

Variant: *NM_000419.4:c.1612G>T*

Version: 1.0

CA399802424 [↗](#)

953055 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 9a3e6efe-261b-412b-ab23-fd1ad5dcde27

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HGVS expressions

NM_000419.4:c.1612G>T

NC_000017.11:g.44380142C>A

CM000679.2:g.44380142C>A

NC_000017.10:g.42457510C>A

CM000679.1:g.42457510C>A

NC_000017.9:g.39813036C>A

NG_008331.1:g.14364G>T

ENST00000262407.6:c.1612G>T

ENST00000648408.1:c.1043G>T

ENST00000262407.5:c.1612G>T

ENST00000592226.5:n.1085G>T

ENST00000592462.5:n.407G>T

NM_000419.3:c.1612G>T

NM_000419.5:c.1612G>T

Pathogenic

Met criteria codes **3**

PP4_Moderate

PVS1

PM2_Supporting

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 nonsense variant, c.1612G>T (p.Glu538Ter), has been reported in one compound heterozygous proband (PMID: 25539746), with a phenotype highly specific to GT, and is absent from population databases. This nonsense variant occurs in exon 17 and is predicted to result in NMD. In summary, this variant meets criteria to be classified as Pathogenic for GT. GT-specific criteria applied: PVS1, PM2_supporting, PP4_moderate.

Met criteria codes

PP4_Moderate			One proband has been described in the literature (PMID: 25539746) with a normal platelet count and a history of bleeding. Platelet aggregation was impaired for all agonists except ristocetin. Based on flow cytometry, α IIb β 3 expression was between 5% and 20% and exhibited no binding of PAC-1 activation-specific anti- α IIb β 3 antibody.
PVS1			This nonsense variant occurs in exon 17 of 30 and is predicted to result in NMD.
PM2_Supporting			This variant is absent from population databases including gnomAD, ExAC, and 1000 Genomes.

Curation History 		
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