The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000132.4:c.1701del

Version: 1.0

CA915940891

2775448 (ClinVar)

Gene: F8 (HGNC:2157)
Condition: hemophilia A (MONDO:0010602)
Inheritance Mode: X-linked inheritance
UUID: 00613ee1-1167-4a78-a9ca-54333efc23bb
Approved on: 2024/2/2
Published on: 2024/7/11

HGVS expressions

NM_000132.4:c.1701del
NC_000023.11:g.154957009del
CM000685.2:g.154957009del
NC_000023.10:g.154185284del
CM000685.1:g.154185284del
NC_000023.9:g.153838478del
NG_011403.1:g.70716del
NG_011403.2:g.70716del
ENST00000360256.9:c.1701del
ENST00000647125.1:c.*1577del
ENST00000360256.8:c.1701del
NM_000132.3:c.1701del

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PS4_Supporting

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.1701del (p.Ile567MetfsTer12) variant is a frameshift variant that is predicted to introduce a premature stop codon in exon 11/26 and expected to result in nonsense-mediated mRNA decay, which meets PVS1 criteria. This variant is completely absent from males in gnomAD v2.1.1 and gnomAD v3 meeting PM2_Supporting criteria. There is one patient from a research cohort with hemophilia A who carries the Ile567MetfsTer12 variant (internal VCEP contributor). In summary, this variant meets criteria to be classified as pathogenic. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F8/F9: PVS1, PS4_Supporting, PM2_Supporting.

Met criteria codes

PVS1	✔	The c.1701del (p.Ile567MetfsTer12) variant is a frameshift variant that is predicted to introduce a premature stop codon in exon 11/26 and expected to result in nonsense-mediated mRNA decay.
PM2_Supporting	✔	The c.1701del (Ile567MetfsTer12) variant is completely absent from males in gnomAD v2.1.1 and gnomAD v3.
PS4_Supporting	✔	There is one patient in a research cohort with hemophilia A with variant.

Curation History

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