

Variant: *NM_000419.5(ITGA2B):c.690A>C (p.Pro230=)*

Version: 1.0

CA8603369 [↗](#)

740616 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 56fc53c2-8f04-4229-bb72-e939f3a4f116

Approved on: 2023/8/15

Published on: 2023/9/21

HGVS expressions

NM_000419.5:c.690A>C

NM_000419.5(ITGA2B):c.690A>C (p.Pro230=)

NC_000017.11:g.44385057T>G

CM000679.2:g.44385057T>G

NC_000017.10:g.42462425T>G

CM000679.1:g.42462425T>G

NC_000017.9:g.39817951T>G

NG_008331.1:g.9449A>C

ENST00000262407.6:c.690A>C

ENST00000648408.1:c.121A>C

ENST00000262407.5:c.690A>C

ENST00000589645.5:n.141A>C

ENST00000591990.5:n.52A>C

ENST00000592075.5:n.59A>C

ENST00000592226.5:n.39+107A>C

ENST00000592253.5:n.198A>C

ENST00000592944.1:n.372A>C

NM_000419.3:c.690A>C

NM_000419.4:c.690A>C

Likely Benign

Met criteria codes **3**

BP4

PM2_Supporting

BP7

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

After a comprehensive literature search of the synonymous variant NM_000419.5(ITGA2B):c.690A>C (p.Pro230=), no individuals with Glanzmann thrombasthenia were reported with the variant. The variant has a minor allele frequency of 0.00002891 (1/34592 alleles) in the Latino population population in gnomAD, which meets the threshold criteria for PM2_supporting. In silico predictor spliceAI revealed that the synonymous mutation is not expected to impact splicing and a PhyloP score of -0.982 shows that the nucleotide position is not highly conserved (BP4, BP7). In summary, this variant is classified as likely benign for autosomal recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD-VCEP: PM2_Supporting, BP4, BP7. (PD VCEP specifications version 2.1).

Met criteria codes

BP4			The c.690A>C (p.Pro230=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP4).
PM2_Supporting			The highest population minor allele frequency in gnomAD v2.1.1 is 0.00002891 (1/34592 alleles) in the Latino population, which is lower than the ClinGen PD VCEP threshold (<0.0001; PM2_Supporting).
BP7			The c.690A>C (p.Pro230=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by phyloP score of -0.982 (BP7).

Curation History



 

 

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