

Variant: NM_001313913.2:c.155T>C

Version: 1.0

CA414435714 [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: ed2faf01-ae5c-4aeb-a9dc-538bf39f3dd4

Approved on: 2025/4/4

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HGVS expressions

NM_001313913.2:c.155T>C
NC_000023.11:g.139537076T>C
CM000685.2:g.139537076T>C
NC_000023.10:g.138619235T>C
CM000685.1:g.138619235T>C
NC_000023.9:g.138446901T>C
NG_007994.1:g.11341T>C
ENST00000218099.7:c.155T>C
ENST00000218099.6:c.155T>C
ENST00000394090.2:c.155T>C
ENST00000479617.2:n.162T>C
NM_000133.3:c.155T>C
NM_001313913.1:c.155T>C
NM_000133.4:c.155T>C

Likely Pathogenic

Met criteria codes **3**

PS4 **PP3** **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.155T>C; p.Leu52Ser missense variant is completely absent from gnomADv2.1.1 and v3.1.1 meeting PM2_supporting. This missense variant has a REVEL score of 0.931 (>0.6) meeting PP3. Three probands with moderate (Turkish) and severe (Italian) and unknown severity (Chinese) hemophilia B are reported in the literature (PMID: 12588353, PMID: 15921378, PMID: 32875744) and two probands were reported from a VCEP member, meeting F9 phenotype criteria. 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, PP3, PM2_Supporting.

Met criteria codes			
PS4			Three patients with moderate (Turkish), severe (Italian) and unknown severity (Chinese) hemophilia B are reported in the literature (PMID: 12588353, PMID: 15921378, PMID: 32875744) and two patients reported from internal lab data, meeting F9 phenotype criteria
PP3			TheLeu52Ser missense variant has a REVEL score of 0.931 (>0.6) and meets PP3 criteria
PM2_Supporting			The c.155T>C (p.Leu52Ser) missense variant is completely absent from gnomADv2.1.1 and v3.1.1, meeting criteria for PM2_Supporting
Not Met criteria codes			
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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