

Variant: *NM_000133.3(F9):c.19A>T (p.Ile7Phe)*

Version: 1.0

CA10529708 [↗](#)

367997 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: b888a6e8-55cd-449a-a9d9-bc586edb7b6c

Approved on: 2024/2/9

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HGVS expressions

NM_000133.3(F9):c.19A>T (p.Ile7Phe)

NM_000133.3:c.19A>T

NC_000023.11:g.139530783A>T

CM000685.2:g.139530783A>T

NC_000023.10:g.138612942A>T

CM000685.1:g.138612942A>T

NC_000023.9:g.138440608A>T

NG_007994.1:g.5048A>T

ENST00000218099.7:c.19A>T

ENST00000218099.6:c.19A>T

ENST00000394090.2:c.19A>T

ENST00000479617.2:n.26A>T

NM_001313913.1:c.19A>T

NM_000133.4:c.19A>T

NM_001313913.2:c.19A>T

Benign

Met criteria codes **2**

BA1 **BS2**

Not Met criteria codes **2**

PS4 **BP4**

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

NM_000133.3:c.19A>T variant results in a missense change in the signal peptide. This variant is reported at a frequency of 0.001643 (152/92537 alleles with 57 hemizygotes) in the non-Finnish European population in gnomAD v2.1.1, meeting the BA1 cut-off of ≥ 0.0000556 . At least 2 individuals with the variant and normal FIX levels from the literature and internal laboratory data are reported

(PMID: 29296726) meeting the BS2 criterion. In summary, the 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, BS2.

Met criteria codes

BA1			The Ile7Phe variant is reported at a frequency of 0.001643 (152/92537 alleles with 57 hemizygotes) in gnomAD v2.1.1 and at a frequency of 0.001600 (81/50618 alleles with 26 hemizygotes) in gnomAD v3, both in the non-Finnish European population, meeting the BA1 cut-off of ≥ 0.0000556 as determined by the CFD VCEP.
BS2			From internal laboratory data: Male individual with normal factor IX that was confirmed in follow-up testing as well. At least 2 individuals with the variant and normal FIX levels from PMID:29296726 and internal laboratory data meet criteria for BS2.

Not Met criteria codes

PS4			PS4 is not applied when BA1 is met. From internal laboratory data: the variant is reported in an individual with severe HB. PMID: 10094553 reports a patient with moderate HB.
BP4			BP4 is not applied since neither CADD score of 17.04 nor REVEL score of 0.582 meet the threshold for BP4 determined by the CFD VCEP. (CADD: BP4 <11 , PP3 <21 ; REVEL: BP4 <0.3 , PP3 >0.6).

Curation History



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