

Variant: NM_001754.5(RUNX1):c.285C>T (p.Pro95=)

Version: 1.0

CA512318858 [↗](#)

3791503 (ClinVar) [↗](#)

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

Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome ([MONDO:0011071](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 8013219d-196e-4576-b2cf-b2c10e0060d3

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HGVS expressions

NM_001754.5(RUNX1):c.285C>T (p.Pro95=)

NM_001754.5:c.285C>T

NC_000021.9:g.34886909G>A

CM000683.2:g.34886909G>A

NC_000021.8:g.36259206G>A

CM000683.1:g.36259206G>A

NC_000021.7:g.35181076G>A

NG_011402.2:g.1102803C>T

ENST00000675419.1:c.285C>T

ENST00000300305.7:c.285C>T

ENST00000344691.8:c.204C>T

ENST00000358356.9:c.204C>T

ENST00000399237.6:c.249C>T

ENST00000399240.5:c.204C>T

ENST00000437180.5:c.285C>T

ENST00000455571.5:c.246C>T

ENST00000482318.5:c.59-6196C>T

NM_001001890.2:c.204C>T

NM_001122607.1:c.204C>T

NM_001754.4:c.285C>T

NM_001001890.3:c.204C>T

NM_001122607.2:c.204C>T

Likely Benign

Met criteria codes **3**

PM2_Supporting BP4 BP7

Not Met criteria codes **23**

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

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

[↗](#) PDF

[↗](#) Criteria Specification Approval History

[↗](#) Criteria Specifications for this VCEP

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.285C>T (p.Pro95=) is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.01) (BP4). This variant has a SpliceAI score ≤ 0.20 (0.01) and evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (1.27) (BP7). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting). In summary, this variant meets criteria to be classified as likely benign. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BP4, BP7, PM2_supporting.

Met criteria codes

PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).
BP4			This synonymous variant has a SpliceAI score ≤ 0.20 (0.01) (BP4).
BP7			This variant has a SpliceAI score ≤ 0.20 (0.01) and evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (1.27) (BP7).

Not Met criteria codes

PS1			This variant is not a missense variant.
PS2			De novo data for this variant has not been reported in literature.
PS3			In vitro or in vivo functional data has not been reported for this variant in the literature.
PS4			Proband data for this variant has not been reported in literature.
BA1			This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
PP1			Segregation data for this variant has not been reported in literature.
PP2			This rule is not applicable for MM-VCEP.
PP3			This synonymous or intronic variant does not have a REVEL score ≥ 0.88 or a SpliceAI score ≥ 0.38 .
PP4			This rule is not applicable for MM-VCEP.
PM1			This variant is not a missense variant.

PM3		✖	This rule is not applicable for MM-VCEP.
PM5		✖	This variant is not a missense variant.
PM4		✖	This variant is not an in-frame deletion/insertion.
PM6		✖	De novo data for this variant has not been reported in literature.
BS2		✖	This rule is not applicable for MM-VCEP.
BS1		✖	This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset.
BS4		✖	Segregation data for this variant has not been reported in literature.
BS3		✖	In vitro or in vivo functional data has not been reported for this variant in the literature.
BP3		✖	This rule is not applicable for MM-VCEP.
BP1		✖	This rule is not applicable for MM-VCEP.
BP2		✖	This variant has not been observed in trans with a pathogenic variant for a fully penetrant dominant gene/disorder or observed in cis with a pathogenic variant in any inheritance pattern.
PVS1		✖	This variant is not a null variant.
BP5		✖	This rule is not applicable for MM-VCEP.

Curation History 		
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