

**Dept. of Molecular Medicine (MOMA), Aarhus University Hospital, Denmark
Thrombocyte defects v1 (2021-10-01), 80 genes**

Approved symbol	Approved name
ABCC4	ATP binding cassette subfamily C member 4
ABCG5	ATP binding cassette subfamily G member 5
ABCG8	ATP binding cassette subfamily G member 8
ACTB	actin beta
ACTN1	actinin alpha 1
ANKRD26	ankyrin repeat domain 26
ANO6	anoctamin 6
AP3B1	adaptor related protein complex 3 subunit beta 1
AP3D1	adaptor related protein complex 3 subunit delta 1
APOH	apolipoprotein H
ARPC1B	actin related protein 2/3 complex subunit 1B
BLOC1S3	biogenesis of lysosomal organelles complex 1 subunit 3
BLOC1S6	biogenesis of lysosomal organelles complex 1 subunit 6
CDC42	cell division cycle 42
COL2A1	collagen type II alpha 1 chain
CYCS	cytochrome c, somatic
DIAPH1	diaphanous related formin 1
DTNBP1	dystrobrevin binding protein 1
ETV6	ETS variant transcription factor 6
FERMT3	FERM domain containing kindlin 3
FLI1	Fli-1 proto-oncogene, ETS transcription factor
FLNA	filamin A
FYB1	FYN binding protein 1
GATA1	GATA binding protein 1
GF11B	growth factor independent 1B transcriptional repressor
GNE	glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase
GP1BA	glycoprotein Ib platelet subunit alpha
GP1BB	glycoprotein Ib platelet subunit beta
GP6	glycoprotein VI platelet
GP9	glycoprotein IX platelet
HABP2	hyaluronan binding protein 2
HOXA11	homeobox A11
HPS1	HPS1 biogenesis of lysosomal organelles complex 3 subunit 1
HPS3	HPS3 biogenesis of lysosomal organelles complex 2 subunit 1
HPS4	HPS4 biogenesis of lysosomal organelles complex 3 subunit 2
HPS5	HPS5 biogenesis of lysosomal organelles complex 2 subunit 2
HPS6	HPS6 biogenesis of lysosomal organelles complex 2 subunit 3
IFNAR2	interferon alpha and beta receptor subunit 2
IKZF5	IKAROS family zinc finger 5
ITGA2B	integrin subunit alpha 2b
ITGB3	integrin subunit beta 3

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KDSR	3-ketodihydrosphingosine reductase
LAT	linker for activation of T cells
LPA	lipoprotein(a)
LYST	lysosomal trafficking regulator
MECOM	MDS1 and EVI1 complex locus
MPIG6B	megakaryocyte and platelet inhibitory receptor G6b
MPL	MPL proto-oncogene, thrombopoietin receptor
MTHFR	methylenetetrahydrofolate reductase
MYH9	myosin heavy chain 9
NBEA	neurobeachin
NBEAL2	neurobeachin like 2
ORAI1	ORAI calcium release-activated calcium modulator 1
P2RY12	purinergic receptor P2Y12
PIGA	phosphatidylinositol glycan anchor biosynthesis class A
PLA2G4A	phospholipase A2 group IVA
PLAU	plasminogen activator, urokinase
PROCR	protein C receptor
PROZ	protein Z, vitamin K dependent plasma glycoprotein
PTPRJ	protein tyrosine phosphatase receptor type J
RASGRP2	RAS guanyl releasing protein 2
RBM8A	RNA binding motif protein 8A
RNU4ATAC	RNA, U4atac small nuclear (U12-dependent splicing)
RUNX1	RUNX family transcription factor 1
SERPINA10	serpin family A member 10
SLC35A1	solute carrier family 35 member A1
SLFN14	schlafen family member 14
SPNS1	sphingolipid transporter 1 (putative)
STIM1	stromal interaction molecule 1
STXBP2	syntaxin binding protein 2
TBXA2R	thromboxane A2 receptor
TBXAS1	thromboxane A synthase 1
TFPI	tissue factor pathway inhibitor
THPO	thrombopoietin
TPM4	tropomyosin 4
TUBB1	tubulin beta 1 class VI
VIPAS39	VPS33B interacting protein, apical-basolateral polarity regulator, spe-39 homolog
VPS33B	VPS33B late endosome and lysosome associated
VWF	von Willebrand factor
WAS	WASP actin nucleation promoting factor