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(ITGB3):c.414G>C (p.Val138=)

Version: 2.0

CA8622959

701285 (ClinVar)

Gene: ITGB3 (HGNC:3690)
Condition: Glanzmann thrombasthenia (MONDO:0100326)
Inheritance Mode: Autosomal recessive inheritance
UUID: 8d9f3ce9-f94c-4ca3-973c-a26a6a6968d4
Approved on: 2023/8/15
Published on: 2026/8/7

HGVS expressions

NM_000212.3(ITGB3):c.414G>C (p.Val138=)
NM_000212.3:c.414G>C
NC_000017.11:g.47284495G>C
CM000679.2:g.47284495G>C
NC_000017.10:g.45361861G>C
CM000679.1:g.45361861G>C
NC_000017.9:g.42716860G>C
NG_008332.2:g.35654G>C
ENST00000696963.1:c.414G>C
ENST00000559488.7:c.414G>C
ENST00000559488.5:c.414G>C
ENST00000560629.1:c.379G>C
ENST00000571680.1:c.414G>C
NM_000212.2:c.414G>C

Likely Benign

Met criteria codes 3

BP7 BP4 PM2_Supporting

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

After a comprehensive literature search of the synonymous variant NM_000212.3(ITGB3):c.414G>C (p.Val138=), no individuals with glanzmann thrombasthenia were reported with the variant. The highest population minor allele frequency in gnomAD v4.1 is 8.475e-7 (1/1179980 alleles) in the European (Non-Finnish) genetic ancestry group which is lower than the ClinGen PD VCEP threshold (<0.0001;

PM2_Supporting). In silico predictor spliceAI revealed that the synonymous mutation is not expected to impact splicing and a PhyloP score of -.103 shows that the nucleotide position is not highly conserved (BP4, BP7). Due to conflicting evidence, this variant is classified as likely benign for autosomal recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD-VCEP: PM2_Supporting, BP4, BP7. (PD VCEP specifications version 2.1).

Met criteria codes

BP7	✔	The c.414G>C (p.Val138=) variant is a synonymous that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by phyloP score of -.103 (BP7).
BP4	✔	The c.414G>C (p.Val138=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP4).
PM2_Supporting	✔	The highest population minor allele frequency in gnomAD v4.1 is 8.475e-7 (1/1179980 alleles) in the European (Non-Finnish) genetic ancestry group which is lower than the ClinGen PD VCEP threshold (<0.0001; PM2_Supporting).

Curation History

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