

Variant: *NM_001754.5(RUNX1):c.990C>T (p.Phe330=)*

Version: 2.0

CA512341388 [↗](#)

1587386 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9583288a-843b-4639-a2eb-226cf1bacb3c

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HGVS expressions

NM_001754.5(RUNX1):c.990C>T (p.Phe330=)

NM_001754.5:c.990C>T

NC_000021.9:g.34792588G>A

CM000683.2:g.34792588G>A

NC_000021.8:g.36164885G>A

CM000683.1:g.36164885G>A

NC_000021.7:g.35086755G>A

NG_011402.2:g.1197124C>T

ENST00000675419.1:c.990C>T

ENST00000300305.7:c.990C>T

ENST00000344691.8:c.909C>T

ENST00000399240.5:c.717C>T

ENST00000437180.5:c.990C>T

ENST00000482318.5:c.*580C>T

NM_001001890.2:c.909C>T

NM_001754.4:c.990C>T

NM_001001890.3:c.909C>T

Likely Benign

Met criteria codes **3**

BP4

BP7

PM2_Supporting

Not Met criteria codes **23**

PM1

PM3

PM5

PM4

PM6

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 for RUNX1 Version 3.1.0

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

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

Evidence submitted by expert panel

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.990C>T (p.Phe330=) is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.0) (BP4, 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

BP4			This synonymous variant has a SpliceAI score ≤ 0.20 (0).
BP7			This synonymous variant has a SpliceAI score ≤ 0.20 (0)
PM2_Supporting			This variant has a MAF ≤ 0.00005 in gnomAD v4.0 in all subpopulations with at least 20X coverage for RUNX1 (PM2_supporting).

Not Met criteria codes

PM1			This variant does not affect any of the following amino acid residues, nor is it located within the RHD: R107, K110, A134, R162, R166, S167, R169, G170, K194, T196, D198, R201, R204 NOR is it within residues 89-204
PM3			This rule is not applicable for the MM-VCEP
PM5			This is not a missense variant, and there has not yet been a different missense change determined to be pathogenic at this amino acid residue.
PM4			This variant is not an inframe indel
PM6			This variant has not been reported in probands in the literature.
BS2			This rule is not applicable for the MM-VCEP
BS1			Highest MAF of 0.00004(0.004%) in South Asian population in gnomAD v4.0.0 is less than 0.00015 (0.015%)
BS4			This variant has not been reported in affected family members.
BS3			This variant has not been featured in in vitro or in vivo functional studies that show no damaging effect on protein function or splicing.
PVS1			This variant is not a null variant, as it does not impact any of the following processes or sites: Nonsense, frameshift, canonical +/- 1 or 2 splice sites, initiation codon, single exon deletion, multiple exon deletions
PS1			This is not a missense variant, and there has not yet been an amino acid change determined to be pathogenic at this amino acid residue.

PS2		✖	This variant has not been found to co-segregate with the disease in the literature.
PS3		✖	This variant has not been featured in in vitro or in vivo functional studies showing a damaging effect on the gene or gene product.
PS4		✖	This variant has not been reported in probands.
BP3		✖	This rule is not applicable for the MM-VCEP
BP1		✖	This rule is not applicable for the MM-VCEP
BP2		✖	This variant has not been detected in a homozygous state in an individual or in a population database (gnomAD).
BP5		✖	This rule is not applicable for the MM-VCEP
BA1		✖	Highest MAF is less than 0.0015 (0.15%) at 0.00004(0.004%, 4 allele count/ 88648 allele number) in the South Asian subpopulation of gnomAD v4.0.0 cohort
PP1		✖	This variant was not found to co-segregate with disease in multiple affected family members.
PP2		✖	This rule is not applicable for the MM-VCEP
PP3		✖	This synonymous variant does not have a SpliceAI score of ≥ 0.38 (0).
PP4		✖	This rule is not applicable for the MM-VCEP

Curation History 		
		
Showing 1 to 2 of 2 rows		

