

Variant: NM_001754.5(RUNX1):c.599C>A (p.Pro200His)

Version: 1.0

CA410207960 [↗](#)

1004688 (ClinVar) [↗](#)

Gene: RUNX1 (HGNC:861)

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 149407af-db7c-48f3-a958-05ee667b179e

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HGVS expressions

NM_001754.5:c.599C>A

NM_001754.5(RUNX1):c.599C>A (p.Pro200His)

NC_000021.9:g.34859488G>T

CM000683.2:g.34859488G>T

NC_000021.8:g.36231785G>T

CM000683.1:g.36231785G>T

NC_000021.7:g.35153655G>T

NG_011402.2:g.1130224C>A

ENST00000675419.1:c.599C>A

ENST00000300305.7:c.599C>A

ENST00000344691.8:c.518C>A

ENST00000358356.9:c.518C>A

ENST00000399237.6:c.563C>A

ENST00000399240.5:c.518C>A

ENST00000437180.5:c.599C>A

ENST00000467577.1:n.91C>A

ENST00000482318.5:c.*189C>A

NM_001001890.2:c.518C>A

NM_001122607.1:c.518C>A

NM_001754.4:c.599C>A

NM_001001890.3:c.518C>A

NM_001122607.2:c.518C>A

Uncertain Significance

Met criteria codes **3**

PP3 PM2_Supporting

PM1_Supporting

Not Met criteria codes **23**

PP1 PP2 PP4 PVS1 PM6

PM3 PM5 PM4 BA1 BS1

BS4 BS3 BS2 PS1 PS2 PS3

PS4 BP5 BP7 BP4 BP3 BP1

BP2

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.599C>A (p.Pro200His) is a missense variant which is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). It has a REVEL score of 0.947 (REVEL score \geq 0.88), meeting PP3. This missense variant is located within the Runt Homology Domain (AA 89-204), but does not occur in an established hotspot residue (PM1_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: PM1_supporting, PM2_supporting, and PP3.

Met criteria codes

PP3			REVEL score= 0.947 (REVELscore \geq 0.88)
PM2_Supporting			Variant is absent from control population databases. PM2_supporting
PM1_Supporting			Meets PM1_supporting. Variant affecting one of the other amino acid residues 89-204 within the RHD.

Not Met criteria codes

PP1			No case study found
PP2			This rule is not applicable for MM-VCEP
PP4			This rule is not applicable for MM-VCEP
PVS1			This is a missense variant
PM6			No case study found
PM3			This rule is not applicable for MM-VCEP
PM5			A Missense change at amino acid residue 200 has not been determined to be pathogenic before
PM4			This is a missense variant
BA1			Variant is absent from control population databases. PM2_supporting met
BS1			Variant is absent from control population databases. PM2_supporting met

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