

Variant: *NM_001754.5(RUNX1):c.301G>C (p.Val101Leu)*

Version: 1.0

[CA410203589](#)

[2864169 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: afbc5661-b1a2-4bbc-91fa-8c38702e6d99

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HGVS expressions

NM_001754.5:c.301G>C

NM_001754.5(RUNX1):c.301G>C (p.Val101Leu)

NC_000021.9:g.34886893C>G

CM000683.2:g.34886893C>G

NC_000021.8:g.36259190C>G

CM000683.1:g.36259190C>G

NC_000021.7:g.35181060C>G

NG_011402.2:g.1102819G>C

ENST00000675419.1:c.301G>C

ENST00000300305.7:c.301G>C

ENST00000344691.8:c.220G>C

ENST00000358356.9:c.220G>C

ENST00000399237.6:c.265G>C

ENST00000399240.5:c.220G>C

ENST00000437180.5:c.301G>C

ENST00000455571.5:c.262G>C

ENST00000482318.5:c.59-6180G>C

NM_001001890.2:c.220G>C

NM_001122607.1:c.220G>C

NM_001754.4:c.301G>C

NM_001001890.3:c.220G>C

NM_001122607.2:c.220G>C

Uncertain Significance

Met criteria codes **3**

PM2_Supporting PP3

PM1_Supporting

Not Met criteria codes **17**

PM6 PM5 PM4 BA1 BS1

BS4 BS3 BP7 BP4 BP3 BP1

PS1 PS2 PS3 PP1 PP2 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




NM_001754.5(RUNX1):c.301G>C (p.Val101Leu) is a missense variant within the Runt Homology domain (AA 89-204), but does not affect an established hotspot variant (PM1_supporting). This missense variant has a REVEL score ≥ 0.88 (0.921) (PP3). 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: PM1_supporting, PP3, 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).
PP3			This missense variant has a REVEL score ≥ 0.88 (0.921) (PP3).
PM1_Supporting			Variant is within the Runt Homology domain (AA 89-204), but does not affect an established hotspot variant (PM1_supporting).

Not Met criteria codes

PM6			De novo data for this variant has not been reported in literature.
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.
BA1			This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
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.
BP7			This variant is not a synonymous or intronic variant.
BP4			This missense variant does not have a REVEL score < 0.50 .
BP3			This rule is not applicable for MM-VCEP.

BP1		✘	This rule is not applicable for MM-VCEP.
PS1		✘	There has not yet been a 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.
PP1		✘	Segregation data for this variant has not been reported in literature.
PP2		✘	This rule is not applicable for MM-VCEP.
PP4		✘	This rule is not applicable for MM-VCEP.

Curation History [↗](#)

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