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Variant: NM_001754.5(RUNX1):c.-9_14dup (p.Ser5fs)

Version: 1.0

CA3054205290

3642848 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: ecfbc170-8d57-4ab1-98a7-cd85ecbf25f8
Approved on: 2025/5/2
Published on: 2025/5/2

HGVS expressions

NM_001754.5(RUNX1):c.-9_14dup (p.Ser5fs)
NM_001754.5:c.-9_14dup
NC_000021.9:g.35048889_35048911dup
CM000683.2:g.35048889_35048911dup
NC_000021.8:g.36421186_36421208dup
CM000683.1:g.36421186_36421208dup
NC_000021.7:g.35343056_35343078dup
NG_011402.2:g.940804_940826dup
ENST00000675419.1:c.-9_14dup
ENST00000300305.7:c.-9_14dup
ENST00000416754.1:c.-9_14dup
ENST00000437180.5:c.-9_14dup
ENST00000455571.5:c.-9_14dup
ENST00000475045.6:c.-9_14dup
ENST00000482318.5:c.-9_14dup
NM_001754.4:c.-9_14dup

Uncertain Significance

Met criteria codes 1

PM2_Supporting

Not Met criteria codes 25

PVS1 PM6 PM1 PM3 PM5 PM4 BA1 BS1 BS4 BS3 BS2 BP5 BP7 BP4 BP3 BP1 BP2 PS1 PS2 PS3 PS4 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.-9_14dup (p.Ser5ArgfsTer22) is a frameshift variant which is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). This variant is in an exon that is absent from RUNX1 isoform a and b (c.1-c.97 as per VCEP specifications) (PVS1_NA). 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: PM2_supporting.

Met criteria codes

PM2_Supporting	✔	This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).

Not Met criteria codes

PVS1	✖	This variant is in an exon that is absent from RUNX1 isoform a and b (c.1-c.97 as per VCEP specifications) (PVS1_NA).
PM6	✖	De novo 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	✖	This variant is a frameshift variant that is NOT downstream of c.98.
PM4	✖	This variant is not an in-frame deletion/insertion.
BA1	✖	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
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.
BP7	✖	This variant is not a synonymous or intronic variant.
BP4	✖	This variant does not have applicable in-silico data available.

BP3		✖	This rule is not applicable for MM-VCEP.
BP1		✖	This rule is not applicable for MM-VCEP.
BP2	ℹ	✖	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.
PS1	ℹ	✖	This variant is not a missense variant.
PS2		✖	De novo data for this variant has not been reported in literature.
PS3	ℹ	✖	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.
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 variant does not have applicable in-silico data available.
PP4		✖	This rule is not applicable for MM-VCEP.

Curation History

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