

Variant: NM_001313913.2:c.197A>T

Version: 1.0

CA414435980 [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 737f396c-3747-4eda-afd0-74746afce526

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HGVS expressions

NM_001313913.2:c.197A>T

NC_000023.11:g.139537118A>T

CM000685.2:g.139537118A>T

NC_000023.10:g.138619277A>T

CM000685.1:g.138619277A>T

NC_000023.9:g.138446943A>T

NG_007994.1:g.11383A>T

ENST00000218099.7:c.197A>T

ENST00000218099.6:c.197A>T

ENST00000394090.2:c.197A>T

ENST00000479617.2:n.204A>T

NM_000133.3:c.197A>T

NM_001313913.1:c.197A>T

NM_000133.4:c.197A>T

Likely Pathogenic

Met criteria codes **4**

PM2_Supporting

PP3

PS4

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.197A>T; p.Glu66Val variant is a missense variant in F9 that is absent from from gnomAD v4.1.0 (PM2_Supporting) and is predicted to have a deleterious effect (REVEL score of 0.98; PP3). The variant has been observed in at least four patients in the literature with moderate to severe hemophilia B (PMID:34880139, 3 points PS4; PMID:7937052, 1 point PS4; Total 4 points PS4, PS4). 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: PS4, PM5, PM2_Supporting, PP3**.

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP3			This variant is predicted to have a deleterious effect (REVEL score of 0.98; 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).
PS4			This variant has been observed in at least 4 patients in the literature with moderate to severe hemophilia B (PMID:34880139, 3 points PS4; PMID:7937052, 1 point PS4; Total 4 points PS4, PS4).
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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