The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_001754.5(RUNX1):c.1432A>C (p.Arg478=)

Version: 1.0

CA512232102

2124494 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: aa9f4785-bf15-4e24-8674-f5a882b029ba
Approved on: 2024/9/10
Published on: 2024/9/10

HGVS expressions

NM_001754.5:c.1432A>C
NM_001754.5(RUNX1):c.1432A>C (p.Arg478=)
NC_000021.9:g.34792146T>G
CM000683.2:g.34792146T>G
NC_000021.8:g.36164443T>G
CM000683.1:g.36164443T>G
NC_000021.7:g.35086313T>G
NG_011402.2:g.1197566A>C
ENST00000675419.1:c.1432A>C
ENST00000300305.7:c.1432A>C
ENST00000344691.8:c.1351A>C
ENST00000399240.5:c.1159A>C
ENST00000437180.5:c.1432A>C
ENST00000482318.5:c.*1022A>C
NM_001001890.2:c.1351A>C
NM_001754.4:c.1432A>C
NM_001001890.3:c.1351A>C

Likely Benign

Met criteria codes 3

BP7 BP4 PM2_Supporting

Not Met criteria codes 23

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

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.1432A>C (p.Arg478=) is a synonymous variant which has a SpliceAI score ≤ 0.20 (0.0) (BP4). This variant has a SpliceAI score ≤ 0.20 (0.0) and evolutionary conservation algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (1.87) (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

BP7	✔	This variant has a SpliceAI score ≤ 0.20 (0.0) and evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score ≤ 2.0 (1.87) (BP7).
BP4	✔	This synonymous variant has a SpliceAI score ≤ 0.20 (0.0) (BP4).
PM2_Supporting	✔	This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).

Not Met criteria codes

BS1	✖	This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset.
BS4	✖	Segregation data for this variant has not been reported in literature.
BS3	✖	In vitro or in vivo functional data has not been reported for this variant in the literature.
BS2	✖	This rule is not applicable for MM-VCEP.
BP5	✖	This rule is not applicable for MM-VCEP.
BP3	✖	This rule is not applicable for MM-VCEP.
BP1	✖	This rule is not applicable for MM-VCEP.
BP2	✖	This variant has not been observed in trans with a pathogenic variant for a fully penetrant dominant gene/disorder or observed in cis with a pathogenic variant in any inheritance pattern.
PM6	✖	De novo data for this variant has not been reported in literature.
PS1	✖	This variant is not a missense variant.
PS2	✖	De novo data for this variant has not been reported in literature.
PS3	✖	In vitro or in vivo functional data has not been reported for this variant in the literature.

PS4	ℹ	✖	Proband data for this variant has not been reported in literature.
PM1		✖	This variant is not a missense variant.
PM3		✖	This rule is not applicable for MM-VCEP.
PM5	ℹ	✖	This variant is not a missense variant.
PM4	ℹ	✖	This variant is not an in-frame deletion/insertion.
PVS1	ℹ	✖	This variant is not a null variant.
BA1	ℹ	✖	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
PP1	ℹ	✖	Segregation data for this variant has not been reported in literature.
PP2		✖	This rule is not applicable for MM-VCEP.
PP3	ℹ	✖	This synonymous or intronic variant does not have a REVEL score ≥ 0.88 or a SpliceAI score ≥ 0.38.
PP4		✖	This rule is not applicable for MM-VCEP.

Curation History

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