

Table S1: Gene mutated in inherited thrombocytopenia

Gene	Human Disease	#N°OMIM	References
Cytoskeleton			
<i>MYH9</i>	MYH9-RD	#600208 #155100 #152640 #605249 #153650	(Balduini et al., 2011; Pecci et al., 2014; Saposnik et al., 2014)
TP0-cMPL axis			
<i>MPL</i>	CAMT	#604498	(Geddis, 2011; Ihara et al., 1999; Varghese et al., 2014)
<i>THPO</i>	THPO-RT		(Noris et al., 2018; Savoia et al., 2007)
Transcription / nuclear factors			
<i>GATA1</i>	GATA1-RD (XLT; XLTT)	#300367 #314050	(Ciovacco et al., 2008; Millikan et al., 2011)
<i>RUNX1</i>	FPD/AML	#601399	(Morgan and Daly, 2017; Schlegelberger and Heller, 2017)
<i>ETV6</i>	ETV6-RT/THC5	#616216	(Karastaneva et al., 2020; Melazzini et al., 2016; Noetzli et al., 2015; Poggi et al., 2017)
<i>GFIIB</i>	GFI1b-RT	#187900	(Beauchemin et al., 2019; Rabbolini et al., 2017; Stevenson et al., 2013)
<i>HOXA11</i>	RUSAT1	#605432	(Horvat-Switzer and Thompson, 2006; Thompson and Nguyen, 2000)
<i>MECOM</i>	RUSAT2	# 616738	(Germeshausen et al., 2018; Niihori et al., 2015)
<i>AI1q23/FLII</i>	Paris-Trousseau Jacobsen syndrome	#188025 #600588 #147791	(Breton-Gorius et al., 1995; Favier et al., 2015)
<i>FLII</i>	FLI1-RT	# 617443	(Stevenson et al., 2015; Vo et al., 2017)
<i>SALL4</i>	IVIC syndrome	# 147750	(Paradisi and Arias, 2007)
<i>RBM8A</i>	TAR	#274000	(Albers et al., 2012; Boussion et al., 2020)
<i>NFE2</i>	NFE2-RT		(Luk et al., 2020)
<i>IKZF5</i>	IKZF5-RT		(Lentaigne et al., 2019)

Kinases/phosphatases

<i>PRKACG</i>	PRKACG-RT	#616176	(Manchev et al., 2014)
<i>SRC</i>	SRC-RT/THC6	# 616937	(Turro et al., 2016)
<i>PTPRJ</i>	PTPRJ-RT		(Marconi et al., 2019)
<i>ANKRD26</i>	ANKRD26-RD/THC2	# 188000	(Bluteau et al., 2014; Ferrari et al., 2017; Noris et al., 2011)
<i>MASTL</i>	Autosomal dominant thrombocytopenia		(Hurtado et al., 2018)

Others

<i>GPIBA</i>	Bernard-Soulier syndrome	#231200	(Bury et al., 2019; Mekchay et al., 2019)
<i>GPIBB</i>	Platelet-type VWD	#231200 #231200	(Nurden, 2014; Savoia et al., 2014)
<i>GP9</i>			
<i>ITGA2B</i>	ITGA2B/ITGB3-RT		(Bury et al., 2016; Favier et al., 2018; Ghevaert et al., 2008; Kunishima et al., 2011; Nurden et al., 2011a)
<i>ITGB3</i>			
<i>FYB1</i>	CARST/THC3		(Levin et al., 2015)
<i>TRPM7</i>	TRPM7-RT		(Stritt et al., 2016a)
<i>NBEAL2</i>	Grey platelet syndrome	#139090	(Bottega et al., 2013; Di Buduo et al., 2016; Gunay-Aygun et al., 2011; Kahr et al., 2011)
<i>SLFN14</i>	SFLN14-RT	# 616913	(Fletcher et al., 2015; Stapley et al., 2020)
<i>CYCS</i>	CYCS-RT/THC4	#612004	(Ledgerwood et al., 2019; Morison et al., 2008; Ong et al., 2017)
<i>GNE</i>	GNE-RT		(Izumi et al., 2014)
<i>STIM1</i>	Stormorken syndrome York platelet syndrome	# 185070	(Bohm and Laporte, 2018; Lacruz and Feske, 2015; Misceo et al., 2014; Morin et al., 2020)
<i>ORAI1</i>	Stormorken syndrome		(Bohm and Laporte, 2018; Lacruz and Feske, 2015; Morin et al., 2020)
<i>ABCG5</i>	ABCG-RT		(Sun et al., 2020)
<i>ABCG8</i>	ABCG-RT		(Sun et al., 2020)
<i>VWF</i>	Type 2B VWF		(Nurden et al., 2009; Nurden et al., 2006)
<i>GALE</i>	GALE-RT		(Seo et al., 2019)
<i>KDSR</i>	KDSR-RT	# 617526	(Takeichi et al., 2017)

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