The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_001754.5(RUNX1):c.1269C>G (p.Arg423=)

Version: 1.0

CA512341156

1160372 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: e00a0f59-2f17-4cc4-b01f-e6b7c9164f4a
Approved on: 2024/6/24
Published on: 2024/6/24

HGVS expressions

NM_001754.5:c.1269C>G
NM_001754.5(RUNX1):c.1269C>G (p.Arg423=)
NC_000021.9:g.34792309G>C
CM000683.2:g.34792309G>C
NC_000021.8:g.36164606G>C
CM000683.1:g.36164606G>C
NC_000021.7:g.35086476G>C
NG_011402.2:g.1197403C>G
ENST00000675419.1:c.1269C>G
ENST00000300305.7:c.1269C>G
ENST00000344691.8:c.1188C>G
ENST00000399240.5:c.996C>G
ENST00000437180.5:c.1269C>G
ENST00000482318.5:c.*859C>G
NM_001001890.2:c.1188C>G
NM_001754.4:c.1269C>G
NM_001001890.3:c.1188C>G

Likely Benign

Met criteria codes 2

BP4 BP7

Not Met criteria codes 24

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

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

Synonymous variant (no REVEL score applicable) with SpliceAI score ≤ 0.20 (0.00) (BP4). Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 0.620 (< 2.0)) (BP7). 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.

Met criteria codes

BP4	✔	Synonymous variant (no REVEL score applicable) with SpliceAI score ≤0.20 (0.00) (BP4)
BP7	✔	Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 0.620 (< 2.0)) (BP7)

Not Met criteria codes

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 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
BP3	✖	This rule is not applicable for MM-VCEP
BP1	✖	This rule is not applicable for MM-VCEP
BP2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5	✖	This rule is not applicable for MM-VCEP
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

PVS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1		✖	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		✖	This rule is not applicable for MM-VCEP
PP3		✖	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
PM6		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3		✖	This rule is not applicable for MM-VCEP
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

Curation History

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