

Variant: NM_000133.4(F9):c.196G>A (p.Glu66Lys)

Version: 1.0

CA414435972 [↗](#)

618643 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 4cb5c6fe-6600-4352-a05c-eedc77bf64c7

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HGVS expressions

NM_000133.4:c.196G>A

NM_000133.4(F9):c.196G>A (p.Glu66Lys)

NC_000023.11:g.139537117G>A

CM000685.2:g.139537117G>A

NC_000023.10:g.138619276G>A

CM000685.1:g.138619276G>A

NC_000023.9:g.138446942G>A

NG_007994.1:g.11382G>A

ENST00000218099.7:c.196G>A

ENST00000218099.6:c.196G>A

ENST00000394090.2:c.196G>A

ENST00000479617.2:n.203G>A

NM_000133.3:c.196G>A

NM_001313913.1:c.196G>A

NM_001313913.2:c.196G>A

Likely Pathogenic

Met criteria codes **4**

PM2_Supporting

PP4_Moderate

PP3

PM5

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 2.0.0

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

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

Evidence submitted by expert panel

Coagulation Factor Deficiency VCEP

The NM_000133.4(F9):c.196G>A; p.Glu66Lys variant is a missense variant in F9 that is absent from gnomAD v4.1.0 (PM2_Supporting) and is predicted to have a deleterious effect (REVEL score of 0.939; PP3). The variant has been observed in at least one proband in the literature with mild hemophilia B (PMID:29296726, 2 points PP4; PP4_Moderate). Another missense variant (c.198A>T, p.Glu66Asp,

CAID:CA414435987) in the same codon has been classified as pathogenic for hemophilia B by the ClinGen Coagulation Factor Deficiency VCEP meeting PM5. In summary, this variant meets the criteria to be classified as likely pathogenic for hemophilia B. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel (specifications version 2.0.0) for F9: PP4_Moderate, PM5, PM2_Supporting, PP3.

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP4_Moderate			There is at least 1 reported proband with mild hemophilia B (PMID:29296726, 2 points PP4; PP4_Moderate).
PP3			This variant is predicted to have a deleterious effect (REVEL score of 0.939; PP3), which is greater than the Clingen CFD threshold for PP3 (>0.6), with splicing predicted to be affected (SpliceAI Donor gain score greater than 0.2).
PM5			Another missense variant (c.198A>T, p.Glu66Asp, CAID:CA414435987) in the same codon has been classified as pathogenic for hemophilia B by the ClinGen Coagulation Factor Deficiency VCEP (PM5), with splicing predicted to be unaffected for the c.198A>T, p.Glu66Asp variant.

Curation History

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