

Variant: *NM_001754.5(RUNX1):c.958C>A (p.Arg320=)*

Version: 2.0

CA512232176 [↗](#)

2814422 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 6964b53c-a237-41b8-b1d3-0366655f3264

Approved on: 2026/4/29

Published on: 2026/4/29

HGVS expressions

NM_001754.5:c.958C>A

NM_001754.5(RUNX1):c.958C>A (p.Arg320=)

NC_000021.9:g.34799310G>T

CM000683.2:g.34799310G>T

NC_000021.8:g.36171607G>T

CM000683.1:g.36171607G>T

NC_000021.7:g.35093477G>T

NG_011402.2:g.1190402C>A

ENST00000675419.1:c.958C>A

ENST00000300305.7:c.958C>A

ENST00000344691.8:c.877C>A

ENST00000399240.5:c.685C>A

ENST00000437180.5:c.958C>A

ENST00000482318.5:c.*548C>A

NM_001001890.2:c.877C>A

NM_001754.4:c.958C>A

NM_001001890.3:c.877C>A

Likely Benign

Met criteria codes 3

BP4 BP7 PM2_Supporting

Not Met criteria codes 23

BP3 BP1 BP2 BP5 BS2 BS1
BS4 BS3 PP1 PP2 PP3 PP4
PM1 PM3 PM5 PM4 PS1
PS2 PS3 PS4 BA1 PM6
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 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.958C>A (p.Arg320=) is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.011) (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 (0.11) (BP4).
BP7			This synonymous variant has a SpliceAI score ≤ 0.20 (0.11)
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).

Not Met criteria codes

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.
BP5			This rule is not applicable for MM-VCEP.
BS2			This rule is not applicable for MM-VCEP.
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.
PP1			Segregation data for this variant has not been reported in literature.
PP2			This rule is not applicable for MM-VCEP.
PP3			This synonymous or intronic variant does not have a REVEL score ≥ 0.88 or a SpliceAI score ≥ 0.38 .
PP4			This rule is not applicable for MM-VCEP.

PM1		✖	This variant is not a missense variant.
PM3		✖	This rule is not applicable for MM-VCEP.
PM5		✖	This variant is not a missense variant.
PM4		✖	This variant is not an in-frame deletion/insertion.
PS1		✖	This variant is not a missense variant.
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.
PM6		✖	De novo data for this variant has not been reported in literature.
PVS1		✖	This variant is not a null variant.

Curation History 		
▼ ▼		
Showing 1 to 2 of 2 rows		

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.