

Variant: *NM\_001754.5(RUNX1):c.999G>T (p.Pro333=)*

Version: 1.0

[CA320251631](#)

[1628628 \(ClinVar\)](#)

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

**Condition:** hereditary thrombocytopenia and hematological cancer predisposition syndrome associated with RUNX1 ([MONDO:0100083](#))

**Inheritance Mode:** Autosomal dominant inheritance

**UID:** cf7937f9-436e-4311-8e81-832624ace4fc

**Approved on:** 2024-06-24

**Published on:** 2024-06-24

### *HGVS expressions*

**NM\_001754.5:c.999G>T**

NM\_001754.5(RUNX1):c.999G>T (p.Pro333=)

NC\_000021.9:g.34792579C>A

CM000683.2:g.34792579C>A

NC\_000021.8:g.36164876C>A

CM000683.1:g.36164876C>A

NC\_000021.7:g.35086746C>A

NG\_011402.2:g.1197133G>T

ENST00000675419.1:c.999G>T

ENST00000300305.7:c.999G>T

ENST00000344691.8:c.918G>T

ENST00000399240.5:c.726G>T

ENST00000437180.5:c.999G>T

ENST00000482318.5:c.\*589G>T

NM\_001001890.2:c.918G>T

NM\_001754.4:c.999G>T

NM\_001001890.3:c.918G>T

**Likely Benign**

**Met criteria codes** **3**

**PM2\_Supporting** **BP4** **BP7**

**Not Met criteria codes** **21**

**PS1** **PS2** **PS3** **PS4** **PM1**  
**PM3** **PM5** **PM4** **PM6** **BP3**  
**BP1** **BP2** **BP5** **BS2** **PVS1**  
**BS4** **BS3** **PP1** **PP2** **PP3** **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*

[Criteria Specification Approval History](#)







[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel




















**Myeloid Malignancy VCEP**
















This synonymous variant has a SpliceAI  $\Delta$  score of 0.00 (BP4). Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score -0.008 (< 2)) (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

<b>PM2_Supporting</b>			0 alleles gnomAD V2, 0 alleles gnomAD V3 This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2).
<b>BP4</b>			SpliceAI $\Delta$ score 0.00 PhyloP score: -0.093 BP4: This synonymous variant has a SpliceAI $\Delta$ score < 0.20 PhyloP score < 2
<b>BP7</b>			SpliceAI $\Delta$ score 0.00 PhyloP score: -0.093 BP7: Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score -0.093) (BP7).

#### Not Met criteria codes

<b>PS1</b>			not applicable
<b>PS2</b>			nil data
<b>PS3</b>			nil data
<b>PS4</b>			nil data
<b>PM1</b>			not applicable
<b>PM3</b>			not applicable
<b>PM5</b>			not applicable
<b>PM4</b>			not applicable
<b>PM6</b>			nil data
<b>BP3</b>			not applicable
<b>BP1</b>			not applicable
<b>BP2</b>			not applicable

<b>BP5</b>			not applicable
<b>BS2</b>			not applicable
<b>PVS1</b>			not applicable
<b>BS4</b>			nil data
<b>BS3</b>			nil data
<b>PP1</b>			nil data
<b>PP2</b>			not applicable
<b>PP3</b>			BP4 met
<b>PP4</b>			nil data

Curation History [↗](#)

Showing 1 to 1 of 1 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.