

Variant: *NM_001754.4(RUNX1):c.183G>A (p.Pro61=)*

Version: 1.0

CA208646 [↗](#)

212088 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9c14a91f-704c-4baf-95d3-a804a2b9ec8e

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HGVS expressions

NM_001754.4(RUNX1):c.183G>A (p.Pro61=)

NM_001754.4:c.183G>A

NC_000021.9:g.34887011C>T

CM000683.2:g.34887011C>T

NC_000021.8:g.36259308C>T

CM000683.1:g.36259308C>T

NC_000021.7:g.35181178C>T

NG_011402.2:g.1102701G>A

ENST00000675419.1:c.183G>A

ENST00000300305.7:c.183G>A

ENST00000344691.8:c.102G>A

ENST00000358356.9:c.102G>A

ENST00000399237.6:c.147G>A

ENST00000399240.5:c.102G>A

ENST00000437180.5:c.183G>A

ENST00000455571.5:c.144G>A

ENST00000482318.5:c.59-6298G>A

NM_001001890.2:c.102G>A

NM_001122607.1:c.102G>A

NM_001001890.3:c.102G>A

NM_001122607.2:c.102G>A

NM_001754.5:c.183G>A

Benign

Met criteria codes 3

BA1 BP4 BP2

Not Met criteria codes 23

BP3 BP1 BP5 BP7 BS2 BS1
BS4 BS3 PP1 PP2 PP3 PP4
PM6 PM2 PM1 PM3 PM5
PM4 PS1 PS2 PS3 PS4
PVS1

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Expert Panel

Myeloid Malignancy VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Myeloid Malignancy VCEP

This synonymous variant is present in gnomAD (v2 and v3) at an allele frequency >0.15% with at least >5 alleles in any general continental population (BA1); in addition, the variant was found in homozygosity in the population database (BP2). Although evolutionary conservation prediction algorithms predict the site as being moderately conserved (PhyloP score: 3.03 > 0.1 [-14.1;6.4]) and the variant is not the reference nucleotide in one primate and/or three mammal species, it is predicted by SSF and MES to lead to either an increase in the canonical splice site score or a decrease of the canonical splice site score by no more than 10% and no putative cryptic splice sites are created (BP4). In summary, the clinical significance of this variant is benign. ACMG/AMP criteria applied, as specified by the ClinGen Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BA1, BP2, and BP4.

Met criteria codes

BA1			ALL:0.14% (379/262036 alleles) - AFR:1.55% (343 het and 5 hom) - AMR:0.063% (22) - NFE:0.0025% (3) - OTH:0.015% (1) (gnomAD v2) ALL:0.4444% (637/143326 alleles) - AFR:1.458% (591 het and 11 hom) - AMR:0.08049% (11) - NFE:0.007742% (5) - OTH:0.3714% (8) (gnomAD v3)
BP4			This synonymous variant is predicted by SSF and MES to lead to either an increase in the canonical splice site score or a decrease of the canonical splice site score by no more than 10% and no putative cryptic splice sites are created.
BP2			Variant found in homozygosity in population database/internal lab: AFR:5 hom (gnomAD v2) and AFR:11 hom (gnomAD v3).

Not Met criteria codes

BP3			Not applicable
BP1			Not applicable
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7			This synonymous variant is predicted by SSF and MES to lead to either an increase in the canonical splice site score or a decrease of the canonical splice site score by no more than 10% and no putative cryptic splice sites are created. However, evolutionary conservation prediction algorithms predict the site as being moderately conserved (PhyloP score: 3.03 > 0.1 [-14.1;6.4]), and the variant is not the reference nucleotide in one primate and/or three mammal species.
BS2			Not applicable
BS1			ALL:0.14% (379/262036 alleles) - AFR:1.55% (343 het and 5 hom) - AMR:0.063% (22) - NFE:0.0025% (3) - OTH:0.015% (1) (gnomAD v2) ALL:0.4444% (637/143326 alleles) - AFR:1.458% (591 het and 11 hom) - AMR:0.08049% (11) - NFE:0.007742% (5) - OTH:0.3714% (8) (gnomAD v3)

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
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			Not applicable
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
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			ALL:0.14% (379/262036 alleles) - AFR:1.55% (343 het and 5 hom) - AMR:0.063% (22) - NFE:0.0025% (3) - OTH:0.015% (1) (gnomAD v2) ALL:0.4444% (637/143326 alleles) - AFR:1.458% (591 het and 11 hom) - AMR:0.08049% (11) - NFE:0.007742% (5) - OTH:0.3714% (8) (gnomAD v3)
PM1			Not located at a hotspot (R107, K110, A134, R162, R166, S167, R169, G170, K194, T196, D198, R210, R204) or within residues 105-204.
PM3			Not applicable
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
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
PS4			

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

Curation History [↗](#)

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