

Variant: *NM_001754.5(RUNX1):c.84T>C (p.Ser28=)*

Version: 2.0

CA512319034 [↗](#)

1156194 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 74b8c298-a7d1-4620-bc17-21b748ea7800

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HGVS expressions

NM_001754.5:c.84T>C

NM_001754.5(RUNX1):c.84T>C (p.Ser28=)

NC_000021.9:g.34892938A>G

CM000683.2:g.34892938A>G

NC_000021.8:g.36265235A>G

CM000683.1:g.36265235A>G

NC_000021.7:g.35187105A>G

NG_011402.2:g.1096774T>C

ENST00000675419.1:c.84T>C

ENST00000300305.7:c.84T>C

ENST00000416754.1:c.84T>C

ENST00000437180.5:c.84T>C

ENST00000455571.5:c.59-5842T>C

ENST00000475045.6:c.84T>C

ENST00000482318.5:c.59-12225T>C

NM_001754.4:c.84T>C

Likely Benign

Met criteria codes 3

BP4

BP7

PM2_Supporting

Not Met criteria codes 23

PS1

PS2

PS3

PS4

BP3

BP1

BP2

BP5

PP1

PP2

PP3

PP4

BA1

PM6

PM1

PM3

PM5

PM4

BS2

BS1

BS4

BS3

PVS1

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**

Evidence submitted by expert panel

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.84T>C is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.11) (BP4, 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			This is a synonymous variant that is not predicted to impact splicing. (SpliceAI < 0.2)
BP7			The SpliceAI score is <0.2
PM2_Supporting			This variant is completely absent from gnomAD with at least 20x coverage for RUNX1 (PM2_supporting).

Not Met criteria codes

PS1			This is a synonymous variant.
PS2			There are no published case studies as of 24/01/2023 that have included this variant.
PS3			There is no literature as of 24/01/2023 that have studied this variant.
PS4			As of 24/02/2023 there are no reports of this variant in the literature.
BP3			This rule is not applicable for MM-VCEP
BP1			This rule is not applicable for MM-VCEP.
BP2			There are no published studies as of 24/01/2023 that have included this variant.
BP5			This rule is not applicable for MM-VCEP
PP1			There are no published case studies as of 24/01/2023 that have included this variant.
PP2			This rule is not applicable for MM-VCEP.
PP3			This synonymous variant is not predicted to impact splicing.
PP4			This rule is not applicable for MM-VCEP
BA1			Allele is absent from ExAC, 1000Genomes and ESP.

