The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000212.3:c.877C>T

Version: 2.1

CA400025022

953048 (ClinVar)

Gene: ITGB3 (HGNC:3690)
Condition: Glanzmann thrombasthenia (MONDO:0100326)
Inheritance Mode: Autosomal recessive inheritance
UUID: 009e5e3b-aeb7-448e-b62e-d46e1bab6c17
Approved on: 2026/9/3
Published on: 2026/9/4

HGVS expressions

NM_000212.3:c.877C>T
NC_000017.11:g.47287169C>T
CM000679.2:g.47287169C>T
NC_000017.10:g.45364535C>T
CM000679.1:g.45364535C>T
NC_000017.9:g.42719534C>T
NG_008332.2:g.38328C>T
ENST00000696963.1:c.877C>T
ENST00000559488.7:c.877C>T
ENST00000559488.5:c.877C>T
ENST00000560629.1:c.842C>T
ENST00000571680.1:c.877C>T
NM_000212.2:c.877C>T

Likely Pathogenic

Met criteria codes 2

PM2_Supporting PVS1

Not Met criteria codes 2

PP4 PM3

Evidence Links 0

Expert Panel

Platelet Disorders VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Platelet Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2.1*
PDF
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Platelet Disorders VCEP

The NM_000212.3(ITGB3):c.877C>T variant results in premature termination of translation at Gln293 in exon 6/15. The resulting transcript is predicted to undergo NMD (PVS1). The highest population minor allele frequency in gnomAD v4.1 is 0.00001098 (1/91072 alleles) in the South Asian genetic ancestry group, which is lower than the ClinGen PD VCEP threshold (<0.0001; PM2_Supporting). The variant is reported in one compound heterozygous individual who does not meet criteria for PP4 (PMID: 30138987). In summary, this variant meets

the criteria to be classified as likely pathogenic for autosomal recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: PM2_Supporting, PVS1 (PD VCEP specifications version 2.1).

Met criteria codes

PM2_Supporting	✔	The highest population minor allele frequency in gnomAD v4.1 is 0.00001098 (1/91072 alleles) in the South Asian genetic ancestry group, which is lower than the ClinGen PD VCEP threshold (<0.0001; PM2_Supporting).
PVS1	✔	The nonsense variant occurs in exon 6 of 15 and the resulting transcript is predicted to undergo NMD.

Not Met criteria codes

PP4	✖	Patient 8 from PMID: 30138987 with hemorrhage, and <5% αIIbβ3 expression by flow cytometry. This patient does not meet criteria for PP4 due to insufficient aggregometry results (impaired aggregation with 1 agonist and normal with ristocetin).
PM3	✖	Patient 8 in PMID: 30138987 is compound het individual with c.1260G>A (Thr420=) phase unknown, which has been classified Likely Pathogenic by the PD-VCEP. 0.25pt

Curation History

Showing 1 to 3 of 3 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.