*Variant: NM_001754.5(RUNX1):c.*3588G>C*

Version: 1.0

CA10650411

339803 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 350cd902-01a5-498d-abbe-5b4b459a51ca
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HGVS expressions

NM_001754.5:c.*3588G>C
NM_001754.5(RUNX1):c.*3588G>C
NC_000021.9:g.34788547C>G
CM000683.2:g.34788547C>G
NC_000021.8:g.36160844C>G
CM000683.1:g.36160844C>G
NC_000021.7:g.35082714C>G
NG_011402.2:g.1201165G>C
ENST00000675419.1:c.*3588G>C
ENST00000300305.7:c.*3588G>C
ENST00000344691.8:c.*3588G>C
ENST00000437180.5:c.*3588G>C
NM_001001890.2:c.*3588G>C
NM_001754.4:c.*3588G>C
NM_001001890.3:c.*3588G>C

Uncertain Significance

Met criteria codes 1

PM2_Supporting

Not Met criteria codes 23

BA1 PS1 PS2 PS3 PS4 PP1 PP2 PP4 PM6 PM1 PM3 PM5 PM4 BS1 BS4 BS3 BS2 PVS1 BP5 BP7 BP3 BP1 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.*3588G>C is a UTR variant is which completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting). 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

BA1	✖	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
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	✖	Proband data for this variant has not been reported in literature.
PP1	✖	Segregation data for this variant has not been reported in literature.
PP2	✖	This rule is not applicable for MM-VCEP.
PP4	✖	This rule is not applicable for MM-VCEP.
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 not a missense variant.
PM4	✖	This variant is not an in-frame deletion/insertion.
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.
PVS1	ⓘ	✖	This variant is not a null variant.
BP5		✖	This rule is not applicable for MM-VCEP.
BP7	ⓘ	✖	This variant is not a synonymous or intronic variant.
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.

Curation History

Showing 1 to 1 of 1 rows