Variant: NM_001754.5(RUNX1):c.1309A>G (p.Thr437Ala)

Version: 1.0

CA410147422

1424427 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 6dc6e605-5b43-4d50-b264-78642e75b5f0
Approved on: 2024-07-11
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HGVS expressions

NM_001754.5:c.1309A>G
NM_001754.5(RUNX1):c.1309A>G (p.Thr437Ala)
NC_000021.9:g.34792269T>C
CM000683.2:g.34792269T>C
NC_000021.8:g.36164566T>C
CM000683.1:g.36164566T>C
NC_000021.7:g.35086436T>C
NG_011402.2:g.1197443A>G
ENST00000675419.1:c.1309A>G
ENST00000300305.7:c.1309A>G
ENST00000344691.8:c.1228A>G
ENST00000399240.5:c.1036A>G
ENST00000437180.5:c.1309A>G
ENST00000482318.5:c.*899A>G
NM_001001890.2:c.1228A>G
NM_001754.4:c.1309A>G
NM_001001890.3:c.1228A>G

Uncertain Significance

Met criteria codes 1

BP4

Not Met criteria codes 25

BS1 **BS4** **BS3** **BS2** **BP5** **BP7**
BP3 **BP1** **BP2** **PS1** **PS2** **PS3**
PS4 **BA1** **PP1** **PP2** **PP3** **PP4**
PM6 **PM2** **PM1** **PM3** **PM5**
PM4 **PVS1**

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.1309A>G (p.Thr437Ala) is a missense variant which has a REVEL score < 0.50 (0.154) and a SpliceAI score ≤ 0.20 (0.0) (BP4). 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.

Met criteria codes

BP4	ⓘ	✓	This missense variant has a REVEL score < 0.50 (0.154) and a SpliceAI score ≤ 0.20 (0.0) (BP4).

Not Met criteria codes

BS1	ⓘ	✗	This variant does not have a MAF between 0.00015 (0.015%) and 0.0015 (0.15%) in any general continental dataset.
BS4	ⓘ	✗	Segregation data for this variant has not been reported in literature.
BS3	ⓘ	✗	In vitro or in vivo functional data has not been reported for this variant in the literature.
BS2		✗	This rule is not applicable for MM-VCEP.
BP5		✗	This rule is not applicable for MM-VCEP.
BP7	ⓘ	✗	This variant is not a synonymous or intronic variant.
BP3		✗	This rule is not applicable for MM-VCEP.
BP1		✗	This rule is not applicable for MM-VCEP.
BP2	ⓘ	✗	This variant has not been observed in trans with a pathogenic variant for a fully penetrant dominant gene/disorder or observed in cis with a pathogenic variant in any inheritance pattern.
PS1	ⓘ	✗	There has not yet been a missense change determined to be pathogenic at this amino acid residue.
PS2		✗	De novo data for this variant has not been reported in literature.
PS3	ⓘ	✗	In vitro or in vivo functional data has not been reported for this variant in the literature.
PS4	ⓘ	✗	Proband data for this variant has not been reported in literature.
BA1	ⓘ	✗	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
PP1	ⓘ	✗	Segregation data for this variant has not been reported in literature.

PP2		✖	This rule is not applicable for MM-VCEP.
PP3	ⓘ	✖	This missense variant does not have a REVEL score of ≥ 0.88.
PP4		✖	This rule is not applicable for MM-VCEP.
PM6	ⓘ	✖	De novo data for this variant has not been reported in literature.
PM2		✖	This variant is present in at least one population database.
PM1		✖	This variant does not affect any of the following amino acid residues, nor is it located within the RHD: R107, K110, A134, R162, R166, S167, R169, G170, K194, T196, D198, R201, R204 OR within residues 89-204.
PM3		✖	This rule is not applicable for MM-VCEP.
PM5	ⓘ	✖	There has not yet been a different missense change determined to be pathogenic at this amino acid residue.
PM4	ⓘ	✖	This variant is not an in-frame deletion/insertion.
PVS1	ⓘ	✖	This variant is not a null variant.

Curation History

Showing 1 to 1 of 1 rows