The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000133.3(F9):c.88+5G>T

Version: 1.1

CA915952341

627156 (ClinVar)

Gene: F9 (HGNC:2158)
Condition: hemophilia B (MONDO:0010604)
Inheritance Mode: X-linked inheritance
UUID: 67d79118-0240-41eb-a752-48b2d3ccaf41
Approved on: 2025/3/10
Published on: 2025/3/28

HGVS expressions

NM_000133.3:c.88+5G>T
NM_000133.3(F9):c.88+5G>T
NC_000023.11:g.139530857G>T
CM000685.2:g.139530857G>T
NC_000023.10:g.138613016G>T
CM000685.1:g.138613016G>T
NC_000023.9:g.138440682G>T
NG_007994.1:g.5122G>T
ENST00000218099.7:c.88+5G>T
ENST00000218099.6:c.88+5G>T
ENST00000394090.2:c.88+5G>T
ENST00000479617.2:n.95+5G>T
NM_001313913.1:c.88+5G>T
NM_000133.4:c.88+5G>T
NM_001313913.2:c.88+5G>T

Likely Pathogenic

Met criteria codes 3

PS4 PP3 PM2_Supporting

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.88+5G>T variant is completely absent in population databases (gnomAD v2.1.1/gnomAD v3.1.2) meeting PM2_Supporting. This intronic variant is predicted to alter splicing based on SpliceAI prediction that the canonical donor site is lost, with a score of 0.91 (>0.5) meeting PP3 criteria. It has been reported in at least 4 probands in the literature with moderate to severe hemophilia B meeting

phenotypic criteria for F9 (PMIDs: 31064749, 9452115, 10094553). 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 F8/F9: PS4, PP3, PM2_Supporting.

Met criteria codes

PS4	✔	The c.88+5G>T variant has been reported in at least 4 probands in the literature with moderate to severe hemophilia B (0 gnomAD alleles) meeting phenotypic criteria for F9.
PP3	✔	The c.88+5G>T intronic variant is predicted to alter splicing based on SpliceAI prediction of canonical donor site loss with score of 0.91 (threshold >0.5)
PM2_Supporting	✔	The c.88+5G>T variant is completely absent in population databases (gnomAD v2.1.1/gnomAD v3).

Curation History

Showing 1 to 2 of 2 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000133.3(F9):c.88+5G>T	Likely Pathogenic	Hemophilia B	2025-03-28	1.1	ClinGen Coagulation Factor Deficiency Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for F9 Version 1.0.0	F9
View	NM_000133.3(F9):c.88+5G>T	Likely Pathogenic	Hemophilia B	2025-03-10	1.0	ClinGen Coagulation Factor Deficiency Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for F9 Version 1.0.0	F9

Showing 1 to 2 of 2 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.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.