Disease	Genes affected	Features	Inheritance
ADP receptor defect	P2RY12	Decreased platelet activation and aggregation in response to ADP	AR
AD thrombocytopenia 2	ANKRD26	Increased likelihood of myelodysplastic syndrome/acute myeloid leukemia (only ankyrin repeat domain 26 mutation)	AD
AD thrombocytopenia 4	CYCS		AD
Amegakaryocytic thrombocytopenia with radioulnar synostosis	HOXA11, MECOM	Limited pronation and supination forearm, hip dysplasia	AD
Arthrogryposis-renal dysfunction-cholestasis (ARC) syndrome 1 and 2	VIPAS39; VPS33B	Joint contractures, Renal tubular acidosis, Cholestasis	AR
Baraitser-Winter syndrome 1 with macrothrombocytopenia	ACTB	Mild developmental disability, minor facial anomalies, microcephaly	AD
Bernard-Soulier syndrome or mild macrothrombocytopenia	GP1BA; GP1BB; GP9	Moderate-severe bleeding Reduced expression of Gp1b on platelets	AR (BSS) AD (Mild macroTP)
Bleeding diathesis due to Gp VI deficiency	GP6	Reduced expression of Gp VI on platelets	AR
Chediak-Higashi syndrome	LYST	Occolucutaneus albinism, ataxia, intellectual disability, autoimmunity, Immunodeficiency	AR
Congenital amegakaryocytic thrombocytopenia (CAMT)	MPL	Progression to aplastic anemia and increased risk of myelodysplastic syndrome/acute myeloid leukemia	AR
Deficiency of phospholipase A2, group IVA	PLA2G4A	Gastroduodenal ulcers	AR
Dense granule abnormality	NBEA	Autism	AD
Familial haemophagocytic lymphohistiocytosis, type 5 (FHL 5)	STXBP2	Fever, hepatosplenomegaly (FHL)	AR (FHL) AD (secretion defect)
Familial platelet disorder with predisposition to AML	RUNX1	Leukemia	AD
Ghosal syndrome	TBXAS1	Anemia and diaphyseal dysplasia of long bones	AR
Glanzmann thrombasthenia, Platelet type bleeding disorder 16	ITGA2B; ITGB3	Moderate-severe bleeding Reduced expression of GpIIbIIIa on platelets	AR (GT) AD (bleeding disorder)
Grey platelet syndrome	NBEAL2	Large spleen, myelofibrosis, autoimmunity	AR

Grey platelet like syndrome	GFI1B		AD
Hermansky-Pudlak syndrome	HPS1; AP3B1 (HPS2); HPS3; HPS4; HPS5; HPS6; DTNBP1 (HPS7); BLOC1S3 (HPS8); BLOC1S6 (HPS9)	Occolu cutaneous albinism, fibrosis, granulomatosis colitis	AR
Leucocyte integrin adhesion deficiency, type III	FERMT3	Infections, aberrant woundhealing, periodontitis. Defect signalling of GpIIbIIIa	AR
Macrothrombocytopenia, ACTN1 related	ACTN1		AD
Macrothrombocytopenia, FLNA related	FLNA	Periventricular nodular heterotopia	XR
Macrothrombocytopenia, TUBB1 related	TUBB1		AD
May-Hegglin and other MYH9-related disorders	MYH9	Renal insufficiency:Haematuria-Proteinuria, High-frequency sensory loss, cataract	AD
Myopathy associated with thrombocytopenia	GNE	Myopathy of lower extremity	AR
Paris-Trousseau thrombocytopenia and Jacobson syndrome	FLI1	Eczema, hand abnormalities, facial abnormalities, heart defect, mental retardation	AR or AD
Platelet abnormalities with eosinophilia and immune mediated inflammatory disease	ARPC1B	Combined immunodeficiency, allergy, and auto inflammation	AR
Platelet-type bleeding disorder 17	GFI1b		AD;AR
Platelet-type bleeding disorder 18	RASGRP2		AR
Platelet-type bleeding disorder 20	SLFN14		
Platelet-type von Willebrand disease	GP1BA		AD
Quebec platelet disorder	PLAU		AD
Roifman syndrome	RNU4ATAC	Skeletal dysplasia, immunodeficiency, retinal dystrophy and developmental delay	AR
Scott syndrome	ANO6	Lack of phosphatidyl serine exposure on platelet activation	AR
Sisterolemia with macrothrombocytoepnia	ABCG5; ABCG8	Tendon xanthomas, premature artheriosclerosis, hemolytic anaemia	AR
Stormorken syndrome (York syndrome)	STIM1; ORAI1	Myopathia	AD

Takenouchi-Kosaki syndrome with thrombocytopenia	CDC42	Intellectual disability, camptodactyly, structural brain abnormalities with sensorineural deafness, hypothyroidism, and frequent infections	AD
Thrombocytopenia 3	FYB1		AR
Thrombocytopenia 6	SRC	Myelofibrosis, bone pathologies, premature toothlessness and mild facial dysmorphia	AD
Thrombocytopenia absent radius (TAR) syndrome	RBM8A	Absence radius, presence thumb, various anomalies of lower extremities, septal defect, renal malformations, milk-protein allergy	AR
Thrombocytopenia and sensorineural hearing loss	DIAPH1	Hearing loss	AD
Thrombocytopenia and susceptibility to cancer	ETV6	Leukemia	AD
Thrombocytopenia progessing to trilineage bone marrow failure	THPO	Bone marrow failure	AR
Thrombocytopenia and erythrokeraderma	KDSR	Hyperkeratosis, ichtiosytosis	AR
Thrombocytopenia anaemia and myelofibrosis	MPIG6B	Anaemia, myelofibrosis and splenomegaly	AR
Thrombocytopenia IKZF5 related	IKZF5		AD
Thromboxane A2 receptor defect	TBXA2R		AR;AD (partial phenotype)
Wiskott-Aldrich syndrome	WAS	Eczema, Immunodeficiency	XR
X-linked macrothrombocytopenia with dyserythropoiesis	GATA1	Myelofibrosis, splenomegaly, cryptorchism	XR