

Variant: *NM_001754.5(RUNX1):c.735C>G (p.Pro245=)*

Version: 2.0

CA512318600 [↗](#)

1124345 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 8651b813-3751-40f3-a05a-327304279dd3

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HGVS expressions

NM_001754.5(RUNX1):c.735C>G (p.Pro245=)

NM_001754.5:c.735C>G

NC_000021.9:g.34834480G>C

CM000683.2:g.34834480G>C

NC_000021.8:g.36206777G>C

CM000683.1:g.36206777G>C

NC_000021.7:g.35128647G>C

NG_011402.2:g.1155232C>G

ENST00000675419.1:c.735C>G

ENST00000300305.7:c.735C>G

ENST00000344691.8:c.654C>G

ENST00000358356.9:c.654C>G

ENST00000399237.6:c.699C>G

ENST00000399240.5:c.532+24994C>G

ENST00000437180.5:c.735C>G

ENST00000469087.1:n.271C>G

ENST00000482318.5:c.*325C>G

NM_001001890.2:c.654C>G

NM_001122607.1:c.654C>G

NM_001754.4:c.735C>G

NM_001001890.3:c.654C>G

NM_001122607.2:c.654C>G

Likely Benign

Met criteria codes **3**

BP7 BP4 PM2_Supporting

Not Met criteria codes **23**

BS2 PVS1 BS1 BS4 BS3
BP5 BP3 BP1 BP2 PS1 PS2
PS3 PS4 BA1 PP1 PP2 PP3
PP4 PM6 PM1 PM3 PM5
PM4

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.735C>G (p.Pro245=) is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.02) (BP4, BP7). This variant has a MAF ≤ 0.00005 in gnomAD v4 across all subpopulations 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

BP7			This a synonymous variant. SpliceAI was <0.20 (Donor Loss score of 0.02)
BP4			This a synonymous variant. SpliceAI was <0.20 (Donor Loss score of 0.02)
PM2_Supporting			This variant has a MAF ≤ 0.00005 in gnomAD v4.0 in all subpopulations with at least 20X coverage for RUNX1 (PM2_supporting).

Not Met criteria codes

BS2			This rule is not applicable for MM-VCEP.
PVS1			This is a synonymous variant.
BS1			This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset.
BS4			No segregation data available.
BS3			This is a synonymous variant with no functional studies reported.
BP5			This rule is not applicable for MM-VCEP.
BP3			This rule is not applicable for MM-VCEP.
BP1			This rule is not applicable for MM-VCEP.
BP2			Other pathogenic variant not known.
PS1			This is a synonymous variant.
PS2			No other de novo cases with phenotype reported.

PS3			This is a synonymous variant with no functional studies reported.
PS4			No published cases reported.
BA1			This variant does not have a MAF $\geq$ 0.0015 (0.15%) in any general continental population dataset.
PP1			No segregation data available.
PP2			This rule is not applicable for MM-VCEP.
PP3			This is a synonymous variant not located within the last 3 bases of an exon splice donor or splice acceptor site. SpliceAI score is <0.20.
PP4			This rule is not applicable for MM-VCEP.
PM6			No other de novo cases with phenotype reported.
PM1			This is a synonymous variant.
PM3			This rule is not applicable for MM-VCEP.
PM5			This is a synonymous variant.
PM4			This is a synonymous variant.

Curation History [↗](#)

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