

Variant: *NM_000419.5:c.2315C>G*

Version: 1.1

CA8602740 [↗](#)

2498355 (ClinVar) [↗](#)

Gene: ITGA2B ([HGNC:3674](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 29f62558-f8d5-4060-8180-f46d31f8443b

Approved on: 2026/8/18

Published on: 2026/8/19

HGVS expressions

NM_000419.5:c.2315C>G

NC_000017.11:g.44376341G>C

CM000679.2:g.44376341G>C

NC_000017.10:g.42453709G>C

CM000679.1:g.42453709G>C

NC_000017.9:g.39809235G>C

NG_008331.1:g.18165C>G

ENST00000262407.6:c.2315C>G

ENST00000648408.1:c.1746C>G

ENST00000262407.5:c.2315C>G

ENST00000592462.5:n.1110C>G

NM_000419.3:c.2315C>G

NM_000419.4:c.2315C>G

Uncertain Significance

Met criteria codes **1**

PP4_Moderate

Not Met criteria codes **5**

PP3

PM3

PM2

BS1

BP4

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_000419.5(ITGA2B):c.2315C>G (p.Pro772Arg) missense variant has been reported in at least one patient (Patient 7 in PMID:19172520/GT database record 164) who displayed mucocutaneous bleeding and impaired aggregation with all agonists except ristocetin, which is highly specific for Glanzmann thrombasthenia (PP4_moderate). Additionally, α Ib β 3 surface expression was reduced to <10%, as measured by flow cytometry. In summary, this variant meets the criteria to be classified as uncertain significance for autosomal

recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: PP4_Moderate (VCEP specifications version 2.1).

Met criteria codes

PP4_Moderate			At least one patient (Patient 7 in PMID:19172520/GT database record 164) with this variant displayed mucocutaneous bleeding and impaired aggregation with all agonists except ristocetin, which is highly specific for Glanzmann thrombasthenia (PP4_moderate). Additionally, α IIb β 3 surface expression was reduced to <10%, as measured by flow cytometry. However, ITGA2B and ITGB3 were not sequenced across all exons and intron/exon boundaries.
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Not Met criteria codes

PP3			The computational predictor REVEL gives a score of 0.321, which is below the ClinGen PD VCEP PP3 threshold of >0.7 and does not predict a damaging effect on ITGA2B function. And the computational splicing predictor SpliceAI indicated that the variant has no predicted impact on splicing.
PM3			The authors report the variant c.2315C>G in patient 7 (PMID: 19172520), the GT database (record 164) confirms it is homozygous. Allele frequency is too high (>PM2) to consider here. Opt
PM2			The highest population minor allele frequency in gnomAD v4.1 is 0.0002416 (22/91078 alleles) in the South Asian population. This intermediate allele frequency is lower than the ClinGen PD VCEP threshold (>0.00158) for BS1 but higher than the threshold (<0.0001) for PM2_Supporting.
BS1			The highest population minor allele frequency in gnomAD v4.1 is 0.0002416 (22/91078 alleles) in the South Asian population. This intermediate allele frequency is lower than the ClinGen PD VCEP threshold (>0.00158) for BS1 but higher than the threshold (<0.0001) for PM2_Supporting.
BP4			The computational predictor REVEL gives a score of 0.321, which is above the ClinGen PD VCEP threshold of <0.25.

Curation History [↗](#)



 

 

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