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Variant: NM_001754.4(RUNX1):c.924C>T (p.Ser308=)

Version: 1.0

CA16616518

415830 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 8568b183-5fdb-4f4a-b47c-4a61cbaad38e
Approved on: 2020-04-10
Published on: 2020-06-02

HGVS expressions

NM_001754.4:c.924C>T
NM_001754.4(RUNX1):c.924C>T (p.Ser308=)
NC_000021.9:g.34799344G>A
CM000683.2:g.34799344G>A
NC_000021.8:g.36171641G>A
CM000683.1:g.36171641G>A
NC_000021.7:g.35093511G>A
NG_011402.2:g.1190368C>T
ENST00000675419.1:c.924C>T
ENST00000300305.7:c.924C>T
ENST00000344691.8:c.843C>T
ENST00000399240.5:c.651C>T
ENST00000437180.5:c.924C>T
ENST00000482318.5:c.*514C>T
NM_001001890.2:c.843C>T
NM_001001890.3:c.843C>T
NM_001754.5:c.924C>T

Likely Benign

Met criteria codes 2

BP7 BP4

Not Met criteria codes 24

BP5 BP3 BP1 BP2 BS1 BS4 BS3 BS2 PVS1 PP1 PP2 PP3 PP4 PS1 PS2 PS3 PS4 PM6 PM2 PM1 PM3 PM5 PM4 BA1

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 synonymous variant is predicted by SSF and MES to lead to either an increase in the canonical splice site score or a decrease of the canonical splice site score by no more than 10% and no putative cryptic splice sites are created; in addition, evolutionary conservation prediction algorithms predict the site as not being highly conserved (PhyloP score: -0.68 < 0.1 [-14.1;6.4]) (BP4+BP7). In summary, this variant meets criteria to be classified as likely benign. ACMG/AMP criteria applied, as specified by the ClinGen Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BP4, BP7.

Met criteria codes

BP7	✔	This synonymous variant is predicted by SSF and MES to lead to either an increase in the canonical splice site score or a decrease of the canonical splice site score by no more than 10% and no putative cryptic splice sites are created. In addition, evolutionary conservation prediction algorithms predict the site as not being highly conserved (PhyloP score: -0.68 < 0.1 [-14.1;6.4]).
BP4	✔	This synonymous variant is predicted by SSF and MES to lead to either an increase in the canonical splice site score or a decrease of the canonical splice site score by no more than 10% and no putative cryptic splice sites are created.

Not Met criteria codes

BP5	✖	Not applicable
BP3	✖	Not applicable
BP1	✖	Not applicable
BP2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	✖	Absent from gnomAD v2 ALL:0.001396% (2/143284 alleles) - AMR:0.007323% (1) - AFR:0.002379% (1) (gnomAD v3)
BS4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2	✖	Not applicable
PVS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2	✖	Not applicable

PP3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4	✖	Not applicable
PS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2	✖	Absent from gnomAD v2 ALL:0.001396% (2/143284 alleles) - AMR:0.007323% (1) - AFR:0.002379% (1) (gnomAD v3)
PM1	✖	Not located at a hotspot (R107, K110, A134, R162, R166, S167, R169, G170, K194, T196, D198, R210, R204) or within residues 105-204.
PM3	✖	Not applicable
PM5	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	✖	Absent from gnomAD v2 ALL:0.001396% (2/143284 alleles) - AMR:0.007323% (1) - AFR:0.002379% (1) (gnomAD v3)

Curation History

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