

Variant: NM_030662.3(MAP2K2):c.893C>T (p.Pro298Leu)

CA180870 [↗](#)

40832 (ClinVar) [↗](#)

Gene: MAP2K2 ([HGNC:5605](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 40ceccb2-5456-4905-8d02-1878c6fe945f

Approved on: 2017-05-09

Published on: 2018-12-10

HGVS expressions

NM_030662.3:c.893C>T

NM_030662.3(MAP2K2):c.893C>T (p.Pro298Leu)

ENST00000262948.9:c.893C>T

ENST00000394867.8:c.602C>T

ENST00000593364.5:n.840C>T

ENST00000595715.1:n.708C>T

ENST00000597263.5:n.169+1792C>T

ENST00000599021.1:n.29+1792C>T

ENST00000600584.5:n.1453C>T

ENST00000601786.5:n.1194C>T

NC_000019.10:g.4099227G>A

CM000681.2:g.4099227G>A

NC_000019.9:g.4099225G>A

CM000681.1:g.4099225G>A

NC_000019.8:g.4050225G>A

NG_007996.1:g.29902C>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.893C>T (p.Pro298Leu) variant in the MAP2K2 gene is 0.953% (68/5776) of East Asian 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

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Curation History

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