

Variant: *NM_000407.5(GP1BB):c.433C>T (p.Pro145Ser)*

Version: 1.0

CA322080045 [↗](#)

2581461 (ClinVar) [↗](#)

Gene: GP1BB ([HGNC:2812](#))

Condition: Bernard-Soulier syndrome ([MONDO:0009276](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: c40b450f-6051-4902-b499-25d7c7bc7b10

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HGVS expressions

NM_000407.5:c.433C>T

NM_000407.5(GP1BB):c.433C>T (p.Pro145Ser)

NC_000022.11:g.19724276C>T

CM000684.2:g.19724276C>T

NC_000022.10:g.19711799C>T

CM000684.1:g.19711799C>T

NC_000022.9:g.18091799C>T

NG_007974.1:g.5734C>T

ENST00000366425.4:c.433C>T

ENST00000366425.3:c.433C>T

ENST00000431044.5:c.*1518C>T

NM_000407.4:c.433C>T

NR_037611.1:n.4173C>T

NR_037612.1:n.2677C>T

Benign

Met criteria codes **2**

BA1

BP4

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 GP1BB Version 1.1*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Platelet Disorders VCEP

The NM_000407.5(GP1BB):c.433C>T (p.Pro145Ser) variant in GP1BB is a missense variant that has a Grpmax Filtering allele frequency in gnomAD v4.1.1 of 0.002995 (based on 215/63908 alleles) in the African/African American population, which is higher than the ClinGen PD VCEP threshold (>0.001), and there for meets benign by frequency in the general population (BA1). The variant is predicted to not affect the protein (REVEL score of 0.115; BP4) In summary, this variant meets the criteria to be classified as benign for autosomal recessive

Bernard-Soulier syndrome. ACMG/AMP criteria applied, as specified by the ClinGen Platelet Disorders VCEP (specifications version 1.1.0): BA1, BP4.

Met criteria codes

BA1			The Grpmax Filtering allele frequency in gnomAD v4.1.1 is 0.002995 (based on 215/63908 alleles) in the African/African American population, which is higher than the ClinGen PD VCEP threshold (>0.001), and there for meets benign by frequency in the general population (BA1).
BP4			The variant is predicted to not affect the protein (REVEL score of 0.115; BP4), with SpliceAI indicating no affect on splicing (all scores equal to zero).

Curation History



 

 

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