*Variant: NM_001754.5(RUNX1):c.*2780A>G*

Version: 1.0

CA10652912

339820 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 71c61932-f02c-46cd-b4d4-4404f0574967
Approved on: 2024-10-29
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HGVS expressions

NM_001754.5:c.*2780A>G
NM_001754.5(RUNX1):c.*2780A>G
NC_000021.9:g.34789355T>C
CM000683.2:g.34789355T>C
NC_000021.8:g.36161652T>C
CM000683.1:g.36161652T>C
NC_000021.7:g.35083522T>C
NG_011402.2:g.1200357A>G
ENST00000675419.1:c.*2780A>G
ENST00000300305.7:c.*2780A>G
ENST00000344691.8:c.*2780A>G
ENST00000437180.5:c.*2780A>G
NM_001001890.2:c.*2780A>G
NM_001754.4:c.*2780A>G
NM_001001890.3:c.*2780A>G

Uncertain Significance

Met criteria codes 1

PM2_Supporting

Not Met criteria codes 25

PVS1 **PS1** **PS2** **PS3** **PS4**
PP1 **PP2** **PP3** **PP4** **PM6**
PM1 **PM3** **PM5** **PM4** **BA1**
BS1 **BS4** **BS3** **BS2** **BP5** **BP7**
BP4 **BP3** **BP1** **BP2**

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.*2780A>G is a UTR variant which is completely absent from all population databases 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: PM2_supporting.

Met criteria codes

PM2_Supporting	✓	This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).

Not Met criteria codes

PVS1	✗	This variant is not a null variant.
PS1	✗	This variant is not a missense, synonymous, or frameshift variant.
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.
PP1	✗	Segregation data for this variant has not been reported in literature
PP2	✗	This rule is not applicable for MM-VCEP.
PP3	✗	This synonymous or intronic variant does not have a REVEL score ≥ 0.88 or a SpliceAI score ≥ 0.38.
PP4	✗	This rule is not applicable for MM-VCEP.
PM6	✗	De novo data for this variant has not been reported in literature.
PM1	✗	This variant is not a missense variant.
PM3	✗	This rule is not applicable for MM-VCEP.
PM5	✗	This variant is not a missense, synonymous, or frameshift variant.
PM4	✗	This variant is not an in-frame deletion/insertion.
BA1	✗	This variant does not have a MAF ≥ 0.0015 (0.15%) in any general continental population dataset.
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	ⓘ	✖	UTR variant.
BP4	ⓘ	✖	UTR 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.

Curation History

Showing 1 to 1 of 1 rows