

Variant: NM_001313913.2:c.688T>G

Version: 1.0

CA414443538 [🔗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: f0c61402-c999-4ccc-a283-8a8afd80213f

Approved on: 2025/10/3

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HGVS expressions

NM_001313913.2:c.688T>G

NC_000023.11:g.139560819T>G

CM000685.2:g.139560819T>G

NC_000023.10:g.138642978T>G

CM000685.1:g.138642978T>G

NC_000023.9:g.138470644T>G

NG_007994.1:g.35084T>G

ENST00000218099.7:c.802T>G

ENST00000643157.1:n.1469T>G

ENST00000218099.6:c.802T>G

ENST00000394090.2:c.688T>G

NM_000133.3:c.802T>G

NM_001313913.1:c.688T>G

NM_000133.4:c.802T>G

Likely Pathogenic

Met criteria codes **5**

PS2_Moderate

PP3

PM5

PS4_Supporting

PM2_Supporting

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

The c.802T>G (NM_000133.4) variant in F9 is a missense variant predicted to cause substitution of cysteine by glycine at amino acid 268 (p.Cys268Gly). There is at least 1 proband with isolated reduction in FIX activity levels meeting phenotypic criteria for F9, with assumed de novo occurrence (PS4_Supporting & PS2_Moderate: PMID: 10647899). This variant is absent from males in population databases (gnomAD v2.1.1/gnomAD v3.1/ gnomAD v4.1.0), meeting PM2_Supporting. The computational predictor REVEL gives a score of 0.961, which is above

the threshold of 0.6, evidence that correlates with impact to F9 function (PP3). A different missense variant causing a change at the same residue (p.Cys268Tyr) has been established as pathogenic for F9 and SpliceAI predicts no impact on splicing (PM5). In summary, this variant meets criteria to be classified as likely pathogenic. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F9: PS4_Supporting + PM2_Supporting + PP3 + PM5 + PS2_Moderate. (ClinGen Coagulation Factor Deficiency Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for F9 Version 1.0.0., Released 10/5/2023).

Met criteria codes

PS2_Moderate			Variant found to originate in mother of proband with severe hemophilia B.
PP3			REVEL score is 0.961, which is above 0.6 cutoff.
PM5			A different missense variant causing a change at the same residue (p.Cys268Tyr) has been established as pathogenic for F9 and SpliceAI predicts no impact on splicing.
PS4_Supporting			Variant identified in 1 proband with severe hemophilia B.
PM2_Supporting			Variant is absent in gnomAD v2.1.1, v3.1, and v4.1.0.

Not Met criteria codes

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

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