

Variant: *NM_000132.3(F8):c.1508G>A (p.Arg503His)*

Version: 1.0

CA10568446 [↗](#)

368123 (ClinVar) [↗](#)

Gene: F8 ([HGNC:2157](#))

Condition: hemophilia A ([MONDO:0010602](#))

Inheritance Mode: X-linked inheritance

UUID: 1de0f1c9-786e-4718-b55f-ed3dbd3daccd

Approved on: 2024/2/1

Published on: 2024/7/11

HGVS expressions

NM_000132.3:c.1508G>A

NM_000132.3(F8):c.1508G>A (p.Arg503His)

NC_000023.11:g.154961104C>T

CM000685.2:g.154961104C>T

NC_000023.10:g.154189379C>T

CM000685.1:g.154189379C>T

NC_000023.9:g.153842573C>T

NG_011403.1:g.66620G>A

NG_011403.2:g.66620G>A

ENST00000360256.9:c.1508G>A

ENST00000647125.1:c.*1384G>A

ENST00000360256.8:c.1508G>A

NM_000132.4:c.1508G>A

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **2**

BP4

BP7

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 F8 Version 1.0.0*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Coagulation Factor Deficiency VCEP

The c.1508G>A (p.Arg503His) variant is reported at an MAF of 0.01797(342/19037 alleles) in the African/African-American population in gnomAD v2.1.1 with 94 hemizygotes and 3 homozygotes, meeting BA1 criteria of MAF >0.000333. 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 for F8/F9: BA1.

Met criteria codes

BA1			The c.1508G>A (p.Arg503His) variant is reported at an MAF of 0.01797(342/19037 alleles) in the African/African-American population in gnomAD v2.1.1 with 94 hemizygotes and 3 homozygotes, meeting BA1 criteria of MAF >0.000333.
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Not Met criteria codes

BP4			The variant has a REVEL score of 0.429 and does not meet criteria for BP4 (<0.3). SpliceAI predicts a donor loss with a score of 0.01, meeting BP4 threshold for F8 (<0.05)
BP7			SpliceAI predicts a donor loss with a score of 0.01, meeting BP4 threshold for F8 (<0.05). The nucleotide has a PhastCons score of 0.4 and a PhyloP score of 0.71 (threshold <0.1), indicating that the nucleotide is weakly conserved. BP7 not met.

Curation History

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