

Variant: NM_000419.5:c.1392A>G

Version: 1.1

CA500273575 [↗](#)

2674649 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: f3aae07d-b5ac-444a-a1fa-c90c71213016

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HGVS expressions

NM_000419.5:c.1392A>G

NC_000017.11:g.44380880T>C

CM000679.2:g.44380880T>C

NC_000017.10:g.42458248T>C

CM000679.1:g.42458248T>C

NC_000017.9:g.39813774T>C

NG_008331.1:g.13626A>G

ENST00000262407.6:c.1392A>G

ENST00000648408.1:c.823A>G

ENST00000262407.5:c.1392A>G

ENST00000592226.5:n.632A>G

ENST00000592462.5:n.187A>G

NM_000419.3:c.1392A>G

NM_000419.4:c.1392A>G

Uncertain Significance

Met criteria codes **2**

PM3_Supporting

PM2_Supporting

Not Met criteria codes **2**

PP3

PP4

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 NM_000419.5(ITGA2B):c.1392A>G (p.Pro464=) has been reported in a patient (Patient 10, PMID: 31029159) with a clinical diagnosis of GT but insufficient information for a highly specific diagnosis. This variant is reported in gnomAD v4.1 in the non-Finnish European population with a MAF of 0.000001695 (2/1180034 alleles). This MAF is less than the 0.0001 MAF established by the PD VCEP to assign this

code (PM2_Supporting). This variant was homozygous in the reported individual (Patient 10, PMID: 31029159) with a clinical diagnosis of GT. Since this was a homozygous occurrence and this variant was not seen in the literature elsewhere; per SVI recommendations this receives 0.5pt since this variant has a MAF of <0.0001 (PM3_Supporting). This variant was identified as heterozygous in parents and not homozygous in healthy individuals in the family. In summary, this variant meets the criteria to be classified as uncertain significance for autosomal recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: PM2_supporting, PM3_Supporting (VCEP specifications version 2.1).

Met criteria codes

PM3_Supporting			This variant was homozygous in the reported individual (Patient 10, PMID: 31029159) with a clinical diagnosis of GT. Since this was a homozygous occurrence and this variant was not seen in the literature elsewhere; per SVI recommendations this receives 0.5pt since this variant has a MAF of <0.0001 (PM3_Supporting). This variant was identified as heterozygous in parents and not homozygous in healthy individuals in the family.
PM2_Supporting			This variant is reported in gnomAD v4.1 in the non-Finnish European population with a MAF of 0.000001695 (2/1180034 alleles). This MAF is less than the 0.0001 MAF established by the PD VCEP to assign this code (PM2_Supporting).

Not Met criteria codes

PP3			This variant occurs in the last codon on exon 13 in ITGA2B. There is no REVEL score calculated for this variant, but two in silico predictors (SpliceAI and varSEAK) both predict a splicing impact. SpliceAI (0.47) and varSEAK (Class 5) both predict possible loss of a Donor site but below the threshold for this criteria (PP3_NotMet).
PP4			The patient has a reported clinical diagnosis of GT, but there is no specific information about their phenotype to assign this code (PP4_NotMet).

Curation History



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