

Variant: NM_001754.5(RUNX1):c.813G>A (p.Arg271=)

Version: 1.0

CA10014289 [↗](#)

1155123 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 58b42f1e-bfa8-4f25-a138-2f0bf3570e34

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HGVS expressions

NM_001754.5(RUNX1):c.813G>A (p.Arg271=)

NM_001754.5:c.813G>A

NC_000021.9:g.34799455C>T

CM000683.2:g.34799455C>T

NC_000021.8:g.36171752C>T

CM000683.1:g.36171752C>T

NC_000021.7:g.35093622C>T

NG_011402.2:g.1190257G>A

ENST00000675419.1:c.813G>A

ENST00000300305.7:c.813G>A

ENST00000344691.8:c.732G>A

ENST00000399240.5:c.540G>A

ENST00000437180.5:c.813G>A

ENST00000482318.5:c.*403G>A

NM_001001890.2:c.732G>A

NM_001754.4:c.813G>A

NM_001001890.3:c.732G>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

[↗](#) PDF

[↗](#) Criteria Specification Approval History

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Myeloid Malignancy VCEP

This synonymous variant (no REVEL score applicable) has SpliceAI scores ≤0.20 (0.01) (BP4). Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score < 2.0 (0.522) (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			Synonymous variant (no REVEL score applicable) and SpliceAI score is ≤0.20 (0.01)
BP7			Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score < 2.0 (0.522) (BP7).

Not Met criteria codes

PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			This rule is not applicable for MM-VCEP
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			This rule is not applicable for MM-VCEP
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			This rule is not applicable for MM-VCEP
BP1			This rule is not applicable for MM-VCEP
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			This rule is not applicable for MM-VCEP

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