

Variant: NM_030662.3(MAP2K2):c.813C>T (p.Asp271=)

CA9090826 [↗](#)

378108 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 241fca61-876f-4b2a-bc64-30fd49bd7ee2

Approved on: 2017-05-09

Published on: 2018-12-10

HGVS expressions

NM_030662.3:c.813C>T

NM_030662.3(MAP2K2):c.813C>T (p.Asp271=)

ENST00000262948.9:c.813C>T

ENST00000394867.8:c.522C>T

ENST00000593364.5:n.760C>T

ENST00000595715.1:n.628C>T

ENST00000597263.5:n.169+1712C>T

ENST00000599021.1:n.29+1712C>T

ENST00000600584.5:n.1373C>T

ENST00000601786.5:n.1114C>T

NC_000019.10:g.4099307G>A

CM000681.2:g.4099307G>A

NC_000019.9:g.4099305G>A

CM000681.1:g.4099305G>A

NC_000019.8:g.4050305G>A

NG_007996.1:g.29822C>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.813C>T (p.Asp271=) variant in the MAP2K2 gene is 0.0045% (1/7096) of Latino 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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✓

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Curation History [↗](#)

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