

Variant: *NM_001754.5(RUNX1):c.1071G>A (p.Pro357=)*

Version: 1.0

CA512341330 [↗](#)

2146447 (ClinVar) [↗](#)

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

Condition: hereditary thrombocytopenia and hematological cancer predisposition syndrome associated with RUNX1 ([MONDO:0100083](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 89d87f97-5584-481e-9d7b-61d6e8fd11c5

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HGVS expressions

NM_001754.5:c.1071G>A

NM_001754.5(RUNX1):c.1071G>A (p.Pro357=)

NC_000021.9:g.34792507C>T

CM000683.2:g.34792507C>T

NC_000021.8:g.36164804C>T

CM000683.1:g.36164804C>T

NC_000021.7:g.35086674C>T

NG_011402.2:g.1197205G>A

ENST00000675419.1:c.1071G>A

ENST00000300305.7:c.1071G>A

ENST00000344691.8:c.990G>A

ENST00000399240.5:c.798G>A

ENST00000437180.5:c.1071G>A

ENST00000482318.5:c.*661G>A

NM_001001890.2:c.990G>A

NM_001754.4:c.1071G>A

NM_001001890.3:c.990G>A

Likely Benign

Met criteria codes **2**

BP4 BP7

Not Met criteria codes **24**

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

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

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

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

Evidence submitted by expert panel

Myeloid Malignancy VCEP

This synonymous variant has a SpliceAI Δ score of 0.00 (BP4). Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score -0.008 (< 2)) (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, BP7.

Met criteria codes

BP4			PhyloP100way= -3.096 Splice AI Δ score = 0.00 BP4: This synonymous variant has a Splice AI Δ score = 0.00
BP7			PhyloP100way= -3.096 Splice AI Δ score = 0.00 BP7: Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score -3.096 (< 2.0) or the variant is the reference nucleotide in one primate and/or three mammal species) (BP7).

Not Met criteria codes

PS1			not applicable
PS2			nil data
PS3			nil data
PS4			nil data
BA1			allele frequency 0.00000911 (gnomAD Version: 20210610)
PP1			nil data
PP2			not applicable
PP3			PhyloP100way= -3.096 Splice AI Δ score = 0.00
PP4			not applicable
PM1			not applicable
PM3			not applicable
PM5			not applicable
PM4			Not applicable
PM6			nil data

PM2		×	allele frequency 0.00000911 (gnomAD Version: 20210610)
BS2		×	not applicable
BS1		×	allele frequency 0.00000911 (gnomAD Version: 20210610)
BS4		×	nil data
BS3		×	nil data
BP3		×	Not applicable
BP1		×	not applicable
BP2		×	not applicable
PVS1		×	not applicable
BP5		×	not applicable

Curation History 		
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