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Variant: NM_000173.7:c.882C>G

Version: 1.0

CA397319104

Gene: GP1BA (HGNC:2811)
Condition: Bernard-Soulier syndrome (MONDO:0009276)
Inheritance Mode: Autosomal recessive inheritance
UUID: 7cda8b81-50ce-4788-9fba-d12bd08dd8de
Approved on: 2025/11/6
Published on: 2025/11/7

HGVS expressions

NM_000173.7:c.882C>G
NC_000017.11:g.4933486C>G
CM000679.2:g.4933486C>G
NC_000017.10:g.4836781C>G
CM000679.1:g.4836781C>G
NC_000017.9:g.4777561C>G
NG_008767.2:g.6192C>G
ENST00000329125.6:c.882C>G
ENST00000649830.1:c.-888+856G>C
ENST00000329125.5:c.882C>G
ENST00000611961.1:c.882C>G
NM_000173.6:c.882C>G

Likely Pathogenic

Met criteria codes 4

PM3_Supporting | PP4 | PVS1_Strong | PM2_Supporting

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 for GP1BA Version 1.0.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Platelet Disorders VCEP

The c.882C>G (p.Tyr294Ter) variant in GP1BA is a nonsense variant that may cause loss of function of the protein, however it is predicted to escape nonsense mediated decay and remove >10% of the protein (PVS1_Strong). At least one patient (Patient V in PMID:22672365) with this variant had less than 10% expression of GPIba measured by flow cytometry, which is highly specific for Bernard-Soulier syndrome. Additionally, the patient had excessive mucocutaneous bleeding and macrothrombocytopenia which are consistent with Bernard-Soulier syndrome (PP4). This individual was homozygous for the variant (0.5 PM3 points, PM3_Supporting). The Grpmax Filtering allele frequency in gnomAD v4.1.0 is 0.00000553 (based on 2/60024 alleles) in the Admixed American population, which is lower than the

ClinGen PD VCEP threshold (<0.0001114; PM2_Supporting). In summary, this variant meets the criteria to be classified as Likely Pathogenic for autosomal recessive Bernard-Soulier syndrome based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: PVS1_Strong, PP4, PM3_Supporting and PM2_Supporting (VCEP specifications version 1).

Met criteria codes

PM3_Supporting ✔	This variant has been detected in at least 1 proband with Bernard-Soulier syndrome. This individual was homozygous for the variant (0.5 PM3 points, PM3_Supporting).
PP4 ✔	At least one patient (Patient V in PMID:22672365) with this variant had less than 10% expression of GPIba measured by flow cytometry, which is highly specific for Bernard-Soulier syndrome. Additionally, the patient had excessive mucocutaneous bleeding and macrothrombocytopenia which are consistent with Bernard-Soulier syndrome (PP4).
PVS1_Strong ✔	The c.882C>G (p.Tyr294Ter) variant in GP1BA is a nonsense variant that may cause loss of function of the protein, however it is predicted to escape nonsense mediated decay and remove >10% of the protein (PVS1_Strong).
PM2_Supporting ✔	The Grpmax Filtering allele frequency in gnomAD v4.1.0 is 0.00000553 (based on 2/60024 alleles) in the Admixed American population, which is lower than the ClinGen PD VCEP threshold (<0.0001114; PM2_Supporting).

Curation History

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