

Variant: *NM\_001754.5(RUNX1):c.519C>T (p.Phe173=)*

Version: 2.0

CA512319131 [↗](#)

1528554 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** f308ef24-97f3-4e7d-a0a8-968bc05a42cc

**Approved on:** 2026/5/4

**Published on:** 2026/5/4

## HGVS expressions

**NM\_001754.5:c.519C>T**

**NM\_001754.5(RUNX1):c.519C>T (p.Phe173=)**

NC\_000021.9:g.34859568G>A

CM000683.2:g.34859568G>A

NC\_000021.8:g.36231865G>A

CM000683.1:g.36231865G>A

NC\_000021.7:g.35153735G>A

NG\_011402.2:g.1130144C>T

ENST00000675419.1:c.519C>T

ENST00000300305.7:c.519C>T

ENST00000344691.8:c.438C>T

ENST00000358356.9:c.438C>T

ENST00000399237.6:c.483C>T

ENST00000399240.5:c.438C>T

ENST00000437180.5:c.519C>T

ENST00000467577.1:n.11C>T

ENST00000482318.5:c.\*109C>T

NM\_001001890.2:c.438C>T

NM\_001122607.1:c.438C>T

NM\_001754.4:c.519C>T

NM\_001001890.3:c.438C>T

NM\_001122607.2:c.438C>T

Likely Benign

Met criteria codes **3**

BP4 BP7 PM2\_Supporting

Not Met criteria codes **22**

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

*Myeloid Malignancy VCEP*

NM\_001754.5(RUNX1):c.519C>T (p.Phe173=) is a synonymous variant which has a SpliceAI score  $\leq 0.20$  (0.00) (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 $\leq 0.20$ (0.0) (BP4).                                                  |
| BP7            |  |  | This synonymous variant has a SpliceAI score $\leq 0.20$ (0.0)                                                         |
| PM2_Supporting |  |  | This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting). |

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 |  |  | Proband data for this variant has not been reported in literature.                                                                                                                          |
| 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.                                                                                                                                                    |
| 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 $\geq 0.88$ or a SpliceAI score $\geq 0.38$ .                                                                               |

|             |                                                                                     |   |                                                                                                                  |
|-------------|-------------------------------------------------------------------------------------|---|------------------------------------------------------------------------------------------------------------------|
| <b>PP4</b>  |                                                                                     | ✖ | This rule is not applicable for MM-VCEP.                                                                         |
| <b>BA1</b>  |    | ✖ | This variant does not have a MAF $\geq$ 0.0015 (0.15%) in any general continental population dataset.            |
| <b>PM6</b>  |    | ✖ | De novo data for this variant has not been reported in literature.                                               |
| <b>PM1</b>  |                                                                                     | ✖ | This variant is not a missense variant.                                                                          |
| <b>PM3</b>  |                                                                                     | ✖ | This rule is not applicable for MM-VCEP.                                                                         |
| <b>PM5</b>  |    | ✖ | There has not yet been a different missense change determined to be pathogenic at this amino acid residue.       |
| <b>PM4</b>  |    | ✖ | This variant is not an in-frame deletion/insertion.                                                              |
| <b>BS1</b>  |    | ✖ | This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset. |
| <b>BS4</b>  |    | ✖ | Segregation data for this variant has not been reported in literature.                                           |
| <b>BS3</b>  |   | ✖ | In vitro or in vivo functional data has not been reported for this variant in the literature.                    |
| <b>PVS1</b> |  | ✖ | This variant is not a null variant.                                                                              |

Curation History 





Showing 1 to 2 of 2 rows

|  |
|--|
|  |
|  |

