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_000173.7(GP1BA):c.624del (p.Phe208fs)

Version: 1.0

CA287216329

3582323 (ClinVar)

Gene: GP1BA (HGNC:2811)
Condition: Bernard-Soulier syndrome (MONDO:0009276)
Inheritance Mode: Autosomal recessive inheritance
UUID: 2326b0f9-7567-45bd-8ccc-5fc415c6b06f
Approved on: 2025/12/16
Published on: 2025/12/16

HGVS expressions

NM_000173.7:c.624del
NM_000173.7(GP1BA):c.624del (p.Phe208fs)
NC_000017.11:g.4933228del
CM000679.2:g.4933228del
NC_000017.10:g.4836523del
CM000679.1:g.4836523del
NC_000017.9:g.4777303del
NG_008767.2:g.5934del
ENST00000329125.6:c.624del
ENST00000649830.1:c.-888+1119del
ENST00000329125.5:c.624del
ENST00000611961.1:c.624del
NM_000173.6:c.624del

Likely Pathogenic

Met criteria codes 4

PM3_Supporting PVS1_Strong PM2_Supporting 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 for GP1BA Version 1.1.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Platelet Disorders VCEP

The c.624del (p.Phe208LeufsTer48) 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 BSS-2 in PMID:23300803) with this variant had aggregation absent for ristocetin and present for all other agonists as well as less than 10% expression of GPIba measured by flow cytometry, which is highly specific for Bernard-Soulier syndrome Additionally, the patient had macrothrombocytopenia which is consistent with Bernard-Soulier syndrome (PP4). This individual was homozygous for the variant (0.5

PM3 points, PM3_Supporting). This variant is absent from gnomAD v4.1.0 (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 1.1).

Met criteria codes

PM3_Supporting	✔	This variant has been detected in at least 1 proband with Bernard-Soulier syndrome (Patient BSS-2 in PMID:23300803). This individual was homozygous for the variant (0.5 PM3 points, PM3_Supporting).
PVS1_Strong	✔	The c.624del (p.Phe208LeufsTer48) 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	✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP4	✔	At least one patient (Patient BSS-2 in PMID:23300803) with this variant had aggregation absent for ristocetin and present for all other agonists as well as less than 10% expression of GPIba measured by flow cytometry, which is highly specific for Bernard-Soulier syndrome Additionally, the patient had macrothrombocytopenia which is consistent with Bernard-Soulier syndrome (PP4).

Curation History

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