

Variant: *NM_001754.5(RUNX1):c.1020C>G (p.Pro340=)*

Version: 1.0

CA512341366 [↗](#)

758049 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 95d282c7-2a56-469b-b26c-ecde2f514069

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HGVS expressions

NM_001754.5:c.1020C>G

NM_001754.5(RUNX1):c.1020C>G (p.Pro340=)

NC_000021.9:g.34792558G>C

CM000683.2:g.34792558G>C

NC_000021.8:g.36164855G>C

CM000683.1:g.36164855G>C

NC_000021.7:g.35086725G>C

NG_011402.2:g.1197154C>G

ENST00000675419.1:c.1020C>G

ENST00000300305.7:c.1020C>G

ENST00000344691.8:c.939C>G

ENST00000399240.5:c.747C>G

ENST00000437180.5:c.1020C>G

ENST00000482318.5:c.*610C>G

NM_001001890.2:c.939C>G

NM_001754.4:c.1020C>G

NM_001001890.3:c.939C>G

Likely Benign

Met criteria codes **3**

BP4 BP7 PM2_Supporting

Not Met criteria codes **23**

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

Evidence Links **0**

Expert Panel

Myeloid Malignancy VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Myeloid Malignancy VCEP

This variant NM_001754.5(RUNX1):c.1020C>G (p.Pro340=) is a synonymous variant. As it is a synonymous variant there is no REVEL score but SpliceAI is ≤0.20 (0.00) (BP4). The PhyloP Score is <2.0 (-0.628) (BP7). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). In summary, this variant meets criteria to be classified as likely benign. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BP4, BP7, PM2_supporting.

Met criteria codes

BP4			Synonymous variant, therefore, has no REVEL score but SpliceAI is ≤0.20 (0.00)
BP7			PhyloP Score is <2.0 (-0.628)
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1.

Not Met criteria codes

PM1			Synonymous variant outside of hotspot/ critical domain
PM3			This rule is not applicable for MM-VCEP
PM5			Synonymous variant does not meet PM5
PM4			Synonymous variant does not meet PM4
PM6			No case studies found
BS2			This rule is not applicable for MM-VCEP
BS1			Meets PM2.
BS4			No case studies found
BS3			No functional studies found
PVS1			Synonymous variant does not meet PVS1
PS1			Amino acid (proline) at 340 change has not been recorded as pathogenic.
PS2			No case studies found
PS3			No functional studies found

