

Variant: *NM_000212.3:c.1990G>T*

Version: 1.0

CA400033003 [↗](#)

2498364 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 987c5c64-6ff8-412f-9906-26cf77b4102c

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HGVS expressions

NM_000212.3:c.1990G>T

NC_000017.11:g.47300554G>T

CM000679.2:g.47300554G>T

NC_000017.10:g.45377920G>T

CM000679.1:g.45377920G>T

NC_000017.9:g.42732919G>T

NG_008332.2:g.51713G>T

ENST00000696963.1:c.1990G>T

ENST00000559488.7:c.1990G>T

ENST00000559488.5:c.1990G>T

ENST00000560629.1:c.1955G>T

NM_000212.2:c.1990G>T

Pathogenic

Met criteria codes **3**

PVS1

PM3_Supporting

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

Platelet Disorders VCEP

The NM_000212.3(ITGB3):c.1990G>T (p.Glu664Ter) nonsense variant is predicted to cause a premature stop codon in biologically-relevant-exon 12/15 and is predicted to lead to nonsense mediated decay in a gene in which loss-of-function is an established mechanism (PVS1). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). Patient 6, of PMID: 31029159, is reported to have a clinical diagnosis of GT and is homozygous for this variant (PM3_supporting). In summary, this variant meets the criteria to be classified as Pathogenic for

autosomal recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: PVS1, PM2_supporting, PM3_supporting (VCEP specifications version 2.1).

Met criteria codes

PVS1			The NM_000212.3(ITGB3):c.1990G>T (p.Glu664Ter) nonsense variant is predicted to cause a premature stop codon in biologically-relevant-exon 12/15 and is predicted to lead to nonsense mediated decay in a gene in which loss-of-function is an established mechanism (PVS1).
PM3_Supporting			Patient 6 is homozygous (by consanguinity) for this variant 0.5pt (PM3_supporting).
PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Not Met criteria codes

PP4			Patient 7 of PMID: 31029159 is reported to have a clinical diagnosis of GT with a bleeding time of 17min, however insufficient information is provided including a lack of platelet aggregation results (PP4_NotMet).
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Curation History

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