

Variant: *NM_000133.4(F9):c.224G>A (p.Arg75Gln)*

Version: 1.0

CA255318 [↗](#)

10573 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 178efdc9-7922-4e7d-8479-352a42eedd02

Approved on: 2024/10/11

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HGVS expressions

NM_000133.4(F9):c.224G>A (p.Arg75Gln)

NM_000133.4:c.224G>A

NC_000023.11:g.139537145G>A

CM000685.2:g.139537145G>A

NC_000023.10:g.138619304G>A

CM000685.1:g.138619304G>A

NC_000023.9:g.138446970G>A

NG_007994.1:g.11410G>A

ENST00000218099.7:c.224G>A

ENST00000218099.6:c.224G>A

ENST00000394090.2:c.224G>A

ENST00000479617.2:n.231G>A

NM_000133.3:c.224G>A

NM_001313913.1:c.224G>A

NM_001313913.2:c.224G>A

Pathogenic

Met criteria codes **4**

PP4_Moderate

PM2_Supporting

PP3

PS4_Very Strong

Not Met criteria codes **1**

PM1

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

NM_000133.4(F9):c.224G>A (p.Arg75Gln) missense variant has a REVEL score of 0.904, which meets criteria for PP3. This variant is absent from males in population databases (gnomAD v2.1.1/gnomAD v3). Approximately 24 patients are reported in the literature with mild hemophilia B and this variant, meeting F9 phenotype criteria for PS4_Very strong (PMID: 31064749, PMID: 2066105, PMID: 8314564, PMID:

29296726, PMID: 27213901, PMID: 2773937). In summary, this variant meets criteria to be classified as pathogenic. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F9: PS4_Very strong, PP4_Moderate, PP3, PM2_supporting.

Met criteria codes

PP4_Moderate			Counting one of the MLOF probands
PM2_Supporting			This variant is absent from males in population databases (gnomAD v2.1.1/gnomAD v3).
PP3			REVEL score is 0.904, which is below the 0.6 cutoff.
PS4_Very Strong			Greater than 16 individuals reported in the literature with mild hemophilia B. The criterion is met at the very strong evidence level.

Not Met criteria codes

PM1			Variant is not within exons 3, 4 or 5.
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Curation History



 

 

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