

Variant: NM_002880.3(RAF1):c.125C>T (p.Ala42Val)

CA2259832 [↗](#)

343103 (ClinVar) [↗](#)

Gene: RAF1 ([HGNC:5894](#))

Condition: RASopathy ([MONDO:0021060](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: da2fd72c-d28d-424b-92b6-eb26ccf60d9f

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Published on: 2018-12-10

HGVS expressions

NM_002880.3:c.125C>T

NM_002880.3(RAF1):c.125C>T (p.Ala42Val)

NM_001354689.1:c.125C>T

NM_001354690.1:c.125C>T

NM_001354691.1:c.-6C>T

NM_001354692.1:c.-6C>T

NM_001354693.1:c.125C>T

NM_001354694.1:c.-6C>T

NM_001354695.1:c.-6C>T

NR_148940.1:n.540C>T

NR_148941.1:n.540C>T

NR_148942.1:n.540C>T

ENST00000251849.8:c.125C>T

ENST00000416093.1:c.125C>T

ENST00000423275.5:c.125C>T

ENST00000442415.6:c.125C>T

NC_000003.12:g.12618597G>A

CM000665.2:g.12618597G>A

NC_000003.11:g.12660096G>A

CM000665.1:g.12660096G>A

NC_000003.10:g.12635096G>A

NG_007467.1:g.50583C>T

Likely Benign

The Expert Panel has overridden the computationally generated classification - "Uncertain Significance - Insufficient Evidence"

Met criteria codes **1**

BS1

Evidence Links **0**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

RASopathy VCEP

The filtering allele frequency of the c.125C>T (p.Ala42Val) variant in the RAF1 gene is 0.026% (25/66738) of European chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BS1; PMID:29493581)

Met criteria codes

BS1



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Curation History [↗](#)

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Showing 1 to 1 of 1 rows

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