

Variant: *NM_000212.3:c.1303G>T*

Version: 1.0

CA400028645 [↗](#)

1879047 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 50c6d852-43dc-45fe-99cd-7eea271ae7d4

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Published on: 2022/12/7

HGVS expressions

NM_000212.3:c.1303G>T

NC_000017.11:g.47292181G>T

CM000679.2:g.47292181G>T

NC_000017.10:g.45369547G>T

CM000679.1:g.45369547G>T

NC_000017.9:g.42724546G>T

NG_008332.2:g.43340G>T

ENST00000696963.1:c.1303G>T

ENST00000559488.7:c.1303G>T

ENST00000559488.5:c.1303G>T

ENST00000560629.1:c.1268G>T

NM_000212.2:c.1303G>T

Pathogenic

Met criteria codes **3**

PM3_Supporting

PVS1

PM2_Supporting

Not Met criteria codes **1**

PP4

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

NM_000212.3(ITGB3):c.1303G>T (p.Glu435Ter) found in a homozygous proband (PMID:31029159; PM3_supporting) causes a premature stop codon at exon 10/15 and is predicted to undergo nonsense mediated decay (PVS1). The mutation was not found in gnomAD v2.1.1 (PM2_supporting). In conclusion the criteria PVS1, PM2_supporting, and PM3_supporting were applied to reach a classification of pathogenic.

Met criteria codes

PM3_Supporting			This variant has been detected in at least 1 proband with Glanzmann Thrombasthenia. One proband was homozygous for the variant, confirmed via paternal testing (PMID: 31029159). Total points: .5 (PM3_supporting).
PVS1			The c.1303G>T (p.Glu435Ter) variant in exon 10 is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 10/15 and is predicted to lead to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).
PM2_Supporting			This variant is absent from gnomAD v[2.1.1] (PM2_Supporting).

Not Met criteria codes

PP4			At least one patient (Patient 4 in PMID:31029159) with this variant displayed bleeding phenotypes including purpura, menorrhagia, bruising susceptibility, and epistaxis, however, there was no platelet aggregation testing information associated with this individual (Not Met).
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Curation History

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