

Variant: *NM_000133.4(F9):c.471T>C (p.Cys157=)*

Version: 1.2

CA518861298 [↗](#)

798429 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 5abc37e1-ba45-4fb1-8f39-596f10a548c6

Approved on: 2026/2/6

Published on: 2026/2/27

HGVS expressions

NM_000133.4:c.471T>C

NM_000133.4(F9):c.471T>C (p.Cys157=)

NC_000023.11:g.139548442T>C

CM000685.2:g.139548442T>C

NC_000023.10:g.138630601T>C

CM000685.1:g.138630601T>C

NC_000023.9:g.138458267T>C

NG_007994.1:g.22707T>C

ENST00000218099.7:c.471T>C

ENST00000643157.1:n.1138T>C

ENST00000218099.6:c.471T>C

ENST00000394090.2:c.357T>C

NM_000133.3:c.471T>C

NM_001313913.1:c.357T>C

NM_001313913.2:c.357T>C

Uncertain Significance

Met criteria codes **2**

BP4

PM2_Supporting

Not Met criteria codes **1**

BP7

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 F9 c.471T>C (p.Cys157=) variant is absent from males in population databases (gnomAD v4.1.0). The Cys157= synonymous variant has an acceptor and donor gain SpliceAI score of 0.08 (<0.1 cutoff) based on meeting the BP4 code. The BP7 is not met due to its conservation scores. We are not aware of any cases of hemophilia B in the literature or in databases at this time. In summary, based on

the evidence available at this time, the clinical significance of this variant is uncertain. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F9 v2.0.0: PM2_Supporting, BP4.

Met criteria codes

BP4			No REVEL prediction available for the synonymous variant. The Cys157= synonymous variant is predicted to have no impact on splicing based on SpliceAI prediction. A cryptic donor site is predicted to be created at position +49 with a delta score of 0.08, which is less than the 0.1 cutoff.
PM2_Supporting			The c.471T>C (p.Cys157=) variant is absent from males in population databases (gnomAD v2.1.1/gnomAD v3).

Not Met criteria codes

BP7			The Cys157= synonymous variant is predicted to have minimal to no impact on splicing based on SpliceAI prediction but the nucleotide is conserved to a degree, based on agreement between PhyloP (0.445) and PhastCons (1) .
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Curation History



 

 

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