

Variant: *NM_000132.3(F8):c.1804C>T (p.Arg602Ter)*

Version: 1.0

CA255124 [↗](#)

10232 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 8d0f7cbb-2982-46ea-b526-c6afd4ec4be3

Approved on: 2024/2/2

Published on: 2024/7/11

HGVS expressions

NM_000132.3:c.1804C>T

NM_000132.3(F8):c.1804C>T (p.Arg602Ter)

NC_000023.11:g.154953991G>A

CM000685.2:g.154953991G>A

NC_000023.10:g.154182266G>A

CM000685.1:g.154182266G>A

NC_000023.9:g.153835460G>A

NG_011403.1:g.73733C>T

NG_011403.2:g.73733C>T

ENST00000360256.9:c.1804C>T

ENST00000647125.1:c.*1680C>T

ENST00000360256.8:c.1804C>T

NM_000132.4:c.1804C>T

Pathogenic

Met criteria codes **4**

PP4_Moderate

PVS1

PS4

PM2_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.1804C>T (p.Arg602Ter) variant is a nonsense variant that is predicted to introduce a premature stop codon in exon 12 and expected to result in nonsense-mediated mRNA decay. This variant is absent from males in gnomAD v2.1.1 and v3.1.1, which meets criteria for PM2_Supporting. More than 25 patients with moderate-severe hemophilia A are reported in the literature and internal laboratory data, meeting criteria for PS4_Very Strong and PP4_Moderate (PMID: 29296726, 18387975, 18691168, 16769589, 8639447, 20331761). 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_Very Strong, PP4_Moderate, PM2_Supporting.

Met criteria codes

PP4_Moderate			Male with severe hemophilia A who had full F8 and F9 gene sequencing and deletion/duplication analysis through MLOF study.
PVS1			The c.1804C>T (p.Arg602Ter) variant is a nonsense variant that is predicted to introduce a premature stop codon in exon 12 and expected to result in nonsense-mediated mRNA decay. Meets PVS1 criteria
PS4			Numerous patients with moderate-severe hemophilia A are reported in the literature and internal laboratory data, meeting criteria for PS4_Very Strong (>8 probands).
PM2_Supporting			The variant is absent from males in gnomAD v2.1.1 and v3.1.1

Curation History



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Showing 1 to 1 of 1 rows

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