*Variant: NM_001754.4(RUNX1):c.*3345A>T*

Version: 1.0

CA10652909

339810 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 86fc220e-350e-4d28-97db-e97caaba1f41
Approved on: 2020-01-13
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HGVS expressions

NM_001754.4:c.*3345A>T
NM_001754.4(RUNX1):c.*3345A>T
NC_000021.9:g.34788790T>A
CM000683.2:g.34788790T>A
NC_000021.8:g.36161087T>A
CM000683.1:g.36161087T>A
NC_000021.7:g.35082957T>A
NG_011402.2:g.1200922A>T
ENST00000675419.1:c.*3345A>T
ENST00000300305.7:c.*3345A>T
ENST00000344691.8:c.*3345A>T
ENST00000437180.5:c.*3345A>T
NM_001001890.2:c.*3345A>T
NM_001001890.3:c.*3345A>T
NM_001754.5:c.*3345A>T

Benign

Met criteria codes 2

BP2 BA1

Not Met criteria codes 24

PM6 PM2 PM1 PM3 PM5 PM4 PVS1 BS2 BS1 BS4 BS3 BP4 BP3 BP1 BP5 BP7 PS1 PS2 PS3 PS4 PP1 PP2 PP3 PP4

Evidence Links 0

Expert Panel

Myeloid Malignancy VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Myeloid Malignancy VCEP

The NM_001754.4:c.*3345A>T variant in the 3' UTR has an MAF of 0.09 (9%, 145/1560 alleles) in the East Asian subpopulation of the gnomAD v3 cohort and is ≥ 0.0015 (0.15%) (BA1). This variant is detected in a homozygous state in 16 individuals in the gnomAD v3

population database (BP2). In summary, this variant meets criteria to be classified as benign. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BA1, BP2.

Met criteria codes

BP2	✔	This variant is reported in 16 homozygotes in gnomAD v3 and 12 homozygotes in gnomAD v2.1.1 in the East Asian population.
BA1	✔	This 3' UTR variant is reported in gnomAD v2.1.1 at a frequency of 0.093 (145/1560 alleles) and in gnomAD v3 at a frequency of 0.08 (254/3134 alleles) in the East Asian population. It meets criteria for BA1.

Not Met criteria codes

PM6	✖	No data currently available
PM2	✖	Meets BA1
PM1	✖	N/A
PM3	✖	MM-VCEP deemed N/A for RUNX1
PM5	✖	N/A
PM4	✖	N/A
PVS1	✖	N/A
BS2	✖	MM-VCEP deemed N/A for RUNX1
BS1	✖	Meets BA1
BS4	✖	No data currently available
BS3	✖	No data currently available
BP4	✖	N/A
BP3	✖	MM-VCEP deemed N/A for RUNX1
BP1	✖	MM-VCEP deemed N/A for RUNX1
BP5	✖	MM-VCEP deemed N/A for RUNX1

BP7	✖	N/A
PS1	✖	N/A
PS2	✖	No data currently available
PS3	✖	No data currently available
PS4	✖	The variant has not been reported in patients with familial platelet disorder with predisposition to hematologic malignancies in the literature, to the best of our knowledge.
PP1	✖	No data currently available
PP2	✖	MM-VCEP deemed N/A for RUNX1
PP3	✖	N/A
PP4	✖	MM-VCEP deemed N/A for RUNX1

Curation History

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