

Variant: NM_001754.5(RUNX1):c.490G>T (p.Val164Phe)

Version: 1.0

CA410202501 [↗](#)

1437603 (ClinVar) [↗](#)

Gene: RUNX1 ([HGNC:861](#))

Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome ([MONDO:0011071](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: ccacf1a75-1215-40dc-9640-a79c4e824cf1

Approved on: 2024/10/29

Published on: 2024/10/29

HGVS expressions

NM_001754.5:c.490G>T

NM_001754.5(RUNX1):c.490G>T (p.Val164Phe)

NC_000021.9:g.34880575C>A

CM000683.2:g.34880575C>A

NC_000021.8:g.36252872C>A

CM000683.1:g.36252872C>A

NC_000021.7:g.35174742C>A

NG_011402.2:g.1109137G>T

ENST00000675419.1:c.490G>T

ENST00000300305.7:c.490G>T

ENST00000344691.8:c.409G>T

ENST00000358356.9:c.409G>T

ENST00000399237.6:c.454G>T

ENST00000399240.5:c.409G>T

ENST00000437180.5:c.490G>T

ENST00000482318.5:c.*80G>T

NM_001001890.2:c.409G>T

NM_001122607.1:c.409G>T

NM_001754.4:c.490G>T

NM_001001890.3:c.409G>T

NM_001122607.2:c.409G>T

Uncertain Significance

Met criteria codes **3**

PP3

PM2_Supporting

PM1_Supporting

Not Met criteria codes **23**

BS2

BS1

BS4

BS3

BP4

BP3

BP1

BP2

BP5

BP7

PVS1

PS1

PS2

PS3

PS4

PP1

PP2

PP4

BA1

PM3

PM5

PM4

PM6

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

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.490G>T (p.Val164Phe) is a missense variant which affects a residue within the Runt Homology Domain (RHD) established by the MM-VCEP for RUNX1 (Val164) (PM1_supporting). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). It has a REVEL score of 0.959, which is greater than 0.88 (PP3). This variant has been reported in ClinVar; however, no phenotypic data was provided. In summary, this variant meets the criteria to be classified as a variant of uncertain significance. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: PM1_supporting, PP3, PM2_supporting.

Met criteria codes

PP3			REVEL score $\geq$ 0.88 (REVEL score=0.959)
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).
PM1_Supporting			This missense variant affects amino acid 164 which is an amino acid residue (89-204) within the RHD.

Not Met criteria codes

BS2			This rule is not applicable for MM-VCEP
BS1			PM2_supporting met
BS4			No case study found
BS3			No studies found
BP4			REVEL score is >0.5
BP3			This rule is not applicable for MM-VCEP
BP1			This rule is not applicable for MM-VCEP
BP2			No case study found
BP5			This rule is not applicable for MM-VCEP
BP7			Missense variant with no predicted splice effect

PVS1			This is not a null variant
PS1			No missense change has been at amino acid 164 has been determined to be pathogenic before
PS2			No case study found
PS3			No studies found
PS4			NO proband meeting RUNX1 FPD-MM phenotypic criteria has been described
PP1			No case study found
PP2			This rule is not applicable for MM-VCEP
PP4			This rule is not applicable for MM-VCEP
BA1			PM2_supporting met
PM3			This rule is not applicable for MM-VCEP
PM5			No missense change has been at amino acid 164 has been determined to be pathogenic before
PM4			This is a missense variant
PM6			No case study found

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.](#)