Variant: NM_000419.5(ITGA2B):c.2348+15G>A

Version: 2.0

CA8602722

890528 (ClinVar)

Gene: ITGA2B (HGNC:3674)
Condition: Glanzmann thrombasthenia (MONDO:0100326)
Inheritance Mode: Autosomal recessive inheritance
UUID: 01b0c1b1-ac6f-4eaa-ac15-14754a14eba4
Approved on: 2025-02-18
Published on: 2025-02-18

HGVS expressions

NM_000419.5:c.2348+15G>A
NM_000419.5(ITGA2B):c.2348+15G>A
NC_000017.11:g.44376293C>T
CM000679.2:g.44376293C>T
NC_000017.10:g.42453661C>T
CM000679.1:g.42453661C>T
NC_000017.9:g.39809187C>T
NG_008331.1:g.18213G>A
ENST00000262407.6:c.2348+15G>A
ENST00000648408.1:c.1779+15G>A
ENST00000262407.5:c.2348+15G>A
ENST00000592462.5:n.1143+15G>A
NM_000419.3:c.2348+15G>A
NM_000419.4:c.2348+15G>A

Likely Benign

Met criteria codes 2

BP7 **BP4**

Not Met criteria codes 2

BS1 **PM2**

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.2348+15G>A variant is an intronic variant that is not predicted by SpliceAI to impact splicing (Delta scores <0.03). In addition, it occurs at a nucleotide that is not conserved as shown by phyloP score of -1.96 (BP7). The highest population minor allele frequency in gnomAD v4.0.0 is 0.0003737 (441/1179990 alleles) in the European (non-Finnish) 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. In summary this variant is classified as likely benign for autosomal recessive Glanzmann thrombasthenia, with ACMG criteria applied as specified by the PD VCEP: BP4, BP7

Met criteria codes

BP7	ℹ	✓	The NM_000419.5(ITGA2B):c.2348+15G>A variant is an intronic variant that is not predicted by SpliceAI to impact splicing (Delta scores <0.03). In addition, it occurs at a nucleotide that is not conserved as shown by phyloP score of -1.96 (BP7).
BP4	ℹ	✓	an intronic variant that is not predicted by SpliceAI to impact splicing (Delta scores <0.03)

Not Met criteria codes

BS1	ℹ	✗	The highest population minor allele frequency in gnomAD v4.0.0 is 0.0003737 (441/1179990 alleles) in the European (non-Finnish) 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.
PM2	ℹ	✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000419.5(ITGA2B):c.2348+15G>A	Likely Benign	Glanzmann Thrombasthenia	2025-02-18	2.0	ClinGen Platelet Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2.1	ITGA2B
View	NM_000419.5(ITGA2B):c.2348+15G>A	Uncertain Significance	Glanzmann Thrombasthenia	2024-04-16	1.0	ClinGen Platelet Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2.1	ITGA2B

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