

Variant: NM_001754.5(RUNX1):c.1158C>T (p.Pro386=)

Version: 1.0

CA512341264 [↗](#)

1150152 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3c34fe88-e5c4-44d8-a423-e43acc7a8bab

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HGVS expressions

NM_001754.5:c.1158C>T

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NC_000021.9:g.34792420G>A

CM000683.2:g.34792420G>A

NC_000021.8:g.36164717G>A

CM000683.1:g.36164717G>A

NC_000021.7:g.35086587G>A

NG_011402.2:g.1197292C>T

ENST00000675419.1:c.1158C>T

ENST00000300305.7:c.1158C>T

ENST00000344691.8:c.1077C>T

ENST00000399240.5:c.885C>T

ENST00000437180.5:c.1158C>T

ENST00000482318.5:c.*748C>T

NM_001001890.2:c.1077C>T

NM_001754.4:c.1158C>T

NM_001001890.3:c.1077C>T

Likely Benign

Met criteria codes 2

BP4 BP7

Not Met criteria codes 24

BA1 BP3 BP1 BP2 BP5 BS2
BS1 BS4 BS3 PP1 PP2 PP3
PP4 PM6 PM2 PM1 PM3
PM5 PM4 PS1 PS2 PS3
PS4 PVS1

Evidence Links 0

Expert Panel

Myeloid Malignancy VCEP [↗](#)

Criteria Specification Information 

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Myeloid Malignancy VCEP

NM_001754.5(RUNX1): c.1158C>T (p.Pro386=) is a synonymous variant; therefore, no REVEL score is applicable. SpliceAI score is ≤ 0.20 (0.00) (BP4). Evolutionary conservation algorithms predict the site as not being conserved (PhyloP score -4.82834 < 2.0), or the variant is the reference nucleotide in one primate and/or three mammal species (BP7). 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.

Met criteria codes

BP4			Synonymous variant therefore no REVEL score is applicable and SpliceAI is ≤0.20 (0.00) (BP4).
BP7			Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score -4.82834 < 2.0 or the variant is the reference nucleotide in one primate and/or three mammal species) (BP7).

Not Met criteria codes

BA1			Variant only present in gnomAD v2.1.1 but does not meet code criteria (MAF 0, 0/10094 in African subpopulation)
BP3			This rule is not applicable for MM-VCEP
BP1			This rule is not applicable for MM-VCEP
BP2			No homozygotes present in gnomAD v2.1.1 or v3.1.2
BP5			This rule is not applicable for MM-VCEP
BS2			This rule is not applicable for MM-VCEP
BS1			Variant only present in gnomAD v2.1.1 but does not meet code criteria (MAF 0, 0/10094 in African subpopulation)
BS4			No case studies found
BS3			No functional studies found
PP1			No case studies found
PP2			This rule is not applicable for MM-VCEP
PP3			Synonymous variant therefore no REVEL score is applicable and SpliceAI is NOT ≥0.38 (0.00)
PP4			This rule is not applicable for MM-VCEP
PM6			No case studies found

PM2		✖	Variant only present in gnomAD v2.1.1 but does not meet code criteria (MAF 0, 0/10094 in African subpopulation)
PM1		✖	Not a missense variant
PM3		✖	This rule is not applicable for MM-VCEP
PM5		✖	Not a missense variant
PM4		✖	Not an inframe deletion/insertion
PS1		✖	Amino acid has not been previously estbalished as pathogenic
PS2		✖	No case studies found
PS3		✖	No functional studies found
PS4		✖	Prevalence of variant in affected individuals is not higher than in controls
PVS1		✖	Not a null variant

Curation History 		
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