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.*3557C>T*

Version: 1.0

CA320248552

897570 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 74db2f83-2875-4293-a23e-02ca1281796d
Approved on: 2024-11-13
Published on: 2024-11-13

HGVS expressions

NM_001754.5:c.*3557C>T
NM_001754.5(RUNX1):c.*3557C>T
NC_000021.9:g.34788578G>A
CM000683.2:g.34788578G>A
NC_000021.8:g.36160875G>A
CM000683.1:g.36160875G>A
NC_000021.7:g.35082745G>A
NG_011402.2:g.1201134C>T
ENST00000675419.1:c.*3557C>T
ENST00000300305.7:c.*3557C>T
ENST00000344691.8:c.*3557C>T
ENST00000437180.5:c.*3557C>T
NM_001001890.2:c.*3557C>T
NM_001754.4:c.*3557C>T
NM_001001890.3:c.*3557C>T

Uncertain Significance

Not Met criteria codes 17

PS2 PS3 PS4 PP1 PP3 BA1 PM6 PM2 PM1 PM4 PVS1 BS1 BS4 BS3 BP7 BP4 BP2

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.*3557C>T is a 3' UTR change which does not meey any ACMg/AMP criteria. 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:

None.

Not Met criteria codes

PS2	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
PS3	✖	To our knowledge, no in vitro or in vivo functional studies are available for this variant.
PS4	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
PP1	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study and we have no information about its cosegregation with the disease.
PP3	✖	This rule cannot be applied since this is a 3' UTR variant.
BA1	✖	This rule cannot be applied since the variant is very rare in all population databases.
PM6	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
PM2	✖	This rule cannot be applied because the variant's MAF is extremely low, at 0.00003192 in gnomAD v2 and 0.000006584 in gnomAD v3.
PM1	✖	This rule cannot be applied since this is a 3' UTR variant.
PM4	✖	This rule cannot be applied since this is a 3' UTR variant.
PVS1	✖	This rule cannot be applied since this is a 3' UTR variant.
BS1	✖	This rule cannot be applied since the variant is very rare in all population databases.
BS4	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study and we have no information about segregation.
BS3	✖	To our knowledge, no in vitro or in vivo functional studies are available for this variant.
BP7	✖	This rule cannot be applied since this is a 3' UTR variant.
BP4	✖	This rule cannot be applied since this is a 3' UTR variant.
BP2	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.

Curation History

Showing 1 to 1 of 1 rows