

Variant: *NM_001754.5(RUNX1):c.496C>A (p.Arg166=)*

Version: 1.0

CA10014502 [↗](#)

570036 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 78d5219e-fd6a-41aa-ad83-9dcf935a1544

Approved on: 2022/8/23

Published on: 2023/11/13

HGVS expressions

NM_001754.5:c.496C>A

NM_001754.5(RUNX1):c.496C>A (p.Arg166=)

NC_000021.9:g.34880569G>T

CM000683.2:g.34880569G>T

NC_000021.8:g.36252866G>T

CM000683.1:g.36252866G>T

NC_000021.7:g.35174736G>T

NG_011402.2:g.1109143C>A

ENST00000675419.1:c.496C>A

ENST00000300305.7:c.496C>A

ENST00000344691.8:c.415C>A

ENST00000358356.9:c.415C>A

ENST00000399237.6:c.460C>A

ENST00000399240.5:c.415C>A

ENST00000437180.5:c.496C>A

ENST00000482318.5:c.*86C>A

NM_001001890.2:c.415C>A

NM_001122607.1:c.415C>A

NM_001754.4:c.496C>A

NM_001001890.3:c.415C>A

NM_001122607.2:c.415C>A

Likely Benign

Met criteria codes **2**

BP4 BP7

Not Met criteria codes **24**

PS1 PS2 PS3 PS4 BP3 BP1
BP2 BP5 PP1 PP2 PP3 PP4
BA1 PM6 PM2 PM1 PM3
PM5 PM4 BS2 BS1 BS4
BS3 PVS1

Evidence Links **0**

Expert Panel

Myeloid Malignancy VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Myeloid Malignancy VCEP

NM_001754.5(RUNX1):c.496C>A (p.Arg166=) is a synonymous variant. Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 1.6 < 2.0 or the variant is the reference nucleotide in one primate and/or three mammal species) (BP7). Synonymous variant therefore no REVEL score and SpliceAI is ≤ 0.20 (0.14) (BP4). 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: BP4, BP7.

Met criteria codes

BP4			Synonymous variant therefore no REVEL score and SpliceAI is ≤ 0.20 (0.14) (BP4)
BP7			Evolutionary conservation prediction algorithms predict the site as not being conserved (PhyloP score 1.6 < 2.0 or the variant is the reference nucleotide in one primate and/or three mammal species) (BP7).

Not Met criteria codes

PS1			No known established pathogenic variants at this codon
PS2			nil data
PS3			nil data
PS4			nil data
BP3			synonymous variant
BP1			not applicable
BP2			not applicable
BP5			not applicable
PP1			nil data
PP2			not applicable
PP3			Synonymous variant therefore no REVEL score and SpliceAI is not ≥0.38 (0.14)
PP4			not applicable

BA1			AF 3.98e-6 gnomAD V2
PM6			nil data
PM2			AF 3.98e-6 gnomAD V2
PM1			Not a missense variant
PM3			not applicable
PM5			No known established pathogenic variants at this codon
PM4			synonymous variant
BS2			not applicable
BS1			AF 3.98e-6 gnomAD V2
BS4			nil data
BS3			nil data
PVS1			synonymous variant not involving splice site

Curation History 		
    		
Showing 1 to 1 of 1 rows		

