The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000419.5(ITGA2B):c.2862G>A (p.Val954=)

Version: 1.0

CA8602549

892241 (ClinVar)

Gene: ITGA2B (HGNC:3674)
Condition: Glanzmann thrombasthenia (MONDO:0100326)
Inheritance Mode: Autosomal recessive inheritance
UUID: 4eaa7e56-77dc-4afa-aa3b-0bcc07cd09aa
Approved on: 2024/12/17
Published on: 2024/12/17

HGVS expressions

NM_000419.5:c.2862G>A
NM_000419.5(ITGA2B):c.2862G>A (p.Val954=)
NC_000017.11:g.44374740C>T
CM000679.2:g.44374740C>T
NC_000017.10:g.42452108C>T
CM000679.1:g.42452108C>T
NC_000017.9:g.39807634C>T
NG_008331.1:g.19766G>A
ENST00000262407.6:c.2862G>A
ENST00000648408.1:c.2293G>A
ENST00000262407.5:c.2862G>A
ENST00000587295.5:c.253+1093G>A
ENST00000592462.5:n.2373G>A
NM_000419.3:c.2862G>A
NM_000419.4:c.2862G>A

Likely Benign

Met criteria codes 2

BP7 BP4

Not Met criteria codes 4

BS1 BA1 PP4 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 c.2862G>A (p.Val954=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not highly conserved as shown by phyloP score of 1.07117 (BP7 and BP4). This variant was observed by Illumina as part

of a predisposition screen in an ostensibly healthy population, however, it is not found in the literature to segregate in any GT patients. In summary, this variant meets the criteria to be classified as Likely benign for autosomal recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: BP4 and BP7 (VCEP specifications version 2; date of approval 12/17/2024).

Met criteria codes

BP7	✔	The c.2862G>A (p.Val954=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not highly conserved as shown by phyloP score of 1.07117 (BP7).
BP4	✔	The c.2862G>A (p.Val954=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing, delta scores <0.06 (BP4).

Not Met criteria codes

BS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4	✖	This variant was observed by Illumina as part of a predisposition screen in an ostensibly healthy population, however, it is not found in the literature to segregate in any GT patients.
PM2	✖	The highest population minor allele frequency in gnomAD v4.1.0 is 0.0001068 (8/74904 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.

Curation History

Showing 1 to 1 of 1 rows

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