The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_001313913.2:c.198A>T

Version: 1.0

CA414435987

Gene: F9 (HGNC:2158)
Condition: hemophilia B (MONDO:0010604)
Inheritance Mode: X-linked inheritance
UUID: 8da5f010-6b77-4b3e-a9b8-895444c4c697
Approved on: 2026/4/10
Published on: 2026/4/10

HGVS expressions

NM_001313913.2:c.198A>T
NC_000023.11:g.139537119A>T
CM000685.2:g.139537119A>T
NC_000023.10:g.138619278A>T
CM000685.1:g.138619278A>T
NC_000023.9:g.138446944A>T
NG_007994.1:g.11384A>T
ENST00000218099.7:c.198A>T
ENST00000218099.6:c.198A>T
ENST00000394090.2:c.198A>T
ENST00000479617.2:n.205A>T
NM_000133.3:c.198A>T
NM_001313913.1:c.198A>T
NM_000133.4:c.198A>T

Pathogenic

Met criteria codes 3

PP3 PS4_Very Strong PM2_Supporting

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.198A>T; p.Glu66Asp 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 0.908; PP3). The variant has been observed in at least eleven patients in the literature with moderate to severe hemophilia B, both with and without inhibitors (PMID:34880139, 11 points PS4, PS4_Very_Strong). In

summary, this variant meets the criteria to be classified as 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_Very strong, PM2_Supporting, PP3.

Met criteria codes

PP3	✔	This variant is predicted to have a deleterious effect (REVEL score of 0.908; PP3), which is greater than the Clingen CFD threshold for PP3 (>0.6), with splicing predicted to be unaffected (SpliceAI scores less than 0.1).
PS4_Very Strong	✔	This variant has been observed in at least 11 patients in the literature with moderate to severe hemophilia B, both with and without inhibitors (PMID:34880139, 11 points PS4, PS4_Very_Strong).
PM2_Supporting	✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting).

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001313913.2:c.198A>T	Pathogenic	Hemophilia B	2026-04-10	1.0	ClinGen Coagulation Factor Deficiency Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for F9 Version 2.0.0	F9

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