

Variant: *NM_000173.7(GP1BA):c.1074A>G (p.Arg358=)*

Version: 1.0

CA8314893 [↗](#)

255463 (ClinVar) [↗](#)

Gene: GP1BA ([HGNC:2811](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: edcbd259-7427-4001-8c6a-5a30e40c8e15

Approved on: 2025/2/11

Published on: 2025/2/12

HGVS expressions

NM_000173.7(GP1BA):c.1074A>G (p.Arg358=)

NM_000173.7:c.1074A>G

NC_000017.11:g.4933678A>G

CM000679.2:g.4933678A>G

NC_000017.10:g.4836973A>G

CM000679.1:g.4836973A>G

NC_000017.9:g.4777753A>G

NG_008767.2:g.6384A>G

ENST00000329125.6:c.1074A>G

ENST00000649830.1:c.-888+664T>C

ENST00000329125.5:c.1074A>G

ENST00000611961.1:c.1074A>G

NM_000173.6:c.1074A>G

Benign

Met criteria codes **3**

BA1

BP7

BP4

Not Met criteria codes **1**

BP2

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 synonymous variant NM_000173.7(GP1BA):c.1074A>G (p.Arg358=) is not predicted by SpliceAI to impact splicing (score 0.00; BP4). In addition, it occurs at a nucleotide that is not highly conserved as shown by phyloP score of 0.134386 (BP7). The Grpmax Filtering allele frequency in gnomAD v4.1 is 0.2116 (based on 9653/44866 alleles, including 1018 homozygotes) in the East Asian population, which is higher than the ClinGen PD VCEP threshold (>0.001; BA1). In summary, this variant meets the criteria to be classified as Benign for

autosomal recessive Bernard-Soulier syndrome based on the ACMG/AMP criteria applied, as specified by the ClinGen PD VCEP: BA1, BP4, BP7 (ClinGen Platelet Disorders VCEP specifications version 1).

Met criteria codes

BA1			The Grpmax Filtering allele frequency in gnomAD v4.1 is 0.2116 (based on 9653/44866 alleles, including 1018 homozygotes) in the East Asian population, which is higher than the ClinGen PD VCEP threshold (>0.001), and therefore meets this criterion (BA1).
BP7			This synonymous variant is not predicted by SpliceAI to impact splicing (score 0.00). In addition, it occurs at a nucleotide that is not highly conserved as shown by phyloP score of 0.134386 (BP7).
BP4			The computational splicing predictor SpliceAI indicated that the variant has no impact on splicing (scores 0.00).

Not Met criteria codes

BP2			This variant has been observed in cis with the variants GP1BA c.1136_1143del and GPIBB c.124_145 del which are asserted to cause BSS in the respective patients (PMID: 19448529 and PMID: 21699652) however pathogenicity has not yet been assessed by the ClinGen PD VCEP.
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Curation History



 

 

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