

Variant: *NM_001754.5(RUNX1):c.1099G>C (p.Gly367Arg)*

Version: 1.0

[CA410148133](#)

[2126320 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 7279e349-fac0-40be-9caf-43641b65ce38

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HGVS expressions

NM_001754.5:c.1099G>C

NM_001754.5(RUNX1):c.1099G>C (p.Gly367Arg)

NC_000021.9:g.34792479C>G

CM000683.2:g.34792479C>G

NC_000021.8:g.36164776C>G

CM000683.1:g.36164776C>G

NC_000021.7:g.35086646C>G

NG_011402.2:g.1197233G>C

ENST00000675419.1:c.1099G>C

ENST00000300305.7:c.1099G>C

ENST00000344691.8:c.1018G>C

ENST00000399240.5:c.826G>C

ENST00000437180.5:c.1099G>C

ENST00000482318.5:c.*689G>C

NM_001001890.2:c.1018G>C

NM_001754.4:c.1099G>C

NM_001001890.3:c.1018G>C

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **18**

BS1 BS4 BS3 BP7 BP4 BP2

PM6 PM1 PM5 PM4 BA1

PS1 PS2 PS3 PS4 PP1 PP3

PVS1

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.1099G>C (p.Gly367Arg) is a missense variant which is completely absent from all population databases (gnomAD v2.1.1 and v3.1.2) with at least 20x coverage for RUNX1 (PM2_supporting). It has a REVEL score of 0.707, which does not allow applying either PP3 (>0.88) or BP4 (<0.50). Additionally, there is no previous report of this variant. 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: PM2_supporting.



Met criteria codes



PM2_Supporting   This variant is completely absent from all population databases (gnomAD v2.1.1 and v3.1.2) with at least 20x coverage for RUNX1 (PM2_supporting).



Not Met criteria codes



BS1   This rule cannot be applied since the variant is completely absent from all population databases.



BS4   This rule cannot be applied since, to our knowledge, this variant has not been reported in any study and we have no information about segregation.


BS3   To our knowledge, no in vitro or in vivo functional studies are available for this variant.



BP7   This rule cannot be applied since this is a missense variant.



BP4   This rule cannot be applied since this missense variant has not a REVEL score <0.50 (0.707).



BP2   This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.



PM6   This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.


PM1  This rule cannot be applied since this missense variant affects amino acid residue 367, which is out of RHD or residues 89-204.











PM5   This rule cannot be applied because, to our knowledge, this missense variant does not impact an amino acid residue where a different pathogenic missense change has previously been observed.

PM4   This rule cannot be applied since this is a missense variant.

BA1   This rule cannot be applied since the variant is completely absent from all population databases.

PS1   This rule cannot be applied because, to our knowledge, this missense variant does not generate the same amino acid change as a previously identified pathogenic variant regardless of nucleotide change.

PS2  This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.

PS3			To our knowledge, no in vitro or in vivo functional studies are available for this variant.
PS4			This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
PP1			This rule cannot be applied since, to our knowledge, this variant has not been reported in any study and we have no information about its cosegregation with the disease.
PP3			This rule cannot be applied since this missense variant has not a REVEL score >0.88 (0.707).
PVS1			This rule cannot be applied since this is a missense variant.

Curation History [↗](#)

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