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Variant: NM_019863.3:c.610A>T

Version: 1.0

CA414896830

Gene: F8 (HGNC:2157)
Condition: hemophilia A (MONDO:0010602)
Inheritance Mode: X-linked inheritance
UUID: 410722b3-4d85-48da-bab9-4d8a259e84d6
Approved on: 2024/6/19
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HGVS expressions

NM_019863.3:c.610A>T
NC_000023.11:g.154837638T>A
CM000685.2:g.154837638T>A
NC_000023.10:g.154065913T>A
CM000685.1:g.154065913T>A
NC_000023.9:g.153719107T>A
NG_011403.1:g.190086A>T
NG_033065.1:g.2025A>T
NG_011403.2:g.190086A>T
ENST00000360256.9:c.7015A>T
ENST00000644698.1:c.748A>T
ENST00000330287.10:c.610A>T
ENST00000360256.8:c.7015A>T
NM_000132.3:c.7015A>T
NM_019863.2:c.610A>T
NM_000132.4:c.7015A>T

Likely Pathogenic

Met criteria codes 3

PS4 PM2_Supporting PP3

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.7015A>T (Arg2339Trp) variant is absent from males in population databases (gnomAD v2.1.1/gnomAD v3). This variant has been reported in at least four probands with severe hemophilia A (>4 probands & 0 gnomAD alleles) meeting phenotypic criteria for F8 (PMID:

30913330, 20331761, 18691168, 18217193). This missense variant has a REVEL score of 0.878 (>0.6). In summary, this variant meets criteria to be classified as likely pathogenic. ACMG/AMP criteria applied, as specified by the Coagulation Factor Deficiency Variant Curation Expert Panel for F8: PS4, PP3, PM2_Supporting.

Met criteria codes

PS4	✔	4 individuals with severe hemophilia A are reported in the literature, meeting criteria for PS4
PM2_Supporting	✔	The c.7015A>T (Arg2339Trp) variant is absent from gnomAD v2.1.1 and v3.1.2
PP3	✔	The Arg2339Trp missense variant meets PP3 with a REVEL score of 0.878 (threshold >0.6)

Curation History

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