

Variant: NM_000133.4(F9):c.720G>A (p.Trp240Ter)

Version: 1.0

CA414441410 [↗](#)

810867 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 9da956d4-9ba6-49ac-884f-f34f4a2f4c86

Approved on: 2024/2/9

Published on: 2024/7/11

HGVS expressions

NM_000133.4:c.720G>A

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NC_000023.11:g.139551261G>A

CM000685.2:g.139551261G>A

NC_000023.10:g.138633420G>A

CM000685.1:g.138633420G>A

NC_000023.9:g.138461086G>A

NG_007994.1:g.25526G>A

ENST00000218099.7:c.720G>A

ENST00000643157.1:n.1387G>A

ENST00000218099.6:c.720G>A

ENST00000394090.2:c.606G>A

NM_000133.3:c.720G>A

NM_001313913.1:c.606G>A

NM_001313913.2:c.606G>A

Pathogenic

Met criteria codes **3**

PM2_Supporting

PVS1

PS4

Not Met criteria codes **1**

PP1

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 NM_000133.4(F9):c.720G>A variant predicts a nonsense change, Trp240Ter, in exon 6 of the F9 gene; NMD is predicted, meeting PVS1 criteria. The variant is absent from gnomAD v2.1.1 and v3, meeting PM2_Supporting criteria. At least 8 severe hemophilia B patients meeting the F9 phenotype criteria have been reported in the literature (PMIDs: 23093250, 18479429, 8091381, 15086324), meeting

PS4_Very strong. Inhibitors to factor replacement therapies have been reported (EAHAD database). In summary, the variant meets criteria to be classified as pathogenic. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency VCEP for F9: PVS1, PS4_Very strong, PM2_Supporting.

Met criteria codes

PM2_Supporting			The variant is absent from gnomAD v2.1.1 and v3.
PVS1			The Trp240Ter variant occurs in exon 6/8 of the F9 gene. NMD is predicted.
PS4			At least 8 severe hemophilia B patients meeting F9 phenotype criteria have been reported in the literature (PMIDs: 23093250, 18479429, 8091381, 15086324) to carry the Trp240Ter variant. Some of the severe patients are noted to have developed inhibitors.

Not Met criteria codes

PP1			2 Irish families (PMID: 18479429) reported with severe hemophilia B and the Trp240Ter variant. Members are male; however information is not available to count the accurate number of meioses to apply PP1.
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Curation History

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