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Variant: NM_001313913.2:c.86C>T

Version: 1.2

CA414434494

2138734 (ClinVar)

Gene: F9 (HGNC:2158)
Condition: hemophilia B (MONDO:0010604)
Inheritance Mode: X-linked inheritance
UUID: 57ee28b4-485e-499c-ac29-4702ddc55d99
Approved on: 2024/11/1
Published on: 2025/3/7

HGVS expressions

NM_001313913.2:c.86C>T
NC_000023.11:g.139530850C>T
CM000685.2:g.139530850C>T
NC_000023.10:g.138613009C>T
CM000685.1:g.138613009C>T
NC_000023.9:g.138440675C>T
NG_007994.1:g.5115C>T
ENST00000218099.7:c.86C>T
ENST00000218099.6:c.86C>T
ENST00000394090.2:c.86C>T
ENST00000479617.2:n.93C>T
NM_000133.3:c.86C>T
NM_001313913.1:c.86C>T
NM_000133.4:c.86C>T

Likely Pathogenic

Met criteria codes 4

PM2_Supporting PP4_Moderate PS4_Moderate PP3

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.86C>T (p.Thr29Ile) variant is completely absent from gnomAD v2.1.1 and gnomAD v3.1.1 meeting criteria for PM2_Supporting. This missense variant has a REVEL score of 0.659 (>0.6) meeting criteria for PP3. At least 4 mild hemophilia B patients, including 1 female patient, are reported in the literature (PMID: 29296726) meeting F9 phenotype criteria for PS4_moderate and PP4_moderate. 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: PS4_Moderate, PP4_Moderate, PP3, PM2_Supporting.

Met criteria codes

PM2_Supporting	✔	The c.86C>T (p.Thr29Ile) variant is completely absent from gnomAD v2.1.1 and gnomAD v3.1.1
PP4_Moderate	✔	Counting one proband from MLOF
PS4_Moderate	✔	At least 3 mild hemophilia B patients, including 1 female patient, are reported in the literature (PMID: 29296726) meeting F9 phenotype criteria. (FIX:C in males = 16%, 28.4%, 34%; FIX:C in female = 33%)
PP3	✔	This missense variant has a REVEL score of 0.659 (>0.6)

Curation History

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