

Variant: *NM_000419.4(ITGA2B):c.2916G>A (p.Pro972=)*

Version: 1.0

CA8602530 [↗](#)

435530 (ClinVar) [↗](#)

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

Condition: Glanzmann's thrombasthenia ([MONDO:0010119](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: a2098f23-c14b-4e8a-a5db-be79298db2f6

Approved on: 2021/5/7

Published on: 2021/5/7

HGVS expressions

NM_000419.4(ITGA2B):c.2916G>A (p.Pro972=)

NM_000419.4:c.2916G>A

NC_000017.11:g.44374686C>T

CM000679.2:g.44374686C>T

NC_000017.10:g.42452054C>T

CM000679.1:g.42452054C>T

NC_000017.9:g.39807580C>T

NG_008331.1:g.19820G>A

ENST00000262407.6:c.2916G>A

ENST00000648408.1:c.2347G>A

ENST00000262407.5:c.2916G>A

ENST00000587295.5:c.253+1147G>A

ENST00000588098.1:c.10G>A

ENST00000592462.5:n.2427G>A

NM_000419.3:c.2916G>A

NM_000419.5:c.2916G>A

Benign

Met criteria codes **3**

BP7

BP4

BA1

Not Met criteria codes **1**

PP4

Evidence Links **0**

Expert Panel

Platelet Disorders VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Platelet Disorders VCEP

The c.2916G>A; p.Pro972= synonymous variant has not been reported in the literature to our knowledge. It is present in a non-Finnish European control population at an allele frequency of 0.005351 and no splice impact is predicted. In summary, this variant meets criteria to be classified as benign for GT. GT-specific criteria applied: BA1, BP4, and BP7.

Met criteria codes			
BP7			No significant splicing motif alteration detected using Human Splicing Finder and MaxEntScan. The nucleotide is not highly conserved (PhyloP score -0.45).
BP4			No significant splicing motif alteration detected using Human Splicing Finder and MaxEntScan.
BA1			The overall allele frequency in gnomAD is 0.002856 with a MAF of 0.005351 (689/128,750 alleles) in the non-Finnish European population. This is above the threshold of >0.0024.
Not Met criteria codes			
PP4			This variant has been observed to be heterozygous in two individuals tested with either a "comprehensive platelet disorder panel" or "inherited thrombocytopenia panel". Both have thrombocytopenia with absent or mild bleeding phenotypes. They do not have phenotypes highly specific for GT.

Curation History



 

 

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See Report	Preferred Variant Title	Classification 	Condition	Published Date	Version 	Criteria Specification	Gene
View	NM_000419.4(ITGA2B):c.2916G>A (p.P...	Benign	Glanzmann's Thrombasthenia 	2021-05-07	1.0	-	ITGA2B 

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