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Variant: NM_000419.5:c.1096C>T

Version: 1.0

CA399804480

1879051 (ClinVar)

Gene: ITGA2B (HGNC:3674)
Condition: Glanzmann thrombasthenia (MONDO:0100326)
Inheritance Mode: Autosomal recessive inheritance
UUID: 649164e8-149c-4032-b343-14e3c680f447
Approved on: 2022/4/7
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HGVS expressions

NM_000419.5:c.1096C>T
NC_000017.11:g.44383607G>A
CM000679.2:g.44383607G>A
NC_000017.10:g.42460975G>A
CM000679.1:g.42460975G>A
NC_000017.9:g.39816501G>A
NG_008331.1:g.10899C>T
ENST00000262407.6:c.1096C>T
ENST00000648408.1:c.527C>T
ENST00000262407.5:c.1096C>T
ENST00000592226.5:n.336C>T
NM_000419.3:c.1096C>T
NM_000419.4:c.1096C>T

Pathogenic

Met criteria codes 4

PVS1 PP4_Moderate PM3_Supporting PM2_Supporting

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 NM_000419.5(ITGA2B):c.1096C>T (p.Arg366Ter) nonsense variant in biologically-relevant-exon 12/30 and is predicted to lead to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). At least one patient (Patient CT in PMID: 12083483) with this variant displayed mucocutaneous bleeding and impaired aggregation with all agonists except ristocetin, which is highly specific for Glanzmann thrombasthenia (PP4_moderate). Patient CT of PMID: 12083483 is homozygous for the Arg366Ter nonsense variant (PM3_supporting). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). In summary, this variant meets the criteria to be classified as Pathogenic for autosomal recessive Glanzmann Thrombasthenia based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: PVS1, PP4_moderate, PM2_supporting, PM3_supporting. (VCEP specifications version 2; date of approval 01/18/2022)

Met criteria codes

PVS1	✔	The c.1096C>T causes the nonsense variant Arg366Ter in biologically-relevant-exon 12/30 and is predicted to lead to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).
PP4_Moderate	✔	At least one patient (Patient CT in PMID: 12083483) with this variant displayed mucocutaneous bleeding and impaired aggregation with all agonists except ristocetin, which is highly specific for Glanzmann thrombasthenia (PP4_moderate).
PM3_Supporting	✔	Patient CT of PMID: 12083483 is homozygous for the Arg366Ter nonsense variant. 0.5pt
PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History

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