Table 1: Lambert, M. (2018). *Platelets* (4th ed.). Boston, MA: Elsevier, Inc.

Inherited Condition	Gene (Location)	Inheritance	Key Features
MICROTHROMBOCYTIC			
Wiskott–Aldrich Syndrome (WAS)	*WAS* (Xp11)	X-linked	Thrombocytopenia, eczema, severe immunodeficiency, small platelets
X-linked thrombocytopenia (XLT)	*WAS* (Xp11-exon2)	X-linked	Small platelets, thrombocytopenia, mild immunodeficiency
FYB-related thrombocytopenia	*FYB* (5p13.1)	AR	Small platelets and mild to moderate bleeding
ARCP1B related thrombocytopenia	*ARCP1B*(7q22.1)	AR	Microthrombocytopenia, eosinophilia, inflammatory disease
NORMOTHROMBOCYTIC			
Congenital amegakaryocytic thrombocytopenia (CAMT)	*MPL* (1p34)	AR	Hypomegakaryocytic thrombocytopenia with eventual development of bone marrow failure
Thrombocytopenia with absent radii (TAR)	*RBM8A*(1q21.1)	AR	Thrombocytopenia that improves with age, limb anomalies (but normal thumbs)
Radio-ulnar synostosis with amegakaryocytic thrombocytopenia (RUSAT)	*HOXA11* (7p15), *MECOM* (3q26.2)	AD	Severe thrombocytopenia that improves with age, skeletal abnormalities (radio-ulnar synostosis, clinodactyly, syndactyly, hip dysplasia), hearing loss
Familial platelet disorder with predisposition to AML (FPD/AML)	*RUNX1* (21q22)	AD	Thrombocytopenia, myelodysplasia or even AML, platelet dysfunction
Paris–Trousseau/Jacobsen syndrome (PT/JS)	*FLI1* (11p24.3)	AR	Thrombocytopenia with large granules and depending on size of deletion if deleted other symptoms arising from deletion of other genes
Familial thrombocytopenia 2 (THC2)	*ANKRD26* (10p12.1)	AD	Mild to moderate thrombocytopenia with mild bleeding symptoms, cancer predisposition with

Inherited Condition	Gene (Location)	Inheritance	Key Features
			risk of myeloid malignancy and MDS
ETV6-related thrombocytopenia (THC5)	*ETV6* (12p13.2)	AD	Mild to moderate thrombocytopenia, increased risk of hematologic malignancy including ALL, AML and MDS
Monoallelic THPO mutation	*THPO* (3q27.1)	AD	Minimal to no bleeding with low platelet count
CYCS-related thrombocytopenia	*CYCS* (7p15)	AD	Thrombocytopenia without significant bleeding due to abnormal platelet release
SLFN14-related thrombocytopenia	*SLFN14* (17q12)	AD	Variable platelet size (sometimes large) with mild to severe bleeding and impaired platelet function
Stomorken Syndrome/York Platelet Syndrome	*STIM1* (11p15) or *ORAI1* (12q24.31)	AD	Tubular aggregate myopathy and platelet disorder with decreased alpha granules, thrombocytopenia and abnormal function and mild to moderate bleeding
MACROTHROMBOCYTOPENIC			
Bernard–Soulier syndrome (BSS)	*GPIBA* (17p13), *GPIBB* (22q11), *GPIX* (3q21)	AR, AD	Platelet dysfunction with large platelets
Velocardiofacial syndrome (22qDS)	22q11	AD	Cardiac anomalies, cleft palate, hypocalcemia, thymic aplasia, and typical facies. BSS-like thrombocytopenia +/– autoimmune
Platelet-type von Willebrand disease	*GPIBA* (17p13)	AD	Decreased high molecular weight VWF multimers with thrombocytopenia because of increased platelet affinity for VWF

Inherited Condition	Gene (Location)	Inheritance	Key Features
MYH9-related disease (MYH9-RD)	*MYH9* (22q11.2)	AD	Large platelets, leukocyte inclusions; may have sensorineural hearing loss, cataracts, glomerulonephritis, or renal failure
Gray platelet syndrome (GPS)	*NBEAL2* (3p21)	AD, AR	Large, pale platelets with absence of α granules
GATA-1 mutation of X-linked thrombocytopenia with thalassemia (GATA-1)	*GATA1* (Xp11.23)	X-linked	Thrombocytopenia with variable anemia
TUBB1- related thrombocytopenia	*TUBB1* (20q13.32)	AD	Spherocytic platelets and decreased cardiovascular disease in males
Macrothrombocytopenia with Filamin A mutations	*FLNA* (Xq28)	X-Linked	Abnormal granule distribution on EM, mild to moderate thrombocytopenia, impaired aggregation to collagen.
GFI1b-related thrombocytopenia	*GFI1b* (9q24)	AD	Moderate to severe bleeding with gray platelet like phenotype with absent alpha granules and variable red cell anisocytosis
TRPM7-related thrombocytopenia	*TRPM7* (15q21.2)	AD	Large platelets with aberrant granule distribution and mild bleeding
ACTN1-related thrombocytopenia	*ACTN1* (14q24)	AD	Large platelets with absent to mild bleeding
PRKACG-related thrombocytopenia	*PRKACG* (9q21)	AR	Large platelets with aberrant FLNA expression and impaired function
TPM4-related thrombocytopenia	*TPM4* (19p13.1)	AD	Large platelets with mild bleeding
DIAPH1-related thrombocytopenia	*DIAPH1* (5q31.3)	AD	Sensorineural hearing loss, large platelets
SRC-related thrombocytopenia	*SRC* (20q11.23)	AD	Moderate to severe bleeding with hypogranular platelets and

Inherited Condition	Gene (Location)	Inheritance	Key Features
			impaired platelet function and juvenile onset myelofibrosis, osteoporosis
ITGA2B/ITGB3 related thrombocytopenia	*ITGA2B* (17q21) or *ITGB3* (17q21)	AD	Moderate bleeding, large platelets and abnormal function with gain of function variants

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