TG1.0 Targeted disorders and related genes

Coagulation Factor Disorders	Genes
Combined 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
Fletcher factor deficiency	KLKB1
Haemophilia A	F8
Haemophilia B	F9
Multiple coagulation factor deficiency type 1	GGCX
Multiple coagulation factor deficiency type 2	VKORC1
Plasminogen activator inhibitor 1 deficiency	SERPINE1
Plasminogen deficiency	PLG
Prothrombin deficiency	F2
Kininogen deficiency	KNG1
von Willebrand disease type 2 or 3	VWF
Platelet Disorders	Genes
ADP receptor defect	P2RY12
Amegakaryocytic thrombocytopenia with radio-ulnar synostosis	HOXA11
Autosomal Dominant thrombocytopenia 4	ANKRD26; CYCS; TUBB1; FLNA
Bernard-Soulier syndrome	GP1BA; GP1BB; GP9
Bleeding diathesis due to glycoprotein VI deficiency	GP6
Chediak–Higashi syndrome	LYST
Congenital amegakaryocytic thrombocytopenia	MPL
Cyclic thrombocytopenia and thrombocythemia 1	THPO
Deficiency of phospholipase A2, group IVA	PLA2G4A
Dense granule abnormalities	NBEA
Familial platelet disorder with predisposition to AML	RUNX1
Ghosal syndrome	TBXAS1
Glanzmann thrombasthenia	ITGA2B; ITGB3
Grey platelet syndrome	NBEAL2
Hermansky–Pudlak syndrome	HPS1; AP3B1; HPS3; HPS4; HPS5; HPS6; DTNBP1; BLOC1S3

May-Hegglin and other MYH9-related disorders	MYH9
Paris-Trousseau thrombocytopenia and Jacobson syndrome	FLI1
Platelet-type von Willebrand disease	GP1BA
Quebec platelet disorder	PLAU
Thrombocytopenia absent radius (TAR) syndrome	RBM8A
Thromboxane A2 receptor defect	TBXA2R
Wiskott-Aldrich syndrome	WAS
X-linked thrombocytopenia with dyserythropoiesis	GATA1
Thrombotic Disorders	Genes
Antithrombin deficiency	SERPINC1
Heparin co-factor 2 deficiency	SERPIND1
Histidine-rich glycoprotein deficiency	HRG
Protein C deficiency	PROC
Protein S deficiency	PROS1
Thrombomodulin deficiency	THBD
Tissue Plasminogen Activator deficiency	PLAT