

Variant: NM_001754.5(RUNX1):c.877T>C (p.Ser293Pro)

Version: 1.0

CA410150061 [↗](#)

2972103 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: f99ff5cb-9026-4a42-b078-a8d06cb17d37

Approved on: 2024/9/12

Published on: 2024/9/12

HGVS expressions

NM_001754.5(RUNX1):c.877T>C (p.Ser293Pro)

NM_001754.5:c.877T>C

NC_000021.9:g.34799391A>G

CM000683.2:g.34799391A>G

NC_000021.8:g.36171688A>G

CM000683.1:g.36171688A>G

NC_000021.7:g.35093558A>G

NG_011402.2:g.1190321T>C

ENST00000675419.1:c.877T>C

ENST00000300305.7:c.877T>C

ENST00000344691.8:c.796T>C

ENST00000399240.5:c.604T>C

ENST00000437180.5:c.877T>C

ENST00000482318.5:c.*467T>C

NM_001001890.2:c.796T>C

NM_001754.4:c.877T>C

NM_001001890.3:c.796T>C

Uncertain Significance

Met criteria codes **2**

BP4

PM2_Supporting

Not Met criteria codes **24**

BA1

BP3

BP1

BP2

BP5

BP7

BS2

BS1

BS4

BS3

PP1

PP2

PP3

PP4

PM6

PM1

PM3

PM5

PM4

PS1

PS2

PS3

PS4

PVS1

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.877T>C (p.Ser293Pro) is a missense variant which is completely absent from gnomAD 2.0 with at least 20x coverage for RUNX1 (PM2_supporting). This missense variant has a REVEL score <0.50 (0.212) 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

BP4			This missense variant has a REVEL score < 0.50 (0.212) and a SpliceAI score ≤ 0.20 (0.01) (BP4).
PM2_Supporting			This variant is completely absent from gnomAD 2.0 with at least 20x coverage for RUNX1 (PM2_Supporting).

Not Met criteria codes

BA1			Absent in gnomAD
BP3			This rule is not applicable for MM-VCEP
BP1			This rule is not applicable for MM-VCEP
BP2			Information not available
BP5			This rule is not applicable for MM-VCEP
BP7			This variant is not a synonymous or intronic variant
BS2			This rule is not applicable for MM-VCEP
BS1			Absent in gnomAD
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.
PP1			Segregation data for this variant has not been reported in literature.
PP2			This rule is not applicable for MM-VCEP
PP3			REVEL Score 0.212 Splice AI 0.01
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 does not affect any of the following amino acid residues, nor is it located within the RHD: R107, K110, A134, R162, R166, S167, R169, G170, K194, T196, D198, R201, R204 OR within residues 89-204.
PM3			This rule is not applicable for MM-VCEP
PM5			There has not yet been a different missense change determined to be pathogenic at this amino acid residue.
PM4			This variant is not an in-frame deletion/insertion.
PS1			There has not yet been a missense change determined to be pathogenic at this amino acid residue.
PS2			This variant does not have a proven de novo occurrence in the 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.
PVS1			This variant is not a null variant.

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.