*Variant: NM_001754.5(RUNX1):c.*3582G>A*

Version: 2.0

CA320248519

897095 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: f14f73c9-6307-4da8-ad3a-b26122d16ba7
Approved on: 2025-01-15
Published on: 2025-01-15

HGVS expressions

NM_001754.5:c.*3582G>A
NM_001754.5(RUNX1):c.*3582G>A
NC_000021.9:g.34788553C>T
CM000683.2:g.34788553C>T
NC_000021.8:g.36160850C>T
CM000683.1:g.36160850C>T
NC_000021.7:g.35082720C>T
NG_011402.2:g.1201159G>A
ENST00000675419.1:c.*3582G>A
ENST00000300305.7:c.*3582G>A
ENST00000344691.8:c.*3582G>A
ENST00000437180.5:c.*3582G>A
NM_001001890.2:c.*3582G>A
NM_001754.4:c.*3582G>A
NM_001001890.3:c.*3582G>A

Likely Benign

Met criteria codes 1

BS1

Not Met criteria codes 11

PP2 PP3 PP4 PM1 PM3 BP4 BP3 BP1 BP5 BP7 BS2

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.*3582G>A is a 3' UTR variant, which has an MAF of 0.01297 (0.1297%, 23/17732 alleles) in the Admixed American subpopulation of the gnomAD v4.1.0 cohort is between 0.00015 (0.015%) and 0.0015 (0.15%) (BS1). 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: BS1.

Met criteria codes

BS1	✔	gnomAD v4.1.0 23/17732 alleles in Admixed American, MAF 0.1297% All: 38/232420 alleles, MAF 0.01635% MAF of 0.01297 (0.1297%, 23/17732 alleles) in the Admixed American subpopulation of the gnomAD v4.1.0 cohort is between 0.00015 (0.015%) and 0.0015 (0.15%) (BS1).

Not Met criteria codes

PP2	✖	This rule is not applicable for MM-VCEP
PP3	✖	UTR variant
PP4	✖	This rule is not applicable for MM-VCEP
PM1	✖	This variant is not a missense variant.
PM3	✖	This rule is not applicable for MM-VCEP
BP4	✖	UTR variant
BP3	✖	This rule is not applicable for MM-VCEP
BP1	✖	This rule is not applicable for MM-VCEP
BP5	✖	This rule is not applicable for MM-VCEP
BP7	✖	UTR variant
BS2	✖	This rule is not applicable for MM-VCEP

Curation History

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