

Variant: *NM_000132.4(F8):c.2696G>A (p.Ser899Asn)*

Version: 1.0

CA10568296 [↗](#)

913257 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 6557ff4d-b1f8-4ce3-8cae-06e3b5b9b78c

Approved on: 2026/8/7

Published on: 2026/9/4

HGVS expressions

NM_000132.4:c.2696G>A

NM_000132.4(F8):c.2696G>A (p.Ser899Asn)

NC_000023.11:g.154931094C>T

CM000685.2:g.154931094C>T

NC_000023.10:g.154159369C>T

CM000685.1:g.154159369C>T

NC_000023.9:g.153812563C>T

NG_011403.1:g.96630G>A

NG_011403.2:g.96630G>A

ENST00000360256.9:c.2696G>A

ENST00000647125.1:c.*2362G>A

ENST00000360256.8:c.2696G>A

NM_000132.3:c.2696G>A

Likely Benign

Met criteria codes **2**

BS1

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 F8 Version 2.0*

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

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

Evidence submitted by expert panel

Coagulation Factor Deficiency VCEP

The c.2696G>A (p.Ser899Asn) is a missense variant is reported in gnomAD v4.1 with a Gprmax FAF = 0.00009278, meeting BS1 criteria of greater than or equal to 0.00333% (or 0.0000333). The variant has a REVEL score of 0.234 (<0.3) and a SpliceAI score of 0.0 meeting BP4. In summary, this variant meets criteria to be classified as likely benign. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F8 v2.0: BS1 and BP4.

Met criteria codes

BS1			The c.2696G>A (p.Ser899Asn) variant is reported in gnomAD v4.1 with a Gprmax FAF = 0.00009278 meeting BS1 criteria of greater than or equal to 0.00333% (or 0.0000333).
BP4			The c.2696G>A (p.Ser899Asn) missense variant has a REVEL score of 0.234 (<0.3) and a SpliceAI score of 0.0 (<0.05).

Curation History



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