

Variant: NM_001754.5(RUNX1):c.40C>A (p.Pro14Thr)

Version: 1.0

CA410208229 [↗](#)

1483319 (ClinVar) [↗](#)

Gene: RUNX1 (HGNC:861)

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

Inheritance Mode: Autosomal dominant inheritance

UID: 7843eb7b-5c25-4e34-bfc2-66c829baab1c

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HGVS expressions

NM_001754.5:c.40C>A

NM_001754.5(RUNX1):c.40C>A (p.Pro14Thr)

NC_000021.9:g.35048860G>T

CM000683.2:g.35048860G>T

NC_000021.8:g.36421157G>T

CM000683.1:g.36421157G>T

NC_000021.7:g.35343027G>T

NG_011402.2:g.940852C>A

ENST00000675419.1:c.40C>A

ENST00000300305.7:c.40C>A

ENST00000416754.1:c.40C>A

ENST00000437180.5:c.40C>A

ENST00000455571.5:c.40C>A

ENST00000475045.6:c.40C>A

ENST00000482318.5:c.40C>A

NM_001754.4:c.40C>A

Uncertain Significance

Met criteria codes **2**

PM2_Supporting BP4

Not Met criteria codes **23**

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

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

Evidence submitted by expert panel
























Myeloid Malignancy VCEP
















NM_001754.5(RUNX1):c.40C>A (p.Pro14Thr) is a missense variant which has a REVEL score of < 0.50 (0.295) (BP4). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting). 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: BP4, 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 missense variant has a REVEL score of 0.295, which is < 0.50 (BP4)

Not Met criteria codes

PS1			There has not yet been a different missense change determined to be pathogenic at this amino acid residue.
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 \geq 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 missense variant does not have a REVEL score of \geq 0.88.
PP4			This rule is not applicable for MM-VCEP.
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 OR within residues 89-204.
PM3			This rule is not applicable for MM-VCEP.
PM5			There has not yet been a different missense change determined to be pathogenic at this amino acid residue.
PM4			This variant is not an in-frame deletion/insertion.
PM6			De novo data for this variant has not been reported in literature.

PVS1			This variant is not a null variant
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.
BP5			This rule is not applicable for MM-VCEP.
BP7			This variant is not a synonymous or intronic variant.
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.

Curation History [↗](#)

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