

Variant: *NM_000133.4(F9):c.130C>T (p.Pro44Ser)*

Version: 1.0

CA10529740 [↗](#)

1085717 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: b6a6a78e-c328-47bf-a216-244b17937741

Approved on: 2025/8/15

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HGVS expressions

NM_000133.4(F9):c.130C>T (p.Pro44Ser)

NM_000133.4:c.130C>T

NC_000023.11:g.139537051C>T

CM000685.2:g.139537051C>T

NC_000023.10:g.138619210C>T

CM000685.1:g.138619210C>T

NC_000023.9:g.138446876C>T

NG_007994.1:g.11316C>T

ENST00000218099.7:c.130C>T

ENST00000218099.6:c.130C>T

ENST00000394090.2:c.130C>T

ENST00000479617.2:n.137C>T

NM_000133.3:c.130C>T

NM_001313913.1:c.130C>T

NM_001313913.2:c.130C>T

Likely Benign

Met criteria codes **2**

BS1 **BP4**

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.130C>T; p.Pro44Ser variant has a Grpmax MAF of 0.00005825 in gnomAD v2.1.1 meeting BS1. The variant also has a REVEL score of 0.144 and Splice AI scores: Acceptor Loss: 0.00, Donor Loss: 0.05, Acceptor Gain: 0.00, Donor Gain: 0.00, meeting BP4. In summary, this

variant meets criteria to be classified as likely benign. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F9: **BS1**, **BP4**.

Met criteria codes

BS1			The c.130C>T, p.Pro44Ser variant has a MAF of 0.00005825 in gnomAD v2.1.1 meeting BS1.
BP4			The c.130C>T, p.Pro44Ser variant has a REVEL score of 0.144 and Splice AI scores: Acceptor Loss: 0.00, Donor Loss: 0.05, Acceptor Gain: 0.00, Donor Gain: 0.00.

Curation History



 

 

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