

Variant: *NM_000133.4(F9):c.60A>G (p.Leu20=)*

Version: 1.0

CA10529715 [↗](#)

701352 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: a1170f31-d66e-4804-818f-4fc81a684a07

Approved on: 2024/2/9

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HGVS expressions

NM_000133.4:c.60A>G

NM_000133.4(F9):c.60A>G (p.Leu20=)

NC_000023.11:g.139530824A>G

CM000685.2:g.139530824A>G

NC_000023.10:g.138612983A>G

CM000685.1:g.138612983A>G

NC_000023.9:g.138440649A>G

NG_007994.1:g.5089A>G

ENST00000218099.7:c.60A>G

ENST00000218099.6:c.60A>G

ENST00000394090.2:c.60A>G

ENST00000479617.2:n.67A>G

NM_000133.3:c.60A>G

NM_001313913.1:c.60A>G

NM_001313913.2:c.60A>G

Benign

Met criteria codes **3**

BP7 **BP4** **BA1**

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.60A>G (p.Leu20=) synonymous variant is reported at an MAF of 0.0009351 (7/7486 alleles) in the Ashkenazi Jewish population in gnomAD v2.1.1 with 4 hemizygotes, meeting BA1 criteria of MAF > 0.0000556. SpliceAI predicts no splicing impact with a score of 0.0, meeting BP4 and BP7 criteria (<0.01). In summary, based on the evidence available at this time, the clinical significance of

this variant is benign. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F9: BA1, BP4 and BP7.

Met criteria codes

BP7			SpliceAI predicts no splicing impact with a score of 0.0, meeting BP7 criteria (<0.01). The variant meets the conservation thresholds with scores of -0.22 for PhyloP (<0.1) and 0.96 for PhastCons (<1).
BP4			The synonymous variant does not have a REVEL score. SpliceAI predicts no splicing impact with a score of 0.0, meeting BP4 criteria (<0.01)
BA1			The c.60A>G (p.Leu20=) variant is reported at an MAF of 0.0009351 (7/7486 alleles) in the Ashkenazi Jewish population in gnomAD v2.1.1 with 4 hemizygotes, meeting BA1 criteria of MAF > 0.0000556. However, it is unclear if inbred populations should be considered. The variant is also reported at an MAF of 0.00001226 (1/81585 alleles) in the non-Finnish European population in gnomAD v2.1.1 with 1 hemizygote, which does not meet BA1 criteria. BA1 is applied but has to be verified with experts that inbred populations may be used for BA1/BS1.

Curation History



 

 

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