

Variant: *NM_000133.4:c.712T>G*

Version: 1.0

CA10529821 [↗](#)

1166447 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 3b02dca7-ec7c-43b8-a80b-8da7a82a2691

Approved on: 2024/2/9

Published on: 2024/7/11

HGVS expressions

NM_000133.4:c.712T>G

NC_000023.11:g.139551253T>G

CM000685.2:g.139551253T>G

NC_000023.10:g.138633412T>G

CM000685.1:g.138633412T>G

NC_000023.9:g.138461078T>G

NG_007994.1:g.25518T>G

ENST00000218099.7:c.712T>G

ENST00000643157.1:n.1379T>G

ENST00000218099.6:c.712T>G

ENST00000394090.2:c.598T>G

NM_000133.3:c.712T>G

NM_001313913.1:c.598T>G

NM_001313913.2:c.598T>G

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **3**

PS4

PP3

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_000133.3:c.712T>G predicts a missense variant, Phe238Val, that is reported at a high MAF of 0.0008074 (12/14862 alleles with 4 hemizygotes) in the East Asian population of gnomAD v2.1.1, meeting the BA1 cut-off of ≥ 0.0000556 . It is reported in the literature in at least one severe Hemophilia B patient who has an additional F9 variant. In summary, this variant meets criteria to be classified as benign. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency VCEP for F9: BA1.

Met criteria codes

BA1

i

✓

Phe238Val is reported in gnomAD v2.1.1 at a frequency of 0.0008074 (12/14862 alleles with 4 hemizygotes) in the East Asian population and in gnomAD v3 at a frequency of 0.00005947 (3/50446 alleles with 1 hemizygote) in the NFE population. gnomAD V2.1.1 MAF meets the BA1 threshold for F9 (≥ 0.0000556).

Not Met criteria codes

PS4

i

✗

The variant has been reported in at least 1 individual of Taiwanese origin. PS4 is not considered while BA1 is met.

PP3

i

✗

REVEL score of 0.547 and CADD score of 18.80 do not meet threshold for PP3 (REVEL >0.6 ; CADD >21)

BP4

i

✗

REVEL score of 0.547 and CADD score of 18.80 do not meet threshold for BP4 (REVEL <0.3 ; CADD <11)

Curation History [↗](#)

⛶

☰

▼

⬇

▼

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.