

Variant: *NM_000419.5(ITGA2B):c.858C>T (p.Val286=)*

Version: 2.1

CA8603312 [↗](#)

323562 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 45d99be7-193f-4943-8eef-03b0407dd395

Approved on: 2024/3/7

Published on: 2024/3/22

HGVS expressions

NM_000419.5(ITGA2B):c.858C>T (p.Val286=)

NM_000419.5:c.858C>T

NC_000017.11:g.44384344G>A

CM000679.2:g.44384344G>A

NC_000017.10:g.42461712G>A

CM000679.1:g.42461712G>A

NC_000017.9:g.39817238G>A

NG_008331.1:g.10162C>T

ENST00000262407.6:c.858C>T

ENST00000648408.1:c.289C>T

ENST00000262407.5:c.858C>T

ENST00000589645.5:n.309C>T

ENST00000591990.5:n.403C>T

ENST00000592075.5:n.227C>T

ENST00000592226.5:n.98C>T

ENST00000592253.5:n.366C>T

ENST00000592944.1:n.540C>T

NM_000419.3:c.858C>T

NM_000419.4:c.858C>T

Likely Benign

Met criteria codes **3**

BP7

BP4

PM2_Supporting

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

NM_000419.5(ITGA2B):c.858C>T (p.Val286=) is a rare synonymous variant. The highest population minor allele frequency in gnomAD v4.0 is 0.00005254 (64/1179956 alleles) in the European (non-Finnish) population, which is lower than the ClinGen PD VCEP threshold (<0.0001; PM2_Supporting). This variant has not been reported in any affected individuals in published literature but has been reported once in a healthy individual as part of a population screen. In silico splicing tools predict this synonymous variant to have no impact on splicing and this is not a highly conserved nucleotide (BP4, BP7). In summary, this variant meets criteria for PM2_supporting, BP4, and BP7; and is therefore classified as likely benign.

Met criteria codes

BP7			Multiple splicing tools (MaxEntScan, HSF, SpliceAI) predict this variant to have no impact on splicing. The nucleotide is not highly conserved (phyloP score = -1.016)
BP4			Multiple splicing tools (MaxEntScan, HSF, SpliceAI) predict this variant to have no impact on splicing. CADD RawScore 0.711042 PHRED 11.24
PM2_Supporting			The highest population minor allele frequency in gnomAD v4.0 is 0.00005254 (64/1179956 alleles) in the European (non-Finnish) population, which is lower than the ClinGen PD VCEP threshold (<0.0001; PM2_Supporting).

Curation History



 

 

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