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_000212.2:c.774_775del

Version: 1.0

CA915940285

953052 (ClinVar)

Gene: ITGB3 (HGNC:3690)
Condition: Glanzmann's thrombasthenia (MONDO:0010119)
Inheritance Mode: Autosomal recessive inheritance
UUID: 4df35ca4-3637-424e-8e92-e66ac5db8751
Approved on: 2020/9/6
Published on: 2021/1/28

HGVS expressions

NM_000212.2:c.774_775del
NC_000017.11:g.47286419_47286420del
CM000679.2:g.47286419_47286420del
NC_000017.10:g.45363785_45363786del
CM000679.1:g.45363785_45363786del
NC_000017.9:g.42718784_42718785del
NG_008332.2:g.37578_37579del
ENST00000696963.1:c.774_775del
ENST00000559488.7:c.774_775del
ENST00000559488.5:c.774_775del
ENST00000560629.1:c.739_740del
ENST00000571680.1:c.774_775del
NM_000212.3:c.774_775del

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PP4_Moderate

Evidence Links 1

Expert Panel

Platelet Disorders VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Platelet Disorders VCEP

The frameshift variant NM_000212.2:c.774_775del results in the immediate creation of a premature stop codon in exon 5 of 15, which is expected to result in NMD. It is absent from population databases but has been reported in one compound heterozygous patient with a phenotype highly specific to GT (PMID: 25539746). In summary, based on the available evidence at this time, the variant is classified as Pathogenic. GT-specific criteria applied: PVS1, PM2_supporting, PP4_moderate.

Met criteria codes

PVS1 ✔

This frameshift variant results in the immediate creation of a premature stop codon in exon 5 of 15, which is expected to result in NMD.

PM2_Supporting ✔ This variant is absent from all population cohorts in gnomAD, ExAC, 1000 Genomes, and ESP.

PP4_Moderate ✔ From PMID: 25539746 The patient had a history of bleeding, normal platelet number and size, impaired aggregation to ADP and arachidonic acid but normal or only mildly reduced agglutination with ristocetin, and αIIbβ3 expression between 5% and 20% (measured by flow cytometry).

PubMed:25539746

Curation History

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