

Variant: NM_001754.5(RUNX1):c.893C>T (p.Pro298Leu)

Version: 1.0

CA410149706 [↗](#)

1972477 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 8cac9689-c611-4ada-9cc6-196038b09afe

Approved on: 2024/8/1

Published on: 2024/8/1

HGVS expressions

NM_001754.5(RUNX1):c.893C>T (p.Pro298Leu)

NM_001754.5:c.893C>T

NC_000021.9:g.34799375G>A

CM000683.2:g.34799375G>A

NC_000021.8:g.36171672G>A

CM000683.1:g.36171672G>A

NC_000021.7:g.35093542G>A

NG_011402.2:g.1190337C>T

ENST00000675419.1:c.893C>T

ENST00000300305.7:c.893C>T

ENST00000344691.8:c.812C>T

ENST00000399240.5:c.620C>T

ENST00000437180.5:c.893C>T

ENST00000482318.5:c.*483C>T

NM_001001890.2:c.812C>T

NM_001754.4:c.893C>T

NM_001001890.3:c.812C>T

Uncertain Significance

Met criteria codes **2**

PM2_Supporting

BP4

Not Met criteria codes **22**

PS1

PS2

PS3

BA1

PP1

PP2

PP3

PP4

PM1

PM3

PM5

PM4

PM6

BS1

BS4

BS3

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**

Evidence submitted by expert panel

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.893C>T (p.Pro298Leu) is a missense variant which is completely absent from all population databases with at least 20x coverage for RUNX1 in gnomAD v2.1.1 and v3.1.2 (PM2_supporting). This missense variant has a REVEL score < 0.50 (0.163) and a SpliceAI score ≤ 0.20 (0.01) (BP4). 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: BP4, PM2_supporting.

Met criteria codes

PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 in gnomAD v2.1.1 and v3.1.2 (PM2_supporting).
BP4			This missense variant has a REVEL score < 0.50 (0.163) and a SpliceAI score ≤ 0.20 (0.01) (BP4).

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
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			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
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			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.