

Variant: *NM_001754.5(RUNX1):c.600C>T (p.Pro200=)*

Version: 1.0

CA512319072 [↗](#)

1120421 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 64bae0ff-3c97-497e-9d10-b4b141c2d476

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HGVS expressions

NM_001754.5(RUNX1):c.600C>T (p.Pro200=)

NM_001754.5:c.600C>T

NC_000021.9:g.34859487G>A

CM000683.2:g.34859487G>A

NC_000021.8:g.36231784G>A

CM000683.1:g.36231784G>A

NC_000021.7:g.35153654G>A

NG_011402.2:g.1130225C>T

ENST00000675419.1:c.600C>T

ENST00000300305.7:c.600C>T

ENST00000344691.8:c.519C>T

ENST00000358356.9:c.519C>T

ENST00000399237.6:c.564C>T

ENST00000399240.5:c.519C>T

ENST00000437180.5:c.600C>T

ENST00000467577.1:n.92C>T

ENST00000482318.5:c.*190C>T

NM_001001890.2:c.519C>T

NM_001122607.1:c.519C>T

NM_001754.4:c.600C>T

NM_001001890.3:c.519C>T

NM_001122607.2:c.519C>T

Likely Benign

Met criteria codes **3**

BP4 BP7 PM2_Supporting

Not Met criteria codes **23**

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

Evidence Links **0**

Expert Panel

Myeloid Malignancy VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Myeloid Malignancy VCEP

This variant NM_001754.5(RUNX1):c.600C>T (p.Pro200=) is a synonymous variant. This is a synonymous variant therefore REVEL score cannot be applied but SpliceAI is ≤ 0.20 (0.00) (BP4). The PhyloP score is <2.0 (0.219) (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			Synonymous variant and SpliceAI is ≤ 0.20 (0.00)
BP7			PhyloP score is <2.0 (0.219)
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1

Not Met criteria codes

PM1			This variant affects one of the other residues (AA 89-204) within the RHD but not a missense variant
PM3			This rule is not applicable for MM-VCEP
PM5			Synonymous variant
PM4			Synonymous variant
PM6			No case studies found
BS2			This rule is not applicable for MM-VCEP
BS1			Meets PM2
BS4			No case studies found
BS3			No functional studies found
PVS1			Synonymous variant thus PVS1 cannot be applied
PS1			Amino Acid (proline) at location 200 has not been established as pathogenic

PS2		✖	No case studies found
PS3		✖	No functional studies found
PS4		✖	No case studies found
BP3		✖	This rule is not applicable for MM-VCEP
BP1		✖	This rule is not applicable for MM-VCEP
BP2		✖	No homozygotes reported on gnomAD
BP5		✖	This rule is not applicable for MM-VCEP
BA1		✖	Meets PM2
PP1		✖	No case studies found
PP2		✖	This rule is not applicable for MM-VCEP
PP3		✖	Synonymous variants and SpliceAI is not ≥ 0.38 (0.00)
PP4		✖	This rule is not applicable for MM-VCEP

Curation History 		
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