

Variant: NM_001754.5(RUNX1):c.216G>A (p.Arg72=)

Version: 1.0

CA512318911 [↗](#)

2853076 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3fb3ded9-665e-4f36-889e-dae83f30f117

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HGVS expressions

NM_001754.5(RUNX1):c.216G>A (p.Arg72=)

NM_001754.5:c.216G>A

NC_000021.9:g.34886978C>T

CM000683.2:g.34886978C>T

NC_000021.8:g.36259275C>T

CM000683.1:g.36259275C>T

NC_000021.7:g.35181145C>T

NG_011402.2:g.1102734G>A

ENST00000675419.1:c.216G>A

ENST00000300305.7:c.216G>A

ENST00000344691.8:c.135G>A

ENST00000358356.9:c.135G>A

ENST00000399237.6:c.180G>A

ENST00000399240.5:c.135G>A

ENST00000437180.5:c.216G>A

ENST00000455571.5:c.177G>A

ENST00000482318.5:c.59-6265G>A

NM_001001890.2:c.135G>A

NM_001122607.1:c.135G>A

NM_001754.4:c.216G>A

NM_001001890.3:c.135G>A

NM_001122607.2:c.135G>A

Likely Benign

Met criteria codes **3**

BP7 BP4 PM2_Supporting

Not Met criteria codes **23**

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

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

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.216G>A (p.Arg72=) 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 (1.43) (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

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 (1.43) (BP7).
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).

Not Met criteria codes

BS2			This rule is not applicable for MM-VCEP.
PVS1			This variant is not a null variant.
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.
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			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.
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.
BA1			This variant does not have a MAF $\geq$ 0.0015 (0.15%) in any general continental population dataset.
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 $\geq$ 0.88 or a SpliceAI score $\geq$ 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.

Curation History 





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