

Variant: NM_001754.5(RUNX1):c.717C>T (p.Ser239=)

Version: 1.1

CA512318611 [↗](#)

747644 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b233c2fd-6bc5-424f-9697-0b3590271ff4

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HGVS expressions

NM_001754.5(RUNX1):c.717C>T (p.Ser239=)

NM_001754.5:c.717C>T

NC_000021.9:g.34834498G>A

CM000683.2:g.34834498G>A

NC_000021.8:g.36206795G>A

CM000683.1:g.36206795G>A

NC_000021.7:g.35128665G>A

NG_011402.2:g.1155214C>T

ENST00000675419.1:c.717C>T

ENST00000300305.7:c.717C>T

ENST00000344691.8:c.636C>T

ENST00000358356.9:c.636C>T

ENST00000399237.6:c.681C>T

ENST00000399240.5:c.532+24976C>T

ENST00000437180.5:c.717C>T

ENST00000469087.1:n.253C>T

ENST00000482318.5:c.*307C>T

NM_001001890.2:c.636C>T

NM_001122607.1:c.636C>T

NM_001754.4:c.717C>T

NM_001001890.3:c.636C>T

NM_001122607.2:c.636C>T

Uncertain Significance

Met criteria codes **2**

PM2_Supporting PP3

Not Met criteria codes **24**

BA1 BP4 BP3 BP1 BP2 BP5
BP7 BS2 BS1 BS4 BS3 PP1
PP2 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

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.717C>T (p.Ser239=) is a synonymous variant therefore no REVEL score and SpliceAI score donor gain 0.52 which is ≥0.38 (PP3). This variant 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, PP3.

Met criteria codes

PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2).
PP3			synonymous variant therefore no REVEL score and SpliceAI score donor gain 0.52 which is ≥0.38 (PP3).

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			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 published in vitro or in vivo functional studies were found
BP5			This rule is not applicable for MM-VCEP
BP7			SpliceAI predicts donor gain (0.52), therefore BP7 is NOT MET.
BS2			This rule is not applicable for MM-VCEP
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			No published in vitro or in vivo functional studies were found
BS3			No published in vitro or in vivo functional studies were found
PP1			No published in vitro or in vivo functional studies were found

			
PP2		✖	This rule is not applicable for MM-VCEP
PP4		✖	This rule is not applicable for MM-VCEP
PM6		✖	No published in vitro or in vivo functional studies were found
PM1		✖	This variant occurs outside the conserved RUNX1 RHD
PM3		✖	This rule is not applicable for MM-VCEP
PM5		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4		✖	Synonymous variant
PS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2		✖	No published in vitro or in vivo functional studies were found
PS3		✖	No published in vitro or in vivo functional studies were found
PS4		✖	No published in vitro or in vivo functional studies were found
PVS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 	
	
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