

Variant: NM_001754.5(RUNX1):c.291C>T (p.Phe97=)

Version: 1.0

CA512318853 [↗](#)

1088304 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 1d2ff086-abdf-4f5e-8985-3cebc995a9d7

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HGVS expressions

NM_001754.5(RUNX1):c.291C>T (p.Phe97=)

NM_001754.5:c.291C>T

NC_000021.9:g.34886903G>A

CM000683.2:g.34886903G>A

NC_000021.8:g.36259200G>A

CM000683.1:g.36259200G>A

NC_000021.7:g.35181070G>A

NG_011402.2:g.1102809C>T

ENST00000675419.1:c.291C>T

ENST00000300305.7:c.291C>T

ENST00000344691.8:c.210C>T

ENST00000358356.9:c.210C>T

ENST00000399237.6:c.255C>T

ENST00000399240.5:c.210C>T

ENST00000437180.5:c.291C>T

ENST00000455571.5:c.252C>T

ENST00000482318.5:c.59-6190C>T

NM_001001890.2:c.210C>T

NM_001122607.1:c.210C>T

NM_001754.4:c.291C>T

NM_001001890.3:c.210C>T

NM_001122607.2:c.210C>T

Likely Benign

Met criteria codes **2**

BP4 BP7

Not Met criteria codes **24**

PM1 PM3 PM5 PM4 PM6
PM2 BS2 BS1 BS4 BS3
PVS1 PS1 PS2 PS3 PS4
BP3 BP1 BP2 BP5 BA1 PP1
PP2 PP3 PP4

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

The NM_001754.5(RUNX1):c.291C>T (p.Phe97=) variant is reported at a MAF of 0.00005 (0.005%, 1/21626 alleles) in the non-Finnish European subpopulation of the gnomAD v2.1.1 cohort, and does not meet thresholds for BA1, BS1, or PM2. The variant has not been reported in patients with familial platelet disorder with predisposition to hematologic malignancies in the literature, to the best of our knowledge. REVEL score is not calculable for a synonymous variant; SpiceAI predicts: Acceptor loss 0.02, Donor loss 0.00, Acceptor gain 0.00, Donor gain 0.00. Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 0.74248< 2.0) In summary, the clinical significance of this variant is likely benign. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BP4, BP7.

Met criteria codes

BP4			REVEL score is not calculable for a synonymous variant; SpiceAI predicts: Acceptor loss 0.02, Donor loss 0.00, Acceptor gain 0.00, Donor gain 0.00. (BP4)
BP7			Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 0.74248< 2.0) (BP7).

Not Met criteria codes

PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			This rule is not applicable for MM-VCEP
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			This variant is present at an allele frequency of 0.00005 (0.005%, 1/21626 alleles) in gnomAD version 2. This does not meet criteria for BA1, BS1, or PM2.
BS2			This rule is not applicable for MM-VCEP
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS3			No literature found for this variant.
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			No literature available for this variant
BP3			This rule is not applicable for MM-VCEP
BP1			This rule is not applicable for MM-VCEP
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			This rule is not applicable for MM-VCEP
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			This rule is not applicable for MM-VCEP
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			This rule is not applicable for MM-VCEP

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