

Variant: *NM_000132.3(F8):c.3864A>C (p.Ser1288=)*

Version: 1.0

CA343867 [↗](#)

41000 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: c93cdc62-e0c9-4b88-ac1b-eb3bed663994

Approved on: 2024/2/1

Published on: 2024/7/11

HGVS expressions

NM_000132.3:c.3864A>C

NM_000132.3(F8):c.3864A>C (p.Ser1288=)

NC_000023.11:g.154929926T>G

CM000685.2:g.154929926T>G

NC_000023.10:g.154158201T>G

CM000685.1:g.154158201T>G

NC_000023.9:g.153811395T>G

NG_011403.1:g.97798A>C

NG_011403.2:g.97798A>C

ENST00000360256.9:c.3864A>C

ENST00000360256.8:c.3864A>C

NM_000132.4:c.3864A>C

Benign

Met criteria codes **2**

BP4 **BA1**

Not Met criteria codes **1**

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.3864A>C (p.Ser1288=) variant is reported at an MAF of 0.3377 (6437/19059 alleles) in the South Asian population in gnomAD v2.1.,1 with 3958 hemizygotes and 424 homozygotes, meeting BA1 criteria of MAF > 0.000333. The synonymous variant is predicted to have no impact on splicing based on SpliceAI score of 0.0, meeting BP4. 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, BP4.

Met criteria codes

BP4			The Ser1288= synonymous variant is predicted to have no impact on splicing based on SpliceAI score of 0.0
BA1			The c.3864A>C (p.Ser1288=) variant is reported at an MAF of 0.3377 (6437/19059 alleles) in the South Asian population in gnomAD v2.1.1/gnomAD v3/ExAC/ESP, etc) with 3958 hemizygotes and 424 homozygotes, meeting BA1 criteria of MAF > 0.000333. Note, MAF in gnomAD v3.1.1 is higher, at 0.35 in the South Asian population with fewer hemi and homozygotes.

Not Met criteria codes

BP7			The Ser1288= synonymous variant is predicted to have no impact on splicing based on SpliceAI score of 0.0; however, the nucleotide is weakly-moderately conserved based on agreement between PhyloP (0.895) and PhastCons (0.465) scores.
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Curation History



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