

Variant: NM_002880.3(RAF1):c.1941C>T (p.Val647=)

CA134718 [↗](#)

44623 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 62438d26-e85a-41a6-ae0f-89550c868512

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HGVS expressions

NM_002880.3(RAF1):c.1941C>T (p.Val647=)

NM_002880.3:c.1941C>T

NC_000003.12:g.12584520G>A

CM000665.2:g.12584520G>A

NC_000003.11:g.12626019G>A

CM000665.1:g.12626019G>A

NC_000003.10:g.12601019G>A

NG_007467.1:g.84660C>T

NM_001354689.1:c.2001C>T

NM_001354690.1:c.1941C>T

NM_001354691.1:c.1698C>T

NM_001354692.1:c.1698C>T

NM_001354693.1:c.1842C>T

NM_001354694.1:c.1758C>T

NM_001354695.1:c.1599C>T

NR_148940.1:n.2469C>T

NR_148941.1:n.2415C>T

NR_148942.1:n.2354C>T

ENST00000251849.8:c.1941C>T

ENST00000423275.5:c.*1618C>T

ENST00000432427.2:n.1578C>T

ENST00000442415.6:c.2001C>T

ENST00000471449.1:n.630C>T

Benign

Met criteria codes **1**

BA1

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.1941C>T (p.Val647=) variant in the RAF1 gene is 1.04% (738/66732) of European chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Met criteria codes

BA1			The filtering allele frequency of the c.1941C>T (p.Val647=) variant in the RAF1 gene is 1.04% (738/66732) of European chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)
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Curation History



 

 

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