

Variant: NM_001754.5(RUNX1):c.155T>A (p.Met52Lys)

Version: 2.1

CA10014581 [↗](#)

239044 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4ceacdfa-56c1-4933-a48a-f8c97e4f92e9

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HGVS expressions

NM_001754.5:c.155T>A

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NC_000021.9:g.34887039A>T

CM000683.2:g.34887039A>T

NC_000021.8:g.36259336A>T

CM000683.1:g.36259336A>T

NC_000021.7:g.35181206A>T

NG_011402.2:g.1102673T>A

ENST00000675419.1:c.155T>A

ENST00000300305.7:c.155T>A

ENST00000344691.8:c.74T>A

ENST00000358356.9:c.74T>A

ENST00000399237.6:c.119T>A

ENST00000399240.5:c.74T>A

ENST00000437180.5:c.155T>A

ENST00000455571.5:c.116T>A

ENST00000482318.5:c.59-6326T>A

NM_001001890.2:c.74T>A

NM_001122607.1:c.74T>A

NM_001754.4:c.155T>A

NM_001001890.3:c.74T>A

NM_001122607.2:c.74T>A

Likely Benign

Met criteria codes **2**

BS1 BP2

Not Met criteria codes **24**

PS1 PS2 PS3 PS4 BA1
PM6 PM2 PM1 PM3 PM5
PM4 PP1 PP2 PP3 PP4
PVS1 BS2 BS4 BS3 BP4
BP3 BP1 BP5 BP7

Evidence Links **1**

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_001754.4(RUNX1):c.155T>A (p.Met52Lys) is a missense variant which has a MAF of 0.0007121 (0.071%, 840/1,179,678 alleles, including 1 homozygote) in the European (non-Finnish) subpopulation of the gnomAD v4.1.0 cohort, meeting the threshold for BS1 and BP2. 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: BS1, BP2.

Met criteria codes

BS1			MAF of 0.0007121 (0.071%, 840/1179678, 840 alleles, 1 homozygote) in the European (non-Finnish) subpopulation of the gnomAD v4.1.0 cohort is between 0.00015 (0.015%) and 0.0015 (0.15%) (BS1).
BP2			This variant is detected in a homozygous state in an individual or in a population database (1 homozygote) in the European (non-Finnish) subpopulation of the gnomAD v4.1.0 cohort). (BP2)

Not Met criteria codes

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.
PS4			PS4 cannot be applied because the variant presents more than 2 times in gnomAD Pt 9 with AML, germline variant on cultured skin fibroblasts PubMed:29365323 
BA1			This variant does not have a MAF \geq 0.0015 (0.15%) in any general continental population dataset.
PM6			This variant does not have two or more assumed de novo occurrences in the literature (one occurrence confirmed as germline; for other occurrences the germline origin was not confirmed).
PM2			This variant is present in at least one population database.
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.
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 ≥ 0.88 .
PP4			This rule is not applicable for MM-VCEP
PVS1			This variant is not a null variant.
BS2			This rule is not applicable for MM-VCEP
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.
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
BP5			This rule is not applicable for MM-VCEP
BP7			This variant is not a synonymous or intronic variant.

Curation History [↗](#)

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