

Variant: *NM_001754.5(RUNX1):c.99T>C (p.Asp33=)*

Version: 1.0

CA512319003 [↗](#)

436612 (ClinVar) [↗](#)

Gene: RUNX1 ([HGNC:861](#))

Condition: hereditary thrombocytopenia and hematological cancer predisposition syndrome associated with RUNX1 ([MONDO:0100083](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 1eb50d20-5255-4396-9165-7e7d0c1afaca

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HGVS expressions

NM_001754.5(RUNX1):c.99T>C (p.Asp33=)

NM_001754.5:c.99T>C

NC_000021.9:g.34887095A>G

CM000683.2:g.34887095A>G

NC_000021.8:g.36259392A>G

CM000683.1:g.36259392A>G

NC_000021.7:g.35181262A>G

NG_011402.2:g.1102617T>C

ENST00000675419.1:c.99T>C

ENST00000300305.7:c.99T>C

ENST00000344691.8:c.18T>C

ENST00000358356.9:c.18T>C

ENST00000399237.6:c.63T>C

ENST00000399240.5:c.18T>C

ENST00000437180.5:c.99T>C

ENST00000455571.5:c.60T>C

ENST00000475045.6:c.99T>C

ENST00000482318.5:c.59-6382T>C

NM_001001890.2:c.18T>C

NM_001122607.1:c.18T>C

NM_001754.4:c.99T>C

NM_001001890.3:c.18T>C

NM_001122607.2:c.18T>C

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **25**

BS2 PVS1 BS1 BS4 BS3
BP5 BP7 BP4 BP3 BP1 BP2
PS1 PS2 PS3 PS4 BA1 PP1
PP2 PP3 PP4 PM6 PM1
PM3 PM5 PM4

Evidence Links **0**

Expert Panel

Myeloid Malignancy VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Myeloid Malignancy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2

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

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

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.99T>C (p.Asp33=) is a synonymous variant which is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). This variant has a SpliceAI predicted acceptor loss of 0.32, which indicates it may impact splicing (BP4/BP7 not applied). In summary, the clinical significance of this variant is uncertain. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: PM2_supporting.

Met criteria codes

PM2_Supporting			absent from gnomAD V2 and V3 This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2).
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Not Met criteria codes

BS2			nil data
PVS1			not applicable
BS1			absent from gnomAD V2 and V3
BS4			nil data
BS3			no data
BP5			not applicable
BP7			PhyloP score: 3.194, SpliceAI Acceptor Loss 0.32 Δ score
BP4			SpliceAI: Δ type Δ score pre-mRNA position Acceptor Loss 0.32 1 bp Donor Loss 0.09 196 bp Acceptor Gain 0.02 1305 bp Donor Gain 0.00
BP3			not applicable
BP1			not applicable
BP2			not applicable
PS1			not applicable

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