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_000133.4(F9):c.88+1G>A

Version: 2.2

CA414434506

627155 (ClinVar)

Gene: F9 (HGNC:2158)
Condition: hemophilia B (MONDO:0010604)
Inheritance Mode: X-linked inheritance
UUID: 9738debd-d565-4111-8eda-77a661e2697a
Approved on: 2026/2/6
Published on: 2026/3/3

HGVS expressions

NM_000133.4(F9):c.88+1G>A
NM_000133.4:c.88+1G>A
NC_000023.11:g.139530853G>A
CM000685.2:g.139530853G>A
NC_000023.10:g.138613012G>A
CM000685.1:g.138613012G>A
NC_000023.9:g.138440678G>A
NG_007994.1:g.5118G>A
ENST00000218099.7:c.88+1G>A
ENST00000218099.6:c.88+1G>A
ENST00000394090.2:c.88+1G>A
ENST00000479617.2:n.95+1G>A
NM_000133.3:c.88+1G>A
NM_001313913.1:c.88+1G>A
NM_001313913.2:c.88+1G>A

Pathogenic

Met criteria codes 3

PM2_Supporting PVS1 PS4_Moderate

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.88+1G>A variant is a canonical splice variant that is predicted to use a cryptic splice donor and introduce a premature stop codon and is expected to result in nonsense-mediated mRNA decay meeting PVS1. This splice site variant is completely absent from gnomAD v2.1.1. and v3.1.2 meeting PM2_Supporting. At least one proband with severe hemophilia B has been reported

(PS4_Supporting; PMID: 31064749). 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 F9: PVS1, PS4_Supporting, PM2_Supporting.

Met criteria codes

PM2_Supporting	✔	The c.88+1G>A splice site variant is completely absent from gnomAD v2.1.1. and v3.1.2
PVS1	✔	The c.88+1G>A variant is a canonical splice variant that is predicted to use a cryptic splice donor and introduce a premature stop codon and is expected to result in nonsense-mediated mRNA decay, PVS1_Strong is applied.
PS4_Moderate	✔	At least two probands with severe hemophilia B have been reported (PMID: 31064749; EAHAD Database).

Curation History

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