The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

*Variant: NM_001754.4(RUNX1):c.*3721A>T*

CA10650410

339801 (ClinVar)

Gene: RUNX1 (HGNC:861)
Condition: hereditary thrombocytopenia and hematologic cancer predisposition syndrome (MONDO:0011071)
Inheritance Mode: Autosomal dominant inheritance
UUID: d6249ec6-8bee-41b0-825c-9d9d2d30ddac
Approved on: 2020-05-13
Published on: 2020-06-02

HGVS expressions

NM_001754.4:c.*3721A>T
NM_001754.4(RUNX1):c.*3721A>T
NC_000021.9:g.34788414T>A
CM000683.2:g.34788414T>A
NC_000021.8:g.36160711T>A
CM000683.1:g.36160711T>A
NC_000021.7:g.35082581T>A
NG_011402.2:g.1201298A>T
NM_001001890.2:c.*3721A>T
NM_001001890.3:c.*3721A>T
ENST00000300305.7:c.*3721A>T
ENST00000344691.8:c.*3721A>T
ENST00000437180.5:c.*3721A>T

Benign

Met criteria codes 2

BP2 BA1

Not Met criteria codes 16

BS1 BS3 BS4 BP4 BP7 PS1 PS3 PS4 PP3 PP1 PM5 PM4 PM1 PVS1 PM2 PM6

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.*3721A>T variant in the 3' UTR has an MAF of 0.001757 (0.18%, 24/13660 alleles) in the Latino subpopulation of the gnomAD v3 cohort and is ≥ 0.0015 (0.15%) (BA1). This variant is detected in a homozygous state in 8 Amish individuals in the gnomAD v3 population database (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

BP2	✔	8 Amish homozygous individuals in gnomAD v3.
BA1	✔	The variant is reported at the highest MAF in the Latino population in gnomAD v3, at a frequency of 0.001757 (24/13660 alleles).

Not Met criteria codes

BS1	✖	Variant meets BA1
BS3	✖	N/A
BS4	✖	N/A
BP4	✖	N/A
BP7	✖	N/A
PS1	✖	N/A
PS3	✖	N/A
PS4	✖	Variant meets BA1
PP3	✖	N/A
PP1	✖	N/A
PM5	✖	N/A
PM4	✖	N/A
PM1	✖	N/A
PVS1	✖	N/A
PM2	✖	Variant meets BA1
PM6	✖	N/A

Curation History

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene

No matching records found