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_000133.4(F9):c.520+13A>G

Version: 1.0

CA915952347

627180 (ClinVar)

Gene: F9 (HGNC:2158)
Condition: hemophilia B (MONDO:0010604)
Inheritance Mode: X-linked inheritance
UUID: 92be25f1-a7ef-4229-b827-147574de1ffb
Approved on: 2024/10/11
Published on: 2024/10/11

HGVS expressions

NM_000133.4:c.520+13A>G
NM_000133.4(F9):c.520+13A>G
NC_000023.11:g.139548504A>G
CM000685.2:g.139548504A>G
NC_000023.10:g.138630663A>G
CM000685.1:g.138630663A>G
NC_000023.9:g.138458329A>G
NG_007994.1:g.22769A>G
ENST00000218099.7:c.520+13A>G
ENST00000643157.1:n.1187+13A>G
ENST00000218099.6:c.520+13A>G
ENST00000394090.2:c.406+13A>G
NM_000133.3:c.520+13A>G
NM_001313913.1:c.406+13A>G
NM_001313913.2:c.406+13A>G

Pathogenic

Met criteria codes 3

PS4_Very Strong PP3 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 F9 Version 1.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Coagulation Factor Deficiency VCEP

The c.520+13A>G in F9 is an intronic variant which located in intron 5. This variant has been reported in 38 probands with abnormal factor IX activity levels meeting the phenotypic criteria for PS4_Very Strong (threshold >=8 probands); PMID: 14675097, PMID: 32155688, PMID: 27213901, PMID: 15921378, PMID: 10090477 and PMID: 2198809. The computational predictor Splice AI gives a delta score of 0.92 for

donor gain (threshold >=0.5), evidence that correlates with impact to F9 function (PP3_supporting).This variant is absent from gnomAD v2.1.1 (PM2_Supporting). In summary, this variant meets the criteria to be classified as Pathogenic for hemophilia B based on the ACMG/AMP criteria applied, as specified by the ClinGen Coagulation Factor Deficiency VCEP for F9: PS4_Very Strong, PP3, PM2_Supporting (version 1.0.0, released 10/5/2023).

Met criteria codes

PS4_Very Strong	✔	This variant has been reported in 38 probands with abnormal factor IX activity levels meeting the phenotypic criteria for PS4_Very Strong (threshold >=8 probands); PMID: 14675097, PMID: 32155688, PMID: 27213901, PMID: 15921378, PMID: 10090477 and PMID: 2198809.
PP3	✔	The computational predictor Splice AI gives a delta score of 0.92 for donor gain (threshold >=0.5), evidence that correlates with impact to F9 function (PP3_supporting).
PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History

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