

Variant: *NM_001754.5(RUNX1):c.967+2_967+5del*

Version: 1.0

[CA645614125](#)

[945290 \(ClinVar\)](#)

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

Condition: hereditary thrombocytopenia and hematological cancer predisposition syndrome associated with RUNX1 ([MONDO:0100083](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 8fa5be74-26b6-4d6e-bb6e-ca90b6cdf97a

Approved on: 2023-12-09

Published on: 2023-12-09

HGVS expressions

NM_001754.5:c.967+2_967+5del

NM_001754.5(RUNX1):c.967+2_967+5del

NC_000021.9:g.34799299_34799302del

CM000683.2:g.34799299_34799302del

NC_000021.8:g.36171596_36171599del

CM000683.1:g.36171596_36171599del

NC_000021.7:g.35093466_35093469del

NG_011402.2:g.1190413_1190416del

ENST00000675419.1:c.967+2_967+5del

ENST00000300305.7:c.967+2_967+5del

ENST00000344691.8:c.886+2_886+5del

ENST00000399240.5:c.694+2_694+5del

ENST00000437180.5:c.967+2_967+5del

ENST00000482318.5:c.*557+2_*557+5del

NM_001001890.2:c.886+2_886+5del

NM_001754.4:c.967+2_967+5del

NM_001001890.3:c.886+2_886+5del

Pathogenic

Met criteria codes 4

PVS1_Strong **PM2_Supporting**

PP1_Strong **PS4_Moderate**

Not Met criteria codes 21

PS1 **PS2** **PS3** **BA1** **PP2** **PP3**

PP4 **PM3** **PM5** **PM4** **PM6**

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*

[Criteria Specification Approval History](#)









[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

















Myeloid Malignancy VCEP



















NM_001754.5(RUNX1):c.967+2_967+5del is a noncoding variant. This variant was found to co-segregate with disease in multiple affected family members, with seven or more (8) meioses observed in one family/across 1 family (PP1_Strong; PMID: 28240786). Skips exon 8 with in frame del270- 323 and D269A deletion in TAD(PVS1_strong). This variant has been reported in two or three probands (X) meeting at least one of the RUNX1-phenotypic criteria (PS4_Moderate; PMID: 28240786, doi: 10.1093/ajcp/aqz121.026) (PS4_moderate). This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2_supporting). 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: PP1_strong, PVS1_strong, PS4_moderate, PM2_supporting

Met criteria codes

PVS1_Strong			Skip exon 8 with in frame del270- 323 and D269A deletion in TAD
PM2_Supporting			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2).
PP1_Strong			This variant was found to co-segregate with disease in multiple affected family members, with seven or more (8) meioses observed in one family/across 1 family (PP1_Strong; PMID: 28240786).
PS4_Moderate			PS4_Moderate: This variant has been reported in two or three probands (X) meeting at least one of the RUNX1-phenotypic criteria (PS4_Moderate; PMID: 28240786, doi: 10.1093/ajcp/aqz121.026).

Not Met criteria codes

PS1			Not applicable
PS2			nil data
PS3			no data
BA1			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2).
PP2			not applicable
PP3			Not applicable
PP4			not applicable
PM3			not applicable
PM5			Not applicable
PM4			Not applicable

PM6			nil data
BS1			This variant is completely absent from all population databases with at least 20x coverage for RUNX1 (PM2).
BS4			not seen in unaffected individuals in reported families
BS3			no data
BS2			not applicable
BP5			not applicable
BP7			Not applicable
BP4			Not applicable
BP3			Not applicable
BP1			not applicable
BP2			not applicable

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.