

## Biological, clinical features and modelization of heterozygous variants of GP1BA and GP1BB genes responsible for constitutional thrombocytopenia

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**Supplemental table 1.**

		ACMG scores			ClinVar
		Varsome	Franklin	InterVar	
<b>GP1BA</b>	p.(Leu73Phe)	4	3.9	4	4/5
	p.(Asn150Ser)	5	3.9	3	4/4
	p.(Leu155Pro)	3.5	3.1	3	fd
<b>GP1BB</b>	p.(Leu137Pro)	4	3.9	4	4
	p.(Cys141*)	4	5	4	4
	p.(Leu16Pro)	4	4.5	3	4/4
	p.(Leu168dup)	3.5	4.5	NC	fd

ACMG scores were calculated according to version 2 (with modulation of the different arguments) based on Richards et al., 2015 and other subsequent publications on the topic. These scores take into account many parameters at the variant locus including population data, literature, protein structure and function, bioinformatic predictions, segregation. ACMG scores are comprised between 1 and 5: 1: benign, 2: likely benign, 3: uncertain significance, 4: likely pathogenic, 5: pathogenic, NC: not calculated.

The three softwares used to perform these calculations can be accessed at

Varsome: <https://varsome.com/>

Franklin: <https://franklin.genoox.com/clinical-db/home>

InterVar: <https://wintervar.wglab.org/>

ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar/>): this database collates the ACMG scores calculated by the teams of experts over the world who have found the variant under consideration. fd: first description.