

Variant: NM_001754.5(RUNX1):c.1428G>T (p.Val476=)

Version: 1.0

CA512232105 [↗](#)

1571183 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 68630317-cd23-4bd2-9d22-9d0ca0965383

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HGVS expressions

NM_001754.5(RUNX1):c.1428G>T (p.Val476=)

NM_001754.5:c.1428G>T

NC_000021.9:g.34792150C>A

CM000683.2:g.34792150C>A

NC_000021.8:g.36164447C>A

CM000683.1:g.36164447C>A

NC_000021.7:g.35086317C>A

NG_011402.2:g.1197562G>T

ENST00000675419.1:c.1428G>T

ENST00000300305.7:c.1428G>T

ENST00000344691.8:c.1347G>T

ENST00000399240.5:c.1155G>T

ENST00000437180.5:c.1428G>T

ENST00000482318.5:c.*1018G>T

NM_001001890.2:c.1347G>T

NM_001754.4:c.1428G>T

NM_001001890.3:c.1347G>T

Likely Benign

Met criteria codes **3**

BP4

PM2_Supporting

BP7

Not Met criteria codes **23**

BA1

BP3

BP1

BP2

BP5

BS2

BS1

BS4

BS3

PP1

PP2

PP3

PP4

PM6

PM1

PM3

PM5

PM4

PS1

PS2

PS3

PS4

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 Version 2

[↗](#) **PDF**

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.1428G>T (p.Val476=) is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.0) (BP4). This variant has a SpliceAI score ≤ 0.20 (0.0) and evolutionary conservation algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (-0.13) (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 synonymous variant has a SpliceAI score ≤ 0.20 (0.0) (BP4).
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).
BP7			This variant has a SpliceAI score ≤ 0.20 (0.0) and evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (-0.13) (BP7).

Not Met criteria codes

BA1			This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
BP3			This rule is not applicable for MM-VCEP.
BP1			This rule is not applicable for MM-VCEP.
BP2			This variant has not been observed in trans with a pathogenic variant for a fully penetrant dominant gene/disorder or observed in cis with a pathogenic variant in any inheritance pattern.
BP5			This rule is not applicable for MM-VCEP.
BS2			This rule is not applicable for MM-VCEP.
BS1			This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset.
BS4			Segregation data for this variant has not been reported in literature.
BS3			In vitro or in vivo functional data has not been reported for this variant in the literature.
PP1			Segregation data for this variant has not been reported in literature.
PP2			This rule is not applicable for MM-VCEP.
PP3			This synonymous or intronic variant does not have a REVEL score ≥ 0.88 or a SpliceAI score ≥ 0.38 .

PP4		✖	This rule is not applicable for MM-VCEP.
PM6		✖	De novo data for this variant has not been reported in literature.
PM1		✖	This variant is not a missense variant.
PM3		✖	This rule is not applicable for MM-VCEP.
PM5		✖	This variant is not a missense variant.
PM4		✖	This variant is not an in-frame deletion/insertion.
PS1		✖	This variant is not a missense variant.
PS2		✖	De novo data for this variant has not been reported in literature.
PS3		✖	In vitro or in vivo functional data has not been reported for this variant in the literature.
PS4		✖	Proband data for this variant has not been reported in literature.
PVS1		✖	This variant is not a null variant.

Curation History 

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