

Variant: *NM_000174.5(GP9):c.423G>A (p.Ala141=)*

Version: 1.0

CA2602711 [↗](#)

900154 (ClinVar) [↗](#)

Gene: GP9 ([HGNC:2815](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 3cbfb15e-6bc3-4678-8d0f-6927b2627923

Approved on: 2025/2/11

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HGVS expressions

NM_000174.5:c.423G>A

NM_000174.5(GP9):c.423G>A (p.Ala141=)

NC_000003.12:g.129062162G>A

CM000665.2:g.129062162G>A

NC_000003.11:g.128781005G>A

CM000665.1:g.128781005G>A

NC_000003.10:g.130263695G>A

NG_008715.1:g.6361G>A

ENST00000307395.5:c.423G>A

ENST00000307395.4:c.423G>A

NM_000174.4:c.423G>A

Likely Benign

Met criteria codes **2**

BP4

BP7

Not Met criteria codes **2**

BS1

PM2

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 GP9 Version 1.0.0

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

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

Evidence submitted by expert panel

Platelet Disorders VCEP

The c.423G>A variant is a synonymous (silent) variant (p.Ala141=) that is not predicted by SpliceAI to impact splicing (score <0.2, BP4). In addition, it occurs at a nucleotide that is not highly conserved as shown by phyloP score of -1.8 (<1.5) (BP7). The Grpmax Filtering allele frequency in gnomAD v4.1 is 0.0001647 (based on 216/1167496alleles) in the European (non-Finnish) population, which is above the ClinGen PD VCEP PM2_supporting threshold (<0.0000329) but below the BS1 threshold (>0.0007). In summary, this variant meets the criteria to be classified as likely benign for autosomal recessive Bernard-Soulier syndrome based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: BP4, BP7

Met criteria codes

BP4			The computational splicing predictor SpliceAI indicated that the variant has no impact on splicing.
BP7			The c.423G>A (p.Ala141=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (score <0.2). In addition, it occurs at a nucleotide that is not highly conserved as shown by phyloP score of -1.8 (<1.5) (BP7).

Not Met criteria codes

BS1			The Grpmax Filtering allele frequency in gnomAD v4.1 is 0.0001647 (based on 216/1167496 alleles) in the European (non-Finnish) population, which is above the ClinGen PD VCEP PM2_supporting threshold (<0.0000329) but below the BS1 threshold (>0.0007).
PM2			The Grpmax Filtering allele frequency in gnomAD v4.1 is 0.0001647 (based on 216/1167496alleles) in the European (non-Finnish) population, which is above the ClinGen PD VCEP PM2_supporting threshold (<0.0000329) but below the BS1 threshold (>0.0007).

Curation History



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