

Variant: NM_002880.3(RAF1):c.923C>T (p.Pro308Leu)

CA134762 [↗](#)

40611 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: ed484574-9b2a-4dda-bd46-ea935a45f83c

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HGVS expressions

NM_002880.3:c.923C>T

NM_002880.3(RAF1):c.923C>T (p.Pro308Leu)

NC_000003.12:g.12600219G>A

CM000665.2:g.12600219G>A

NC_000003.11:g.12641718G>A

CM000665.1:g.12641718G>A

NC_000003.10:g.12616718G>A

NG_007467.1:g.68961C>T

NM_001354689.1:c.983C>T

NM_001354690.1:c.923C>T

NM_001354691.1:c.680C>T

NM_001354692.1:c.680C>T

NM_001354693.1:c.824C>T

NM_001354694.1:c.740C>T

NM_001354695.1:c.581C>T

NR_148940.1:n.1338C>T

NR_148941.1:n.1338C>T

NR_148942.1:n.1338C>T

ENST00000251849.8:c.923C>T

ENST00000423275.5:c.*600C>T

ENST00000432427.2:n.560C>T

ENST00000442415.6:c.983C>T

ENST00000465826.5:n.167C>T

ENST00000491290.1:n.552C>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.923C>T (p.Pro308Leu) variant in the RAF1 gene is **1.516% (179/10394)** of African 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.923C>T (p.Pro308Leu) variant in the RAF1 gene is 1.516% (179/10394) of African 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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