

Variant: NM_001754.5(RUNX1):c.1400C>A (p.Ala467Glu)

Version: 1.0

CA410146806 [↗](#)

463983 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: ce0016e0-4c01-4ac9-8859-fe7b3348533e

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HGVS expressions

NM_001754.5(RUNX1):c.1400C>A (p.Ala467Glu)

NM_001754.5:c.1400C>A

NC_000021.9:g.34792178G>T

CM000683.2:g.34792178G>T

NC_000021.8:g.36164475G>T

CM000683.1:g.36164475G>T

NC_000021.7:g.35086345G>T

NG_011402.2:g.1197534C>A

ENST00000675419.1:c.1400C>A

ENST00000300305.7:c.1400C>A

ENST00000344691.8:c.1319C>A

ENST00000399240.5:c.1127C>A

ENST00000437180.5:c.1400C>A

ENST00000482318.5:c.*990C>A

NM_001001890.2:c.1319C>A

NM_001754.4:c.1400C>A

NM_001001890.3:c.1319C>A

Uncertain Significance

Met criteria codes 1

BP4

Not Met criteria codes 25

BA1

BS2

BS1

BS4

BS3

BP3

BP1

BP2

PVS1

BP5

BP7

PS1

PS2

PS3

PS4

PP1

PP2

PP3

PP4

PM1

PM3

PM5

PM4

PM6

PM2

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.1400C>A (p.Ala467Glu) is a missense variant which has a REVEL score ≤ 0.50 (0.09) and a SpliceAI score ≤ 0.20 (0.00) (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.

Met criteria codes

BP4			This missense variant has a REVEL score ≤ 0.50 (0.09) and a SpliceAI score ≤ 0.20 (0.00).
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Not Met criteria codes

BA1			Highest MAF is less than 0.0015 (0.15%) at 0.00004 (0.004%, 1/24686,) in the Latino subpopulation of gnomAD v2 cohort
BS2			This rule is not applicable for the MM-VCEP
BS1			Highest MAF of 0.00004 (0.004%, 1/24686,) in the Latino subpopulation of gnomAD v2 cohort is less than 0.00015 (0.15%)
BS4			This variant has not been reported in affected family members.
BS3			This variant has not been featured in in vitro or in vivo functional studies that show no damaging effect on protein function or splicing.
BP3			This rule is not applicable for the MM-VCEP
BP1			This rule is not applicable for the MM-VCEP
BP2			This variant has not been detected in a homozygous state in an individual or in a population database (gnomAD).
PVS1			This variant has not been featured in in vitro or in vivo functional studies showing a damaging effect on the gene or gene product.
BP5			This rule is not applicable for the MM-VCEP
BP7			This variant is not a synonymous or intronic variant.
PS1			There has not yet been a missense change determined to be pathogenic at this amino acid residue.
PS2			This variant has not been found to co-segregate with the disease in the literature.
PS3			This variant has not been featured in in vitro or in vivo functional studies showing a damaging effect on the gene or gene product.

PS4		✖	This variant has not been reported in probands.
PP1		✖	This variant was not found to co-segregate with disease in multiple affected family members.
PP2		✖	This rule is not applicable for the MM-VCEP
PP3		✖	This missense variant does not have a REVEL score >0.88 (0.085)
PP4		✖	This rule is not applicable for the MM-VCEP
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 NOR is it within residues 89-204
PM3		✖	This rule is not applicable for the MM-VCEP
PM5		✖	There has not yet been a different missense change determined to be pathogenic at this amino acid residue.
PM4		✖	This variant does not affect one of the other residues within the RHD.
PM6		✖	This variant has not been reported in probands in the literature.
PM2		✖	Variant is present in database (gnomAD) with at least 20x coverage for RUNX1: 0.00004 (0.004%, 1/24686) in the Latino subpopulation of gnomAD v2 cohort

Curation History 		
  		
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