*Variant: NM_001754.5(RUNX1):c.*3417A>G*

Version: 1.0

CA10653549

339808 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: b2a548a9-9fba-4673-9b10-36d59c099869
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HGVS expressions

NM_001754.5:c.*3417A>G
NM_001754.5(RUNX1):c.*3417A>G
NC_000021.9:g.34788718T>C
CM000683.2:g.34788718T>C
NC_000021.8:g.36161015T>C
CM000683.1:g.36161015T>C
NC_000021.7:g.35082885T>C
NG_011402.2:g.1200994A>G
ENST00000675419.1:c.*3417A>G
ENST00000300305.7:c.*3417A>G
ENST00000344691.8:c.*3417A>G
ENST00000437180.5:c.*3417A>G
NM_001001890.2:c.*3417A>G
NM_001754.4:c.*3417A>G
NM_001001890.3:c.*3417A>G

Likely Benign

Met criteria codes 1

BS1

Not Met criteria codes 25

BA1 BS2 BS4 BS3 BP5 BP7 BP4 BP3 BP1 BP2 PS1 PS2 PS3 PS4 PP1 PP2 PP3 PP4 PVS1 PM6 PM2 PM1 PM3 PM5 PM4

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.*3417A>G is a 3' UTR variant which has a MAF of 0.0001764 (0.01764%, 12/68008, 12 alleles) in the European (non-Finnish) subpopulation of the gnomAD v4 cohort which 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	✔	MAF of 0.0001764 (0.01764%, 12/68008, 12 alleles) in the European (non-Finnish) subpopulation of the gnomAD v4 cohort is between 0.00015 (0.015%) and 0.0015 (0.15%) (BS1).

Not Met criteria codes

BA1	✖	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
BS2	✖	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.
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	✖	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	✖	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.
PP3	ⓘ	✖	This variant does not have applicable in-silico data available.
PP4		✖	This rule is not applicable for MM-VCEP.
PVS1	ⓘ	✖	This variant is not a null variant.
PM6	ⓘ	✖	De novo data for this variant has not been reported in literature.
PM2		✖	This variant is present in at least one population database.
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.

Curation History

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