

Variant: *NM_000133.3(F9):c.108C>T (p.Asn36=)*

Version: 1.0

CA10529735 [↗](#)

367998 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 427ba16e-f4c5-4c03-a24f-46db9355483b

Approved on: 2024/2/9

Published on: 2024/7/11

HGVS expressions

NM_000133.3(F9):c.108C>T (p.Asn36=)

NM_000133.3:c.108C>T

NC_000023.11:g.139537029C>T

CM000685.2:g.139537029C>T

NC_000023.10:g.138619188C>T

CM000685.1:g.138619188C>T

NC_000023.9:g.138446854C>T

NG_007994.1:g.11294C>T

ENST00000218099.7:c.108C>T

ENST00000218099.6:c.108C>T

ENST00000394090.2:c.108C>T

ENST00000479617.2:n.115C>T

NM_001313913.1:c.108C>T

NM_000133.4:c.108C>T

NM_001313913.2:c.108C>T

Benign

Met criteria codes **3**

BP4

BP7

BA1

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.108C>T (p.Asn36=) variant is reported at an MAF of 0.0002820 (5/17730 alleles) in the South Asian population in gnomAD v2.1.1 with 1 hemizygote, meeting BA1 criteria of MAF > 0.0000556. SpliceAI predicts no splicing impact, with delta scores of 0, meeting BP4. The nucleotide is not conserved, with PhyloP score of -0.14 and PhastCons score of 0.031, meeting BP7. In summary, this variant meets criteria

to be classified as benign. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F9: BA1, BP4, BP7.

Met criteria codes

BP4			SpliceAI predicts no splicing impact, with delta scores of 0.
BP7			SpliceAI predicts no splicing impact, with delta scores of 0. The nucleotide is not conserved, with PhyloP score of -0.14 and PhastCons score of 0.031
BA1			The c.108C>T (p.Asn36=) variant is reported at an MAF of 0.0002820 (5/17730 alleles) in the South Asian population in gnomAD v2.1.1 with 1 hemizygote, meeting BA1 criteria of MAF > 0.0000556.

Curation History



 ▾

 ▾

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.