

Variant: *NM_000419.5(ITGA2B):c.2946G>A (p.Val982=)*

Version: 1.0

CA8602501 [↗](#)

1803209 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8b2babb0-eaf0-4d97-a6b3-bf5b77d28a63

Approved on: 2023/12/19

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HGVS expressions

NM_000419.5:c.2946G>A

NM_000419.5(ITGA2B):c.2946G>A (p.Val982=)

NC_000017.11:g.44374468C>T

CM000679.2:g.44374468C>T

NC_000017.10:g.42451836C>T

CM000679.1:g.42451836C>T

NC_000017.9:g.39807362C>T

NG_008331.1:g.20038G>A

ENST00000262407.6:c.2946G>A

ENST00000648408.1:c.2374+191G>A

ENST00000262407.5:c.2946G>A

ENST00000587295.5:c.253+1365G>A

ENST00000588098.1:c.37+191G>A

ENST00000592462.5:n.2645G>A

NM_000419.3:c.2946G>A

NM_000419.4:c.2946G>A

Likely Benign

Met criteria codes **2**

BP4

BP7

Not Met criteria codes **3**

BA1

PM2

BS1

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 c.2946G>A (p.Val982=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing and the nucleotide is only moderately conserved, as shown by phyloP score of 0.703 (BP4, BP7). The highest population minor allele frequency in gnomAD v3.1.2 is 0.0001448 (6/41436 alleles) in the African/African American 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, so no allele frequency criteria were met. In addition, no cases of the variant segregating with Glanzmann thrombasthenia were found in the literature. In summary, this variant meets the criteria to be classified as Likely Benign for autosomal recessive Glanzmann Thrombasthenia with the following ACMG/AMP applied, as specified by the ClinGen PD VCEP: BP4, BP7 (VCEP specifications version 2.1)

Met criteria codes

BP4			The c.2946G>A (p.Val982=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (delta scores <=0.05).
BP7			The c.2946G>A (p.Val982=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (delta scores <=0.05) and the nucleotide is only moderately conserved, as shown by phyloP score of 0.703.

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			The highest population minor allele frequency in gnomAD v3.1.2 is 0.0001448 (6/41436 alleles) in the African/African American 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			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



 

 

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