

Variant: *NM_001754.4(RUNX1):c.*3977G>A*

Version: 1.0

[CA10650402](#)

[339799 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: da0bcbca-a4b1-4e32-a2e0-701807535636

Approved on: 2020-05-13

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HGVS expressions

NM_001754.4:c.*3977G>A

NM_001754.4(RUNX1):c.*3977G>A

NC_000021.9:g.34788158C>T

CM000683.2:g.34788158C>T

NC_000021.8:g.36160455C>T

CM000683.1:g.36160455C>T

NC_000021.7:g.35082325C>T

NG_011402.2:g.1201554G>A

ENST00000675419.1:c.*3977G>A

ENST00000300305.7:c.*3977G>A

ENST00000344691.8:c.*3977G>A

ENST00000437180.5:c.*3977G>A

NM_001001890.2:c.*3977G>A

NM_001001890.3:c.*3977G>A

NM_001754.5:c.*3977G>A

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **25**

PS1 PS2 PS3 PS4 PVS1

PP1 PP2 PP3 PP4 PM1

PM3 PM5 PM4 PM6 PM2

BS2 BS1 BS4 BS3 BP4 BP3

BP1 BP2 BP5 BP7

Evidence Links **0**

Expert Panel

[Myeloid Malignancy VCEP](#)

Criteria Specification Information

[Criteria Specifications for this VCEP](#)


Evidence submitted by expert panel

Myeloid Malignancy VCEP


The c.*3977G>A variant in the 3' UTR has an MAF of 0.003831 (0.38%, 12/3132 alleles) in the East Asian subpopulation of the gnomAD v3 cohort and is ≥ 0.0015 (0.15%) (BA1). In summary, this variant meets criteria to be classified as benign. ACMG/AMP criteria applied, as


specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BA1.


Met criteria codes


BA1  The variant is reported at the highest MAF in the East Asian population in gnomAD v3, at a frequency of 0.003831 (12/3132 alleles), with 0 homozygotes.


Not Met criteria codes


PS1  N/A


PS2  N/A


PS3  N/A


PS4  Variant meets BA1


PVS1  N/A


PP1  N/A


PP2  MM-VCEP deemed N/A for RUNX1


PP3  N/A


PP4  MM-VCEP deemed N/A for RUNX1


PM1  N/A


PM3  MM-VCEP deemed N/A for RUNX1


PM5  N/A

PM4  N/A

PM6  N/A

PM2  N/A

BS2  MM-VCEP deemed N/A for RUNX1

BS1  N/A

BS4	✘	N/A
BS3	✘	N/A
BP4	✘	N/A
BP3	✘	MM-VCEP deemed N/A for RUNX1
BP1	✘	MM-VCEP deemed N/A for RUNX1
BP2	✘	N/A
BP5	✘	MM-VCEP deemed N/A for RUNX1
BP7	✘	N/A

Curation History [↗](#)



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