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_000212.3:c.792G>A

Version: 1.1

CA400024376

2498356 (ClinVar)

Gene: ITGB3 (HGNC:3690)
Condition: Glanzmann thrombasthenia (MONDO:0100326)
Inheritance Mode: Autosomal recessive inheritance
UUID: 84a0eea3-c89d-4f26-a970-a80d4ac1abe4
Approved on: 2023/2/23
Published on: 2026/2/2

HGVS expressions

NM_000212.3:c.792G>A
NC_000017.11:g.47287084G>A
CM000679.2:g.47287084G>A
NC_000017.10:g.45364450G>A
CM000679.1:g.45364450G>A
NC_000017.9:g.42719449G>A
NG_008332.2:g.38243G>A
ENST00000696963.1:c.792G>A
ENST00000559488.7:c.792G>A
ENST00000559488.5:c.792G>A
ENST00000560629.1:c.757G>A
ENST00000571680.1:c.792G>A
NM_000212.2:c.792G>A

Pathogenic

Met criteria codes 3

PM2_Supporting PVS1 PM3_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 ITGB3 variant c.792G>A (p.Trp264Ter) is a nonsense variant causing a premature stop codon in exon 6/15, and is predicted to undergo nonsense mediated decay (PVS1). The highest population minor allele frequency in gnomAD v2.1.1 is 0.00005782 (2/34590 alleles) in the Latino population, which meets threshold for PM2_supporting. At least one homozygous patient (Patient II-2 in PMID:24357714) with this variant displayed mucocutaneous bleeding and impaired aggregation with all agonists with no αIIbβ3 surface expression as measured by

flow cytometry (PM3_supporting). This patient was also diagnosed with primary ciliary dyskinesia. In summary, based on the available evidence at this time, the c.792G>A variant is classified Pathogenic. GT-specific criteria applied: PM3_supporting, PM2_Supporting, PVS1.

Met criteria codes

PM2_Supporting	✔	The highest population minor allele frequency in gnomAD v2.1.1 is 0.00005782 (2/34590 alleles) in the Latino population, which is lower than the ClinGen PD VCEP threshold (<0.0001; PM2_Supporting).
PVS1	✔	The c.792G>A (p.Trp264Ter) variant in exon 6 is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 6/15 and is predicted to lead to nonsense mediated decay in a gene in which loss-of-function is an established mechanism (PVS1).
PM3_Supporting	✔	One proband was homozygous for the variant (PMID:24357714). Total points: .5 (PM3_Supporting).

Not Met criteria codes

PP4	✖	At least one patient (Patient II-2 in PMID:24357714) with this variant displayed mucocutaneous bleeding and impaired aggregation with all agonists. However, response to ristocetin was not reported. Additionally, αIIbβ3 surface expression was reduced to 0% (<25%), as measured by flow cytometry.

Curation History

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