

Variant: *NM_001754.5(RUNX1):c.1098C>T (p.Ile366=)*

Version: 1.0

CA10014203 [↗](#)

1129441 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: deb5fbf1-e5eb-46a5-b1f6-ccaa7def24fd

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HGVS expressions

NM_001754.5(RUNX1):c.1098C>T (p.Ile366=)

NM_001754.5:c.1098C>T

NC_000021.9:g.34792480G>A

CM000683.2:g.34792480G>A

NC_000021.8:g.36164777G>A

CM000683.1:g.36164777G>A

NC_000021.7:g.35086647G>A

NG_011402.2:g.1197232C>T

ENST00000675419.1:c.1098C>T

ENST00000300305.7:c.1098C>T

ENST00000344691.8:c.1017C>T

ENST00000399240.5:c.825C>T

ENST00000437180.5:c.1098C>T

ENST00000482318.5:c.*688C>T

NM_001001890.2:c.1017C>T

NM_001754.4:c.1098C>T

NM_001001890.3:c.1017C>T

Likely Benign

Met criteria codes **2**

BP7 BP4

Not Met criteria codes **24**

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

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.1098C>T (p.Ile366=) is a synonymous variant therefore no REVEL score and SpliceAI is ≤0.20 (0.00) (BP4). Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score -0.378488 < 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 and BP7

Met criteria codes

BP7			Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score -0.378488 < 2.0 or the variant is the reference nucleotide in one primate and/or three mammal species) (BP7).
BP4			Synonymous variant therefore no REVEL score and SpliceAI is ≤0.20 (0.00)

Not Met criteria codes

BS2			This rule is not applicable for MM-VCEP
PVS1			Not a null variant
BS1			Present in gnomAD v2.1.1 and v3.1.2 but does not meet criteria for code
BS4			No case studies found
BS3			No functional studies found
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			No homozygotes present in gnomAD v2.1.1 and v3.1.2.
PS1			Amino acid has not been previously established as pathogenic
PS2			no case studies found
PS3			No functional studies found
PS4			No case studies found
BA1			Present in gnomAD v2.1.1 and v3.1.2 but does not meet criteria for code

PP1			No case studies found
PP2			This rule is not applicable for MM-VCEP
PP3			Synonymous variant therefore no REVEL score and SpliceAI is NOT ≥ 0.38 (0.00)
PP4			This rule is not applicable for MM-VCEP
PM6			No case studies found
PM2			Present in gnomAD v2.1.1 and v3.1.2
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

Curation History 		
  		
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