

Variant: *NM_000133.4(F9):c.7C>T (p.Arg3Cys)*

Version: 1.0

CA10529705 [↗](#)

765903 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: e7e8cade-6031-4598-a3f1-257a702d1088

Approved on: 2026/2/6

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HGVS expressions

NM_000133.4(F9):c.7C>T (p.Arg3Cys)

NM_000133.4:c.7C>T

NC_000023.11:g.139530771C>T

CM000685.2:g.139530771C>T

NC_000023.10:g.138612930C>T

CM000685.1:g.138612930C>T

NC_000023.9:g.138440596C>T

NG_007994.1:g.5036C>T

ENST00000218099.7:c.7C>T

ENST00000218099.6:c.7C>T

ENST00000394090.2:c.7C>T

ENST00000479617.2:n.14C>T

NM_000133.3:c.7C>T

NM_001313913.1:c.7C>T

NM_001313913.2:c.7C>T

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **1**

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

The NM_000133.4(F9):c.7C>T; p.Arg3Cys variant is a missense variant in F9 that has a MAF of 0.0006498 (9/13851 alleles) in the East Asian population in gnomAD v2.1.1 with 3 hemizygotes, meeting BA1 criteria of MAF > 0.0000556. 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 v1.0 rule specifications for F9: BA1.

Met criteria codes

BA1			The c.7C>T (p.Arg3Cys) variant is reported at an MAF of 0.0006498 (9/13851 alleles) in the East Asian population in gnomAD v2.1.1 with 3 hemizygotes, meeting BA1 criteria of MAF > 0.0000556.
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Not Met criteria codes

BP4			The c.7C>T (p.Arg3Cys) missense variant has a REVEL score of 0.157 (less than 0.3), however, it has a SpliceAI greater than 0.01.
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Curation History



 

 

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