

Variant: *NM_000419.4(ITGA2B):c.1848G>A (p.Val616=)*

Version: 1.1

CA8602928 [↗](#)

323550 (ClinVar) [↗](#)

Gene: ITGA2B ([HGNC:3674](#))

Condition: Glanzmann's thrombasthenia ([MONDO:0010119](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 4b977a47-a3af-452c-bf3f-c9d6a9c2b38b

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HGVS expressions

NM_000419.4:c.1848G>A

NM_000419.4(ITGA2B):c.1848G>A (p.Val616=)

NC_000017.11:g.44379719C>T

CM000679.2:g.44379719C>T

NC_000017.10:g.42457087C>T

CM000679.1:g.42457087C>T

NC_000017.9:g.39812613C>T

NG_008331.1:g.14787G>A

ENST00000262407.6:c.1848G>A

ENST00000648408.1:c.1279G>A

ENST00000262407.5:c.1848G>A

ENST00000592462.5:n.643G>A

NM_000419.3:c.1848G>A

NM_000419.5:c.1848G>A

Benign

Met criteria codes **3**

BP4 **BP7** **BA1**

Not Met criteria codes **1**

PP4

Evidence Links **0**

Expert Panel

Platelet Disorders VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Platelet Disorders VCEP

The c.1848G>A (p.Val616=) synonymous variant has not been reported in the literature to our knowledge. It is present in the gnomAD African population at an allele frequency of 0.01899. No splice impact is predicted and the variant is not highly conserved. In summary, this variant meets criteria to be classified as benign for GT. GT-specific criteria applied: BA1, BP4, and BP7.

Met criteria codes

BP4			HSF and MaxEntScan do not predict there to be an impact to the splice consensus sequence nor the creation of a new splice site.
BP7			HSF and MaxEntScan do not predict there to be an impact to the splice consensus sequence nor the creation of a new splice site. The nucleotide is not highly conserved (PhyloP score 0.71).
BA1			The overall allele frequency in gnomAD is 0.001924 with a MAF of 0.01899 (472/24,854 alleles) in the African population. This is above the >0.0024 threshold.
Not Met criteria codes			
PP4			This variant has been identified in one patient from internal lab data by an inherited thrombocytopenia panel. The patient had thrombocytopenia with no history of bleeding. Laboratory studies from 2011-2018 have shown platelet counts from 98,000/uL- 127,000/uL, MPVs from 8.8 fL-11.3 fL (reference of 8-12 fL) and WBC from 4.0 k/uL- 5.3 k/uL. There were not phenotypes highly specific to GT, such as impaired aggregation to all agonists except ristocetin or reduced surface expression of GPIIb-GPIIIa.

Curation History 	
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