

Variant: *NM_001754.5(RUNX1):c.*3580T>G*

Version: 1.0

[CA10650412](#)

[339804 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 8fc36324-ad0d-4371-aad3-4afc8cf17234

Approved on: 2025-01-15

Published on: 2025-01-15

HGVS expressions

NM_001754.5:c.*3580T>G

NM_001754.5(RUNX1):c.*3580T>G

NC_000021.9:g.34788555A>C

CM000683.2:g.34788555A>C

NC_000021.8:g.36160852A>C

CM000683.1:g.36160852A>C

NC_000021.7:g.35082722A>C

NG_011402.2:g.1201157T>G

ENST00000675419.1:c.*3580T>G

ENST00000300305.7:c.*3580T>G

ENST00000344691.8:c.*3580T>G

ENST00000437180.5:c.*3580T>G

NM_001001890.2:c.*3580T>G

NM_001754.4:c.*3580T>G

NM_001001890.3:c.*3580T>G

Likely Benign

Met criteria codes **1**

BS1

Not Met criteria codes **25**

BP4 BP3 BP1 BP2 BP5 BP7
PS1 PS2 PS3 PS4 BA1 PP1
PP2 PP3 PP4 PM1 PM3
PM5 PM4 PM6 PM2 BS2
PVS1 BS4 BS3

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.*3580T>G is a 3' UTR variant which has a MAF of 0.00057 (0.057%, 5/8716, alleles) in the African subpopulation of the 20210610 (gnomAD) cohort which is between 0.00015 (0.015%) and 0.0015 (0.15%) (BS1). In summary, this variant meets criteria to be

classified as likely benign. ACMG/AMP criteria applied, as specified by the Myeloid Malignancy Variant Curation Expert Panel for RUNX1: BS1.

Met criteria codes

BS1   MAF of 0.00057 (0.057%, 5/8716, alleles) in the African subpopulation of the 20210610 (gnomAD) cohort is between 0.00015 (0.015%) and 0.0015 (0.15%) (BS1).

Not Met criteria codes

BP4   Not applicable

BP3  Not applicable

BP1  Not applicable

BP2   Not applicable

BP5  Not applicable

BP7   Not applicable

PS1   Nil data

PS2  Nil data

PS3   Nil data

PS4   Nil data

BA1   BS1 met


PP1   Nil data

PP2  Not applicable

PP3   Not applicable

PP4  Not applicable

PM1  Not applicable

PM3		✘	Not applicable
PM5		✘	Nil data
PM4		✘	Not applicable
PM6		✘	Nil data
PM2		✘	BS1 met
BS2		✘	Not applicable
PVS1		✘	Not applicable
BS4		✘	Nil data
BS3		✘	Nil data

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.