

Variant: NM_001754.5(RUNX1):c.781C>T (p.Pro261Ser)

Version: 1.0

CA410206700 [↗](#)

1515908 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 55b2ea7f-2880-4508-8f5f-f5af9204e0fc

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HGVS expressions

NM_001754.5(RUNX1):c.781C>T (p.Pro261Ser)

NM_001754.5:c.781C>T

NC_000021.9:g.34834434G>A

CM000683.2:g.34834434G>A

NC_000021.8:g.36206731G>A

CM000683.1:g.36206731G>A

NC_000021.7:g.35128601G>A

NG_011402.2:g.1155278C>T

ENST00000675419.1:c.781C>T

ENST00000300305.7:c.781C>T

ENST00000344691.8:c.700C>T

ENST00000358356.9:c.700C>T

ENST00000399237.6:c.745C>T

ENST00000399240.5:c.532+25040C>T

ENST00000437180.5:c.781C>T

ENST00000469087.1:n.317C>T

ENST00000482318.5:c.*371C>T

NM_001001890.2:c.700C>T

NM_001122607.1:c.700C>T

NM_001754.4:c.781C>T

NM_001001890.3:c.700C>T

NM_001122607.2:c.700C>T

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **25**

PS1 PS2 PS3 PS4 BA1 PP1
PP2 PP3 PP4 PM1 PM3
PM5 PM4 PM6 BS2 BS1
BS4 BS3 BP4 BP3 BP1 BP2
PVS1 BP5 BP7

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.781C>T (p.Pro261Ser) is a missense variant, which is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting). In summary, the clinical significance of this variant is uncertain. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: PM2_Supporting.

Met criteria codes

PM2_Supporting			Completely absent from gnomAD v2+v3+v4 with a mean coverage of at least 20X.
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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 relevant literature was found in LOVD, HGMD, ClinVar, COSMIC, Mastermind, and Google/Google Scholar searches.
PS3			No relevant literature was found in LOVD, HGMD, ClinVar, COSMIC, Mastermind, and Google/Google Scholar searches.
PS4			No relevant literature was found in LOVD, HGMD, ClinVar, COSMIC, Mastermind, and Google/Google Scholar searches.
BA1			Completely absent from gnomAD v2+v3+v4 with a mean coverage of at least 20X.
PP1			No relevant literature was found in LOVD, HGMD, ClinVar, COSMIC, Mastermind, and Google/Google Scholar searches.
PP2			Not applicable
PP3			The REVEL score = 0, which is uninformative. SpliceAI doesn't predict any significant splicing impact (Δ scores $\leq$ 0.20).
PP4			Not applicable
PM1			Not located at a hotspot (R107, K110, A134, R162, R166, S167, R169, G170, K194, T196, D198, R210, R204) or within residues 89-204.
PM3			Not applicable

PM5			P261/P234 variants are reported in COSMIC and/or Mastermind, but not at a high enough frequency that they would be likely classified as LP/P.
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No relevant literature was found in LOVD, HGMD, ClinVar, COSMIC, Mastermind, and Google/Google Scholar searches.
BS2			Not applicable
BS1			Completely absent from gnomAD v2+v3+v4 with a mean coverage of at least 20X.
BS4			No relevant literature was found in LOVD, HGMD, ClinVar, COSMIC, Mastermind, and Google/Google Scholar searches.
BS3			No relevant literature was found in LOVD, HGMD, ClinVar, COSMIC, Mastermind, and Google/Google Scholar searches.
BP4			The REVEL score = 0, which is uninformative. SpliceAI doesn't predict any significant splicing impact (Δ scores $\leq$ 0.20).
BP3			Not applicable
BP1			Not applicable
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			Not applicable
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

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