

Variant: *NM_001313913.2:c.1189T>G*

Version: 1.0

CA414447216 [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 791488da-e5d8-4596-95b1-53079e87e817

Approved on: 2024/5/9

Published on: 2024/7/11

HGVS expressions

NM_001313913.2:c.1189T>G

NC_000023.11:g.139561988T>G

CM000685.2:g.139561988T>G

NC_000023.10:g.138644147T>G

CM000685.1:g.138644147T>G

NC_000023.9:g.138471813T>G

NG_007994.1:g.36253T>G

ENST00000218099.7:c.1303T>G

ENST00000643157.1:n.1723+247T>G

ENST00000218099.6:c.1303T>G

ENST00000394090.2:c.1189T>G

NM_000133.3:c.1303T>G

NM_001313913.1:c.1189T>G

NM_000133.4:c.1303T>G

Likely Pathogenic

Met criteria codes **4**

PP3

PM5

PS4_Moderate

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 F9 c.1303T>G; p.Cys435Gly variant is completely absent from gnomAD v2.1.1 and v3.1.1. This missense variant has a REVEL score of 0.964 and meets PP3 criteria (threshold >0.6). One proband is reported in the literature with moderate hemophilia B and this variant and a second proband was reported with severe hemophilia B, meeting F9 phenotype criteria for PS4_Moderate. Cys35Tyr is another pathogenic

variant at the same residue (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_Moderate, PM5, PP3, PM2_Supporting.

Met criteria codes

PP3			REVEL score is 0.964, which is above the 0.6 cutoff.
PM5			The F9 c. Cys435Tyr variant reaches a pathogenic classification by the CFD VCEP rule specifications.
PS4_Moderate			2 probands with hemophilia B (one severe and one moderate) in the literature.
PM2_Supporting			Variant is absent from gnomAD v2.1.1 and 3.1.

Not Met criteria codes

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



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