

Variant: NM_001754.5(RUNX1):c.411dup (p.Glu138Ter)

Version: 1.0

CA2739267616 [↗](#)

2820865 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3a6bb6cb-0628-40cf-bc39-2f4f9ade5014

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HGVS expressions

NM_001754.5:c.411dup

NM_001754.5(RUNX1):c.411dup (p.Glu138Ter)

NC_000021.9:g.34880654dup

CM000683.2:g.34880654dup

NC_000021.8:g.36252951dup

CM000683.1:g.36252951dup

NC_000021.7:g.35174821dup

NG_011402.2:g.1109058dup

ENST00000675419.1:c.411dup

ENST00000300305.7:c.411dup

ENST00000344691.8:c.330dup

ENST00000358356.9:c.330dup

ENST00000399237.6:c.375dup

ENST00000399240.5:c.330dup

ENST00000437180.5:c.411dup

ENST00000455571.5:c.372dup

ENST00000482318.5:c.*1dup

NM_001001890.2:c.330dup

NM_001122607.1:c.330dup

NM_001754.4:c.411dup

NM_001001890.3:c.330dup

NM_001122607.2:c.330dup

Pathogenic

Met criteria codes 4

PVS1 PM1_Supporting

PM2_Supporting PM5

Not Met criteria codes 22

BA1 BP5 BP7 BP4 BP3 BP1

BP2 BS1 BS4 BS3 BS2 PP1

PP2 PP3 PP4 PS1 PS2 PS3

PS4 PM3 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.411dup (p.Glu138Ter) is a nonsense variant which is predicted to result in nonsense-mediated mRNA decay (PVS1). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting). It is downstream of c.98 (PM5_Supporting) and impacts amino acid 138, which is located within the Runt Homology Domain (RHD) but does not occur in an established hotspot residue (PM1_Supporting). Clinical and functional data are not available for this variant. In summary, this variant meets criteria to be classified as pathogenic. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: PVS1, PM2_Supporting, PM5_Supporting, PM1_Supporting.

Met criteria codes

PVS1			This variant is a nonsense variant and is predicted to undergo NMD.
PM1_Supporting			This variant is located between AAs 89-204 and is within the RHD.
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_Supporting).
PM5			This is a nonsense/frameshift variant that is downstream of c.98.

Not Met criteria codes

BA1			This variant does not have a MAF $\geq$ 0.0015 (0.15%) in any general continental population dataset.
BP5			This rule is not applicable for MM-VCEP
BP7			This is not a silent or intronic variant
BP4			This is not a missense variant
BP3			This is not an in-frame deletion/insertion.
BP1			This rule is not applicable for MM-VCEP
BP2			This variant has not been reported.
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 is not available for this variant.

BS3			Functional studies for this variant are not available.
BS2			This rule is not applicable for the MM-VCEP.
PP1			Segregation data is not available for this variant.
PP2			This rule is not applicable for MM-VCEP
PP3			This is not a missense variant
PP4			This rule is not applicable for MM-VCEP
PS1			Pathogenic variants have not been reported at this AA.
PS2			De novo data is not available for this variant.
PS3			Functional studies for this variant are not available.
PS4			Proband data is not available for this variant.
PM3			This rule is not applicable for MM-VCEP
PM4			This is not an in-frame deletion/insertion.
PM6			De novo data is not available for this variant.

Curation History 

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