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Variant: NM_001754.5(RUNX1):c.249C>T (p.Ala83=)

Version: 1.0

CA512318889

1153047 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 55792a37-c1ea-4f3f-bfd7-8fba9ad57db5
Approved on: 2022/4/25
Published on: 2022/7/5

HGVS expressions

NM_001754.5(RUNX1):c.249C>T (p.Ala83=)
NM_001754.5:c.249C>T
NC_000021.9:g.34886945G>A
CM000683.2:g.34886945G>A
NC_000021.8:g.36259242G>A
CM000683.1:g.36259242G>A
NC_000021.7:g.35181112G>A
NG_011402.2:g.1102767C>T
ENST00000675419.1:c.249C>T
ENST00000300305.7:c.249C>T
ENST00000344691.8:c.168C>T
ENST00000358356.9:c.168C>T
ENST00000399237.6:c.213C>T
ENST00000399240.5:c.168C>T
ENST00000437180.5:c.249C>T
ENST00000455571.5:c.210C>T
ENST00000482318.5:c.59-6232C>T
NM_001001890.2:c.168C>T
NM_001122607.1:c.168C>T
NM_001754.4:c.249C>T
NM_001001890.3:c.168C>T
NM_001122607.2:c.168C>T

Likely Benign

Met criteria codes 3

BP7 PM2_Supporting BP4

Not Met criteria codes 23

PM1 PM3 PM5 PM4 PM6
BS2 BS1 BS4 BS3 PVS1
BP5 BP3 BP1 BP2 BA1 PS1
PS2 PS3 PS4 PP1 PP2 PP3
PP4

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.249C>T (p.Ala83=) is a synonymous variant. REVEL score not calculable. SpliceAI predicts: Acceptor loss 0.00, Donor loss 0.00, Acceptor gain 0.00, Donor gain 0.00. (BP4) Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 0.324528 < 2.0 or the variant is the reference nucleotide in one primate and/or three mammal species) (BP7). PM2_supporting: This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). 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: BP4, BP7, PM2_supporting.

Met criteria codes

BP7	✔	Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 0.324528 < 2.0 or the variant is the reference nucleotide in one primate and/or three mammal species) (BP7).
PM2_Supporting	✔	PM2_supporting: This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2).
BP4	✔	REVEL score not calculable. SpliceAI predicts: Acceptor loss 0.00, Donor loss 0.00, Acceptor gain 0.00, Donor gain 0.00. (BP4)

Not Met criteria codes

PM1	✖	This variant is outside the RHD.
PM3	✖	This rule is not applicable for MM-VCEP
PM5	✖	Synonymous change, not applicable
PM4	✖	Synonymous variant, not applicable
PM6	✖	No published literature could be found on this variant
BS2	✖	This rule is not applicable for MM-VCEP
BS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4	✖	No published literature could be found on this variant
BS3	✖	No published literature could be found on this variant

PVS1	ℹ	✖	Synonymous change, not applicable
BP5		✖	This rule is not applicable for MM-VCEP
BP3		✖	This rule is not applicable for MM-VCEP
BP1		✖	This rule is not applicable for MM-VCEP
BP2	ℹ	✖	No published literature could be found on this variant
BA1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1	ℹ	✖	Synonymous change, not applicable
PS2		✖	No published literature could be found on this variant
PS3	ℹ	✖	No published literature could be found on this variant
PS4	ℹ	✖	No published literature could be found on this variant
PP1	ℹ	✖	No published literature could be found on this variant
PP2		✖	This rule is not applicable for MM-VCEP
PP3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4		✖	This rule is not applicable for MM-VCEP

Curation History

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