

Variant: *NM_000132.4(F8):c.6305G>A (p.Gly2102Asp)*

Version: 1.0

CA414900171 [↗](#)

1163208 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 445032ae-201c-47eb-be4a-4f11a33e5338

Approved on: 2026/4/10

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HGVS expressions

NM_000132.4(F8):c.6305G>A (p.Gly2102Asp)

NM_000132.4:c.6305G>A

NC_000023.11:g.154896201C>T

CM000685.2:g.154896201C>T

NC_000023.10:g.154124476C>T

CM000685.1:g.154124476C>T

NC_000023.9:g.153777670C>T

NG_011403.1:g.131523G>A

NG_011403.2:g.131523G>A

ENST00000360256.9:c.6305G>A

ENST00000360256.8:c.6305G>A

NM_000132.3:c.6305G>A

Likely Pathogenic

Met criteria codes **4**

PP3

PS4_Moderate

PM2_Supporting

PP4_Moderate

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 2.0.0*

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

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

Evidence submitted by expert panel

Coagulation Factor Deficiency VCEP

The c.6305G>A variant in F8 is a missense variant predicted to cause substitution of aspartic acid for glycine at amino acid 2102. This variant is completely absent from gnomAD v4.1.1 meeting PM2_Supporting. The missense variant has a REVEL score of 0.877 (>0.6) meeting PP3. At least 3 probands from the literature and internal laboratory data meet F8 phenotype criteria for PS4_Moderate and PP4_Moderate (PMID: 23926300, 29296726 & 23913812). In summary, based on the evidence available at this time, this variant is classified as likely pathogenic for hemophilia A. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel v2.0.0: PP4_Moderate, PS4_Moderate, PP3, PM2_Supporting.

Met criteria codes

PP3			REVEL score is 0.877, which above the 0.6 cutoff. SpliceAI prediction for donor gain = 0.01 and 0 for others.
PS4_Moderate			Two probands with mild hemophilia with factor VIII activity levels at 20% meeting PS4_Moderate (PMID: 23913812 & 23926300).
PM2_Supporting			Variant is absent in gnomAD v4.1.1.
PP4_Moderate			Variant identified in proband with 3% factor VIII activity level in MLOF study.

Curation History

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