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.182C>G (p.Pro61Arg)

Version: 1.0

CA410203987

1951250 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: 3601d04a-af75-4f56-9ee2-bac3a7f1a2ff
Approved on: 2024/7/17
Published on: 2024/7/17

HGVS expressions

NM_001754.5(RUNX1):c.182C>G (p.Pro61Arg)
NM_001754.5:c.182C>G
NC_000021.9:g.34887012G>C
CM000683.2:g.34887012G>C
NC_000021.8:g.36259309G>C
CM000683.1:g.36259309G>C
NC_000021.7:g.35181179G>C
NG_011402.2:g.1102700C>G
ENST00000675419.1:c.182C>G
ENST00000300305.7:c.182C>G
ENST00000344691.8:c.101C>G
ENST00000358356.9:c.101C>G
ENST00000399237.6:c.146C>G
ENST00000399240.5:c.101C>G
ENST00000437180.5:c.182C>G
ENST00000455571.5:c.143C>G
ENST00000482318.5:c.59-6299C>G
NM_001001890.2:c.101C>G
NM_001122607.1:c.101C>G
NM_001754.4:c.182C>G
NM_001001890.3:c.101C>G
NM_001122607.2:c.101C>G

Uncertain Significance

Met criteria codes 2

PM2_Supporting BP4

Not Met criteria codes 24

PS1 PS2 PS3 PS4 BA1 PP1 PP2 PP3 PP4 PM6 PVS1 PM1 PM3 PM5 PM4 BS1 BS4 BS3 BS2 BP5 BP7 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.182C>G (p.Pro61Arg) is a missense variant which is absent from gnomAD v2.1.1 and v3.1.2 (PM2_supporting). This variant has a REVEL score < 0.50 (0.316), 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, PM2_supporting.

Met criteria codes

PM2_Supporting	✔	Absent from gnomAD.
BP4	✔	REVEL= 0.316 which is below the threshold for pathogenicity and SpliceAI=0 which indicates that there is a very low chance that the variant impacts splicing.

Not Met criteria codes

PS1	✖	Not reported.
PS2	✖	Variant is not found in published studies.
PS3	✖	Variant is not found in published studies.
PS4	✖	Variant is not found in published studies.
BA1	✖	Absent from gnomAD.
PP1	✖	Variant is not found in published studies.
PP2	✖	This rule is not applicable for MM-VCEP
PP3	✖	REVEL= 0.316 and SpliceAI=0
PP4	✖	This rule is not applicable for MM-VCEP
PM6	✖	Variant is not found in published studies.
PVS1	✖	Variant is missense
PM1	✖	Variant is not in a established functional domain.

PM3		✖	This rule is not applicable for MM-VCEP
PM5	ℹ	✖	Not reported.
PM4	ℹ	✖	Variant is a missense variant.
BS1	ℹ	✖	Absent from gnomAD.
BS4	ℹ	✖	Variant is not found in published studies.
BS3	ℹ	✖	Variant is not found in published studies.
BS2		✖	This rule is not applicable for MM-VCEP
BP5		✖	This rule is not applicable for MM-VCEP
BP7	ℹ	✖	Variant is missense.
BP3		✖	This rule is not applicable for MM-VCEP
BP1		✖	This rule is not applicable for MM-VCEP
BP2	ℹ	✖	Variant is not found in published studies.

Curation History

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.