

Variant: NM_030662.3(MAP2K2):c.1194C>T (p.Thr398=)

CA137913 [↗](#)

40846 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4dda494b-f286-41e8-b20f-481268ec7d54

Approved on: 2017-05-09

Published on: 2018-12-10

HGVS expressions

NM_030662.3(MAP2K2):c.1194C>T (p.Thr398=)

NM_030662.3:c.1194C>T

NC_000019.10:g.4090607G>A

CM000681.2:g.4090607G>A

NC_000019.9:g.4090605G>A

CM000681.1:g.4090605G>A

NC_000019.8:g.4041605G>A

NG_007996.1:g.38522C>T

ENST00000262948.9:c.1194C>T

ENST00000394867.8:c.903C>T

ENST00000597263.5:n.379C>T

ENST00000599021.1:n.304C>T

ENST00000600584.5:n.2643C>T

ENST00000601786.5:n.1495C>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.1194C>T (p.Thr398=) variant in the MAP2K2 gene is 0.159% (19/7836) 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.1194C>T (p.Thr398=) variant in the MAP2K2 gene is 0.159% (19/7836) 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)

Curation History



 ▾

 ▾

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.