

Variant: *NM_000212.3:c.1402G>T*

Version: 1.0

CA400029121 [↗](#)

1330338 (ClinVar) [↗](#)

Gene: ITGB3 ([HGNC:3690](#))

Condition: Glanzmann thrombasthenia ([MONDO:0100326](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 95edf71c-af25-4f7c-9c91-da0934a75e1e

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HGVS expressions

NM_000212.3:c.1402G>T

NC_000017.11:g.47292280G>T

CM000679.2:g.47292280G>T

NC_000017.10:g.45369646G>T

CM000679.1:g.45369646G>T

NC_000017.9:g.42724645G>T

NG_008332.2:g.43439G>T

ENST00000696963.1:c.1402G>T

ENST00000559488.7:c.1402G>T

ENST00000559488.5:c.1402G>T

ENST00000560629.1:c.1367G>T

NM_000212.2:c.1402G>T

Pathogenic

Met criteria codes **4**

PP4_Moderate

PM3_Supporting

PM2_Supporting

PVS1

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 ITGB3 nonsense variant NM_000212.3:c.1402G>T (p.Glu468Ter) introduces a premature termination codon in exon 10 of 15 total exons and the resulting mRNA product is predicted to undergo nonsense mediated decay, leading to loss of normal protein function. This variant has been observed in homozygosity in one individual with a phenotype specific for Glanzmann's thrombasthenia (GT) (GT34, PMID: 16463284). Furthermore, this variant is absent from population databases. In summary, this variant meets criteria to be classified as pathogenic for GT. GT-specific criteria applied: PVS1, PM2_Supporting, PM3_Supporting, PP4_Moderate.

Met criteria codes

PP4_Moderate



All requirements for PP4_Moderate are met (GT34 in PMID: 16463284): history of bleeding and impaired aggregation to at least two agonists, but normal or only mildly reduced agglutination with ristocetin.

PM3_Supporting			This variant was reported in homozygosity in one individual (GT34 in PMID: 16463284), sufficient to apply PM3_Supporting.
PM2_Supporting			This variant is absent from all population cohorts in gnomAD, ExAC, 1000 Genomes, and ESP.
PVS1			This variant introduces a termination codon at amino acid position 468 in exon 10 of 15 total exons. The resulting mRNA product is predicted to undergo nonsense mediated decay, leading to loss of normal protein function.

Curation History

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Showing 1 to 1 of 1 rows

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