

# Bleeding Thrombotic and Platelet Disorder TIER1 genes (ISTH\_2024)

Category	Gene symbol	Associated disorder(s)	Inheritance	Transcript	Location
Bleeding/coagulation	F10	Factor X deficiency	AR; AD	NM_000504.3	13q34
Bleeding/coagulation	F11	Factor XI deficiency	AR; AD	NM_000128.3	4q35.2
Coagulation <i>Angioedema</i>	F12	Factor XII deficiency <i>Angioedema</i>	AR (coagulation) <i>AD (angioedema)</i>	NM_000505.3	5q35.3
Bleeding/coagulation	F13A1	Factor XIII deficiency	AR	NM_000129.3	6p25.1
Bleeding/coagulation	F13B	Factor XIII deficiency	AR	NM_001994.2	1q31.3
Bleeding/coagulation <i>Thrombosis</i>	F2	Prothrombin deficiency <i>Thrombophilia due to thrombin defect</i>	AR (bleeding/coagulation) <i>AD (thrombosis)</i>	NM_000506.4	11p11.2
Bleeding/coagulation <i>Thrombosis</i>	F5	Factor V deficiency <i>Thrombophilia due to activated protein C resistance</i>	AR (bleeding/coagulation) <i>AD (thrombosis)</i>	NM_000130.4	1q24.2
Bleeding/coagulation	F7	Factor VII deficiency	AR; AD	NM_000131.4	13q34
Bleeding/coagulation	F8	Haemophilia A	XLR	NM_000132.3	Xq28
Bleeding/coagulation	F9	Haemophilia B	XLR	NM_000133.3	Xq27.1
Bleeding	FGA	Fibrinogen deficiency	AR (afibrinogenemia); AD (hypo/dysfibrinogenemia)	NM_000508.3	4q31.3
Bleeding	FGB	Fibrinogen deficiency	AR (afibrinogenemia); AD (hypo/dysfibrinogenemia)	NM_005141.4	4q31.3
Bleeding	FGG	Fibrinogen deficiency	AR (afibrinogenemia); AD (hypo/dysfibrinogenemia)	NM_021870.2	4q32.1
Bleeding/coagulation	GGCX	Vitamin K-dependent clotting factors deficiency 1	AR	NM_000821.6	2p11.2
Coagulation	KLKB1	Prekallikrein (Fletcher) deficiency	AR	NM_000892.5	4q35.2
Coagulation	KNG1	Kininogen Deficiency	AR	NM_000893.4	3q27.3
Bleeding/coagulation	LMAN1	Combined factor V and VIII deficiency	AR	NM_005570.3	18q21.32
Bleeding/coagulation	MCFD2	Combined factor V and VIII deficiency	AR	NM_139279.5	2p21
Bleeding	SERPINE1	Plasminogen activator Inhibitor 1 deficiency	AR; AD	NM_000602.4	7q22.1
Bleeding	SERPINF2	Alpha 2 antiplasmin deficiency	AR	NM_000934.3	17p13.3
Bleeding/coagulation	VKORC1	Vitamin K-dependent clotting factors deficiency 2	AR	NM_024006.5	16p11.2
Bleeding <i>Platelet</i>	VWF	Von Willebrand disease	AD (VWD type 1 and 2) AR (VWD type 3) <i>AD (VWD type 2B)</i>	NM_000552.3	12p13.31
Thrombosis	ADAMTS13	Thrombotic thrombocytopenic purpura (TTP)	AR	NM_139025.4	9q34.2
Thrombosis	HRG	Histidine-rich glycoprotein deficiency	AD	NM_000412.4	3q27.3
Thrombosis	PIGA	Paroxysmal nocturnal hemoglobinuria	acquired (somatic)	NM_002641.3	Xp22.2
Thrombosis	PLG	Plasminogen deficiency	AR	NM_000301.3	3q27.3
Thrombosis	PROC	Protein C deficiency	AR; AD	NM_000312.3	2q14.3
Thrombosis	PROS1	Protein S deficiency	AR; AD	NM_000313.3	3q11.1
Thrombosis	SERPINC1	Antithrombin deficiency	AR; AD	NM_000488.3	1q25.1

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Thrombosis	SERPIND1	Heparin cofactor 2 deficiency	AD	NM_000185.3	22q11.21
Thrombosis	THBD	Thrombomodulin deficiency	AD	NM_000361.2	20p11.21
Bleeding		<i>Bleeding due to high soluble thrombomodulin</i>			
Platelet	ABCC4	Reduced ADP-induced platelet aggregation	AR	NM_005845.4	13q32.1
Platelet	ABCG5	Sitosterolemia with macrothrombocytopenia	AR	NM_022436.2	2p21
Platelet	ABCG8	Sitosterolemia with macrothrombocytopenia	AR	NM_022437.2	2p21
Platelet	ACTB	Baraitser-Winter syndrome 1 with macrothrombocytopenia	AD	NM_001101.3	7p22.1
Platelet	ACTN1	Macrothrombocytopenia	AD	NM_001130004.1	14q24.1
Platelet	ANKRD26	AD thrombocytopenia 2	AD	NM_014915.2	10p12.1
Platelet	ANO6	Scott syndrome	AR	NM_001025356.2	12q12
Platelet	AP3B1	Hermansky-Pudlak syndrome	AR	NM_003664.4	5q14.1
Platelet	AP3D1	Hermansky-Pudlak syndrome	AR	NM_001261826.3	19p13.3
Platelet	ARPC1B	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	AR	NM_005720.4	7q22.1
Platelet	BLOC1S3	Hermansky-Pudlak syndrome	AR	NM_212550.4	19q13.32
Platelet	BLOC1S5	Hermansky-Pudlak syndrome	AR	NM_201280.3	6p24.3
Platelet	BLOC1S6	Hermansky-Pudlak syndrome	AR	NM_012388.3	15q21.1
Platelet	CDC42	Takenouchi-Kosaki syndrome with thrombocytopenia	AD	NM_001791.4	1p36.12
Platelet	CYCS	AD thrombocytopenia 4	AD	NM_018947.5	7p15.3
Platelet	DIAPH1	Macrothrombocytopenia and sensorineural hearing loss	AD	NM_001079812.2	5q31.3
Platelet	DTNBP1	Hermansky-Pudlak syndrome	AR	NM_032122.4	6p22.3
Platelet	ERG	Thrombocytopenia and susceptibility to cancer	AD	NM_182918.4	21q22.2
Platelet	ETV6	Thrombocytopenia and susceptibility to cancer	AD	NM_001987.4	12p13.2
Platelet	FERMT3	Leukocyte integrin adhesion deficiency, type 3	AR	NM_178443.2	11q13.1
Platelet	FLI1	Paris-Trousseau and Jacobson syndrome	AR; AD	NM_002017.4	11q24.3
Platelet	FLNA	Syndrome with macrothrombocytopenia	XLD; XLR	NM_001110556.2	Xq28
Platelet	FYB1	Thrombocytopenia 3	AR	NM_001465.6	5p13.1
Platelet	GATA1	X-linked thrombocytopenia with dyserythropoiesis	XR	NM_002049.3	Xp11.23
Platelet	GF11B	Platelet-type bleeding disorder 17	AD; AR	NM_004188.5	9q34.13
Platelet	GNE	Myopathy associated with TP	AR	NM_005476.6	9p13.3
Platelet	GP1BA	Bernard-Soulier syndrome Mild macrothrombocytopenia Platelet-type von Willebrand disease	AR (BSS) AD (mild macroTP) AD (platelet-type von Willebrand)	NM_000173.5	17p13.2
Platelet	GP1BB	Bernard-Soulier syndrome Mild macrothrombocytopenia	AR (BSS) AD (mild macroTP)	NM_000407.4	22q11.21
Platelet	GP6	Bleeding diathesis due to glycoprotein VI deficiency	AR	NM_016363.5	19q13.42

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Platelet	GP9	Bernard-Soulier syndrome	AR	NM_000174.4	3q21.3
Platelet	HOXA11	Amegakaryocytic thrombocytopenia with radioulnar synostosis	AD	NM_005523.5	7p15.2
Platelet	HPS1	Hermansky-Pudlak syndrome	AR	NM_000195.4	10q24.2
Platelet	HPS3	Hermansky-Pudlak syndrome	AR	NM_032383.4	3q24
Platelet	HPS4	Hermansky-Pudlak syndrome	AR	NM_022081.5	22q12.1
Platelet	HPS5	Hermansky-Pudlak syndrome	AR	NM_181507.1	11p15.1
Platelet	HPS6	Hermansky-Pudlak syndrome	AR	NM_024747.5	10q24.32
Platelet	IKZF5	Thrombocytopenia	AD	n/a	10q26.13
Platelet	ITGA2B	Glanzmann thrombasthenia Platelet-type bleeding disorder 16	AR (GT) AD (bleeding disorder)	NM_000419.3	17q21.31
Platelet	ITGB3	Glanzmann thrombasthenia Platelet-type bleeding disorder 16	AR (GT) AD (bleeding disorder)	NM_000212.2	17q21.32
Platelet	KDSR	Thrombocytopenia and erythroderma	AR	NM_002035.4	18q21.33
Platelet	LYST	Chediak-Higashi syndrome	AR	NM_000081.3	1q42.3
Platelet	MECOM	Amegakaryocytic thrombocytopenia with radioulnar synostosis 2	AD	NM_004991.3	3q26.2
Platelet	MPIG6B	Thrombocytopenia, anaemia and myelofibrosis	AR	NM_025260.3	6p21.33
Platelet	MPL	Congenital amegakaryocytic thrombocytopenia	AR	NM_005373.2	1p34.2
Platelet	MYH9	MYH9-related disorders	AD	NM_002473.5	22q12.3
Platelet	NBEA	Autism with platelet dense granule defect	AD	NM_015678.4	13q13.3
Platelet	NBEAL2	Gray platelet syndrome	AR	NM_015175.2	3p21.31
Platelet	P2RY12	ADP receptor defect	AR	NM_022788.4	3q25.1
Platelet	PLA2G4A	Deficiency of phospholipase A2, group IV A	AR	NM_024420.2	1q31.1
Platelet	PLAU	Quebec platelet disorder	AD	NM_002658.3	10q22.2
Platelet	PTGS1	Bleeding disorder platelet type 12	AR; AD	NM_000962.4	9q33.2
Platelet	RASGRP2	Platelet-type bleeding disorder 18	AR	NM_153819.1	11q13.1
Platelet	RAP1B	Syndromic thrombocytopenia	AD	NM_001010942.3	12q15
Platelet	RBM8A	TP-absent radius syndrome	AR	NM_005105.4	1q21.1
Platelet	RNU4AT AC	Roifman Syndrome	AR	NR_023343.1	2q14.2
Platelet	RUNX1	Familial platelet disorder with predisposition to AML	AD	NM_001754.4	21q22.12
Platelet	SLFN14	Platelet-type bleeding disorder 20	AD	NM_001129820.1	17q12
Platelet	SRC	Thrombocytopenia 6	AD	NM_198291.2	20q11.23
Platelet	STIM1	Stormorken syndrome (York platelet syndrome)	AD	NM_003156.3	11p15.4
Platelet	STXBP2	Familial hemophagocytic lymphohistiocytosis type 5	AR	NM_006949.2	19p13.2
Platelet	TBXA2R	Thromboxane A2 receptor defect	AR; AD (partial phenotype)	NM_001060.5	19p13.3
Platelet	TBXAS1	Ghosal syndrome	AR	NM_030984.3	7q34

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Platelet	THPO	Thrombocytopenia progressing to trilineage bone marrow failure	AR, AD	NM_000460.4	3q27.1
Platelet	TPM4	Macrothrombocytopenia	AD	NM_003290.3	19p13.12
Platelet	TUBB1	Macrothrombocytopenia	AD	NM_030773.3	20q13.32
Platelet	VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 1	AR	NM_001193315.1	14q24.3
Platelet	VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 2	AR	NM_018668.4	15q26.1
Platelet	WAS	Wiskott-Aldrich syndrome	XLR	NM_000377.2	Xp11.23

AD: autosomal dominant. AR: autosomal recessive. XLD: X-linked dominant. XLR: X-linked recessive. Categories in italics indicate a rarer occurrence for a specific gene.