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*Variant: NM_001754.4(RUNX1):c.*4032T>C*

Version: 1.0

CA10653545

339797 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 3c95e75d-735c-49cf-9bef-f6bd97b0da95
Approved on: 2020/1/13
Published on: 2020/1/14

HGVS expressions

NM_001754.4(RUNX1):c.*4032T>C
NM_001754.4:c.*4032T>C
NC_000021.9:g.34788103A>G
CM000683.2:g.34788103A>G
NC_000021.8:g.36160400A>G
CM000683.1:g.36160400A>G
NC_000021.7:g.35082270A>G
NG_011402.2:g.1201609T>C
ENST00000675419.1:c.*4032T>C
ENST00000300305.7:c.*4032T>C
ENST00000344691.8:c.*4032T>C
ENST00000437180.5:c.*4032T>C
NM_001001890.2:c.*4032T>C
NM_001001890.3:c.*4032T>C
NM_001754.5:c.*4032T>C

Benign

Met criteria codes 2

BA1 BP2

Not Met criteria codes 24

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

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.*4032T>C variant reported at an MAF of 0.096 (9%, 835/8702 alleles) in the African population in gnomAD cohort is ≥ 0.0015 (0.15%) (BA1). This variant is detected in a homozygous state in 25 individuals in gnomAD (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

BA1	✔	The variant is reported in gnomAD at a frequency of 0.09595 (835/8702 alleles) in the African population, with 25 homozygotes, meeting criteria for BA1 (AF > 0.0015)
BP2	✔	The variant is reported in 25 homozygotes in gnomAD

Not Met criteria codes

PP1	✖	No data currently available
PP2	✖	MM-VCEP deemed N/A for RUNX1
PP3	✖	N/A
PP4	✖	MM-VCEP deemed N/A for RUNX1
PM6	✖	No data currently available
PM2	✖	Variant meets BA1
PVS1	✖	N/A
PM1	✖	N/A
PM3	✖	MM-VCEP deemed N/A for RUNX1
PM5	✖	N/A
PM4	✖	N/A
BS1	✖	Variant meets BA1
BS4	✖	No data currently available
BS3	✖	No data currently available
BS2	✖	MM-VCEP deemed N/A for RUNX1

PS1	ℹ	✖	N/A
PS2		✖	No data currently available
PS3	ℹ	✖	No data currently available
PS4	ℹ	✖	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.
BP5		✖	MM-VCEP deemed N/A for RUNX1
BP7	ℹ	✖	N/A
BP4	ℹ	✖	N/A
BP3		✖	MM-VCEP deemed N/A for RUNX1
BP1		✖	MM-VCEP deemed N/A for RUNX1

Curation History

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