

Variant: *NM\_001754.4(RUNX1):c.\*4032T>C*

Version: 1.0

CA10653545 [↗](#)

339797 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** 3c95e75d-735c-49cf-9bef-f6bd97b0da95

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### HGVS expressions

**NM\_001754.4:c.\*4032T>C**

**NM\_001754.4(RUNX1):c.\*4032T>C**

NC\_000021.9:g.34788103A>G

CM000683.2:g.34788103A>G

NC\_000021.8:g.36160400A>G

CM000683.1:g.36160400A>G

NC\_000021.7:g.35082270A>G

NG\_011402.2:g.1201609T>C

ENST00000675419.1:c.\*4032T>C

ENST00000300305.7:c.\*4032T>C

ENST00000344691.8:c.\*4032T>C

ENST00000437180.5:c.\*4032T>C

NM\_001001890.2:c.\*4032T>C

NM\_001001890.3:c.\*4032T>C

NM\_001754.5:c.\*4032T>C

**Benign**

#### Met criteria codes **2**

BA1 BP2

#### Not Met criteria codes **24**

BP4 BP3 BP1 BP5 BP7 BS2  
BS1 BS4 BS3 PP1 PP2 PP3  
PP4 PS1 PS2 PS3 PS4 PM1  
PM3 PM5 PM4 PM6 PM2  
PVS1

#### 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 NM\_001754.4:c.\*4032T>C variant reported at an MAF of 0.096 (9%, 835/8702 alleles) in the African population in gnomAD cohort is  $\geq 0.0015$  (0.15%) (BA1). This variant is detected in a homozygous state in 25 individuals in gnomAD (BP2). 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, BP2.

Met criteria codes

|     |                                                                                   |                                                                                   |                                                                                                                                                                       |
|-----|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| BA1 |  |  | The variant is reported in gnomAD at a frequency of 0.09595 (835/8702 alleles) in the African population, with 25 homozygotes, meeting criteria for BA1 (AF > 0.0015) |
| BP2 |  |  | The variant is reported in 25 homozygotes in gnomAD                                                                                                                   |

Not Met criteria codes

|     |                                                                                     |                                                                                     |                              |
|-----|-------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------|------------------------------|
| BP4 |    |    | N/A                          |
| BP3 |                                                                                     |    | MM-VCEP deemed N/A for RUNX1 |
| BP1 |                                                                                     |    | MM-VCEP deemed N/A for RUNX1 |
| BP5 |                                                                                     |    | MM-VCEP deemed N/A for RUNX1 |
| BP7 |  |  | N/A                          |
| BS2 |                                                                                     |  | MM-VCEP deemed N/A for RUNX1 |
| BS1 |  |  | Variant meets BA1            |
| BS4 |  |  | No data currently available  |
| BS3 |  |  | No data currently available  |
| PP1 |  |  | No data currently available  |
| PP2 |                                                                                     |  | MM-VCEP deemed N/A for RUNX1 |
| PP3 |  |  | N/A                          |
| PP4 |                                                                                     |  | MM-VCEP deemed N/A for RUNX1 |
| PS1 |  |  | N/A                          |
| PS2 |                                                                                     |  | No data currently available  |

|      |                                                                                   |   |                                                                                                                                                                                |
|------|-----------------------------------------------------------------------------------|---|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PS3  |    | ✖ | No data currently available                                                                                                                                                    |
| PS4  |  | ✖ | The variant has not been reported in patients with familial platelet disorder with predisposition to hematologic malignancies in the literature, to the best of our knowledge. |
| PM1  |                                                                                   | ✖ | N/A                                                                                                                                                                            |
| PM3  |                                                                                   | ✖ | MM-VCEP deemed N/A for RUNX1                                                                                                                                                   |
| PM5  |  | ✖ | N/A                                                                                                                                                                            |
| PM4  |  | ✖ | N/A                                                                                                                                                                            |
| PM6  |  | ✖ | No data currently available                                                                                                                                                    |
| PM2  |                                                                                   | ✖ | Variant meets BA1                                                                                                                                                              |
| PVS1 |  | ✖ | N/A                                                                                                                                                                            |

| Curation History  |  |  |
|------------------------------------------------------------------------------------------------------|--|--|
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