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.*3638G>A*

CA914658966

897094 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: c2328855-b9b5-4fb0-97f4-2e6f36b64c1b
Approved on: 2024-09-16
Published on: 2024-09-16

HGVS expressions

NM_001754.5:c.*3638G>A
NM_001754.5(RUNX1):c.*3638G>A
NC_000021.9:g.34788497C>T
CM000683.2:g.34788497C>T
NC_000021.8:g.36160794C>T
CM000683.1:g.36160794C>T
NC_000021.7:g.35082664C>T
NG_011402.2:g.1201215G>A
ENST00000675419.1:c.*3638G>A
ENST00000300305.7:c.*3638G>A
ENST00000344691.8:c.*3638G>A
ENST00000437180.5:c.*3638G>A
NM_001001890.2:c.*3638G>A
NM_001754.4:c.*3638G>A
NM_001001890.3:c.*3638G>A

Uncertain Significance

Not Met criteria codes 26

PP1 PP4 PP3 PP2 PM1 PM5 PM3 PM4 PM6 PM2 PS2 PS4 PS3 PS1 BA1 PVS1 BP7 BP5 BP2 BP3 BP4 BP1 BS4 BS3 BS1 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.*3638G>A is a UTR variant which does not meet ant 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

PP1	✖	Segregation data for this variant has not been reported in literature.
PP4	✖	This rule is not applicable for MM-VCEP.
PP3	✖	This variant does not have applicable in-silico data available.
PP2	✖	This rule is not applicable for MM-VCEP.
PM1	✖	This variant is not a missense variant.
PM5	✖	This variant is not a missense variant.
PM3	✖	This rule is not applicable for MM-VCEP.
PM4	✖	This variant is not an in-frame deletion/insertion.
PM6	✖	De novo data for this variant has not been reported in literature.
PM2	✖	This variant is present in at least one population database.
PS2	✖	De novo data for this variant has not been reported in literature.
PS4	✖	Proband 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.
PS1	✖	This variant is not a missense variant.
BA1	✖	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
PVS1	✖	This variant is not a null variant.
BP7	✖	This variant is not a synonymous or intronic variant.
BP5	✖	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.

BP3		✖	This rule is not applicable for MM-VCEP.
BP4	ⓘ	✖	This variant does not have applicable in-silico data available.
BP1		✖	This rule is not applicable for MM-VCEP.
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.
BS1	ⓘ	✖	This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset.
BS2		✖	This rule is not applicable for MM-VCEP.

Curation History

See Report	Preferred Variant Title	Classification ⓘ	Condition	Published Date	Version ⓘ	Criteria Specification	Gene

No matching records found