

Variant: *NM_000133.4(F9):c.769G>A (p.Val257Ile)*

Version: 1.0

CA10529834 [↗](#)

225354 (ClinVar) [↗](#)

Gene: F9 ([HGNC:2158](#))

Condition: hemophilia B ([MONDO:0010604](#))

Inheritance Mode: X-linked inheritance

UUID: 38622bb4-32e1-4c95-a640-f3cd85e5a081

Approved on: 2025/4/4

Published on: 2025/4/4

HGVS expressions

NM_000133.4(F9):c.769G>A (p.Val257Ile)

NM_000133.4:c.769G>A

NC_000023.11:g.139560786G>A

CM000685.2:g.139560786G>A

NC_000023.10:g.138642945G>A

CM000685.1:g.138642945G>A

NC_000023.9:g.138470611G>A

NG_007994.1:g.35051G>A

ENST00000218099.7:c.769G>A

ENST00000643157.1:n.1436G>A

ENST00000218099.6:c.769G>A

ENST00000394090.2:c.655G>A

NM_000133.3:c.769G>A

NM_001313913.1:c.655G>A

NM_001313913.2:c.655G>A

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **2**

BP4

PP3

Evidence Links **0**

Expert Panel

Coagulation Factor Deficiency VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** *ClinGen Coagulation Factor Deficiency Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for F9 Version 1.0.0*

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

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

Evidence submitted by expert panel

Coagulation Factor Deficiency VCEP

The c.769G>A variant in F9 is a missense variant predicted to cause substitution of Valine by Isoleucine at amino acid 257 (p.Val257Ile). The highest population minor allele frequency in gnomAD v2.1.1 is 0.001548 (23/14862 alleles) in the East Asian population a grpmax FAF for exomes = 0.001015 and genomes = 0.0003535, which is higher than the ClinGen Coagulation Factor Deficiency VCEP threshold

(≥ 0.0000556) for BA1, and therefore meets this criterion (BA1). In summary, this variant meets the criteria to be classified as Benign for Hemophilia B based on the ACMG/AMP criteria applied, as specified by the ClinGen Coagulation Factor Deficiency VCEP for F9 (version 1.0.0, released 10/5/2023): BA1.

Met criteria codes

BA1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.001548 (23/14862 alleles) in the East Asian population with a grpmx FAF for exomes = 0.001015 and genomes= 0.0003535, which is higher than the ClinGen Coagulation Factor Deficiency VCEP threshold (≥ 0.0000556) for BA1, and therefore meets this criterion (BA1).
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Not Met criteria codes

BP4			The computational predictor REVEL gives a score of 0.483, which is neither above nor below the thresholds predicting a damaging (≥ 0.6) or benign (≤ 0.3) impact on F9 function.
PP3			The computational predictor REVEL gives a score of 0.483, which is neither above nor below the thresholds predicting a damaging (≥ 0.6) or benign (≤ 0.3) impact on F9 function.

Curation History



 

 

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