

Variant: *NM_001754.5(RUNX1):c.270G>A (p.Val90=)*

Version: 2.0

CA512318868 [↗](#)

1116571 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 142360ab-0f52-479d-abb9-f98eca7aa7aa

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HGVS expressions

NM_001754.5:c.270G>A

NM_001754.5(RUNX1):c.270G>A (p.Val90=)

NC_000021.9:g.34886924C>T

CM000683.2:g.34886924C>T

NC_000021.8:g.36259221C>T

CM000683.1:g.36259221C>T

NC_000021.7:g.35181091C>T

NG_011402.2:g.1102788G>A

ENST00000675419.1:c.270G>A

ENST00000300305.7:c.270G>A

ENST00000344691.8:c.189G>A

ENST00000358356.9:c.189G>A

ENST00000399237.6:c.234G>A

ENST00000399240.5:c.189G>A

ENST00000437180.5:c.270G>A

ENST00000455571.5:c.231G>A

ENST00000482318.5:c.59-6211G>A

NM_001001890.2:c.189G>A

NM_001122607.1:c.189G>A

NM_001754.4:c.270G>A

NM_001001890.3:c.189G>A

NM_001122607.2:c.189G>A

Likely Benign

Met criteria codes **3**

BP7 BP4 PM2_Supporting

Not Met criteria codes **23**

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

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**

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.270G>A (p.Val90=) is a synonymous variant which has a MAF ≤ 0.00005 in gnomAD v4.0 across all subpopulations with at least 20X coverage for RUNX1 (PM2_supporting). This variant has a SpliceAI score ≤ 0.20 (0.14) (BP4, 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: PM2_supporting, BP4, BP7.

Met criteria codes

BP7			SpliceAi = 0.14
BP4			SpliceAI = 0.14
PM2_Supporting			This variant has a MAF ≤ 0.00005 in gnomAD v4.0 in all subpopulations with at least 20X coverage for RUNX1 (PM2_supporting).

Not Met criteria codes

PVS1			Not a null variant
BA1			This variant is completely absent from all population databases with at least 20x coverage for RUNX1
BP5			This rule is not applicable for the MM-VCEP
BP3			This rule is not applicable for the MM-VCEP
BP1			This rule is not applicable for the MM-VCEP
BP2			Variant absent in gnomAD therefore no homozygotes present
BS1			This variant is completely absent from all population databases with at least 20x coverage for RUNX1
BS4			No case studies found
BS3			No functional studies found
BS2			This rule is not applicable for the MM-VCEP
PP1			no case studies found

PP2		✖	This rule is not applicable for the MM-VCEP
PP3		✖	Synonymous variant therefore no REVEL score applicable and SpliceAI is not ≥ 0.38 (0.26 Donor Gain)
PP4		✖	This rule is not applicable for the MM-VCEP
PS1		✖	Amino acid location has not been previously established as pathogenic
PS2		✖	no case studies found
PS3		✖	No functional studies found
PS4		✖	prevalence of variant is not significantly increased compared to prevalence in controls
PM1		✖	Not a missense variant
PM3		✖	This rule is not applicable for the MM-VCEP
PM5		✖	Not a missense variant
PM4		✖	Not an in-frame deletion/insertion
PM6		✖	No case studies found

Curation History 		
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