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Variant: NM_001754.5(RUNX1):c.1085C>T (p.Ser362Leu)

Version: 1.0

CA410148181

2008544 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: aa730b3a-a6ad-408c-b234-a7d923af2179
Approved on: 2024/8/1
Published on: 2024/8/1

HGVS expressions

NM_001754.5:c.1085C>T
NM_001754.5(RUNX1):c.1085C>T (p.Ser362Leu)
NC_000021.9:g.34792493G>A
CM000683.2:g.34792493G>A
NC_000021.8:g.36164790G>A
CM000683.1:g.36164790G>A
NC_000021.7:g.35086660G>A
NG_011402.2:g.1197219C>T
ENST00000675419.1:c.1085C>T
ENST00000300305.7:c.1085C>T
ENST00000344691.8:c.1004C>T
ENST00000399240.5:c.812C>T
ENST00000437180.5:c.1085C>T
ENST00000482318.5:c.*675C>T
NM_001001890.2:c.1004C>T
NM_001754.4:c.1085C>T
NM_001001890.3:c.1004C>T

Uncertain Significance

Met criteria codes 2

BP4 PM2_Supporting

Not Met criteria codes 13

BP2 BP7 PS2 PS3 PS4 PP3 PM1 PM4 PM6 PVS1 BA1 BS1 BS3

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.1085C>T (p.Ser362Leu) is a missense variant which has a REVEL score <0.50 (0.223) (BP4). This variant is completely absent from all population databases (gnomAD v2.1.1 and v3.1.2) with at least 20x coverage for RUNX1 (PM2_supporting). In summary, the clinical significance of this variant is uncertain. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BP4, PM2_supporting.

Met criteria codes

BP4	✔	This missense variant has a REVEL score <0.50 (0.223) (BP4).
PM2_Supporting	✔	This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting).

Not Met criteria codes

BP2	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
BP7	✖	This rule cannot be applied since this is a missense variant.
PS2	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
PS3	✖	To our knowledge, no in vitro or in vivo functional studies are available for this variant.
PS4	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
PP3	✖	This rule cannot be applied since this missense variant has not a REVEL score >0.88 (0.223).
PM1	✖	This rule cannot be applied since this missense variant affects amino acid residue 362, which is out of RHD or residues 89-204.
PM4	✖	This rule cannot be applied since the variant is a missense change.
PM6	✖	This rule cannot be applied since, to our knowledge, this variant has not been reported in any study.
PVS1	✖	This rule cannot be applied since this is a missense variant.
BA1	✖	This rule cannot be applied since the variant is completely absent from all population databases.
BS1	✖	This rule cannot be applied since the variant is completely absent from all population databases.
BS3	✖	To our knowledge, no in vitro or in vivo functional studies are available for this variant.

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001754.5(RUNX1):c.1085C>T (p.S...	Uncertain Significance	Hereditary Thrombocytopenia And Hematologic Cancer Predisposition Syndrome	2024-08-01	1.0	ClinGen Myeloid Malignancy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2	RUNX1

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