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.414C>A (p.Cys138Ter)

Version: 2.0

CA399806043

854735 (ClinVar)

Gene: ITGA2B (HGNC:3674)
Condition: Glanzmann thrombasthenia (MONDO:0100326)
Inheritance Mode: Autosomal recessive inheritance
UUID: eb81f6e4-263e-4297-b459-3141e374cfb5
Approved on: 2024/4/16
Published on: 2024/4/16

HGVS expressions

NM_000419.5(ITGA2B):c.414C>A (p.Cys138Ter)
NM_000419.5:c.414C>A
NC_000017.11:g.44385711G>T
CM000679.2:g.44385711G>T
NC_000017.10:g.42463079G>T
CM000679.1:g.42463079G>T
NC_000017.9:g.39818605G>T
NG_008331.1:g.8795C>A
ENST00000262407.6:c.414C>A
ENST00000262407.5:c.414C>A
ENST00000592944.1:n.99C>A
NM_000419.3:c.414C>A
NM_000419.4:c.414C>A

Likely Pathogenic

Met criteria codes 2

PVS1 PM2_Supporting

Not Met criteria codes 1

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.414C>A (p.Cys138Ter) nonsense variant occurs in exon 4 of 30 and is predicted to result in NMD. This variant has not been reported in the literature, to our knowledge. This highest MAF in gnomADv4.0.0 is 0.00005652 (1/17694 alleles) in the African/African American population, which is below the threshold of <0.0001 for PM2_supporting. In summary, this variant meets criteria to be classified as likely pathogenic for GT. GT-specific criteria applied: PM2_Supporting and PVS1.

Met criteria codes

PVS1	✔	The nonsense variant occurs in exon 4 of 30 and is predicted to result in NMD.
PM2_Supporting	✔	This highest MAF in gnomADv4.0.0 is 0.00005652 (1/17694 alleles) in the African/African American population, which is below the threshold of <0.0001 for PM2_supporting.

Not Met criteria codes

PP4	✖	This variant has been reported once by Invitae in ClinVar (SCV001224491.1) for Glanzmann thrombasthenia. However no patient information is provided regarding phenotype or zygosity.

Curation History

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