

Variant: NM_030662.3(MAP2K2):c.818A>G (p.Lys273Arg)

CA296139 [↗](#)

40824 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 905a415a-6df7-4dab-b492-c7ae4a886f9f

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HGVS expressions

NM_030662.3(MAP2K2):c.818A>G (p.Lys273Arg)

NM_030662.3:c.818A>G

ENST00000262948.9:c.818A>G

ENST00000394867.8:c.527A>G

ENST00000593364.5:n.765A>G

ENST00000595715.1:n.633A>G

ENST00000597263.5:n.169+1717A>G

ENST00000599021.1:n.29+1717A>G

ENST00000600584.5:n.1378A>G

ENST00000601786.5:n.1119A>G

NC_000019.10:g.4099302T>C

CM000681.2:g.4099302T>C

NC_000019.9:g.4099300T>C

CM000681.1:g.4099300T>C

NC_000019.8:g.4050300T>C

NG_007996.1:g.29827A>G

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.818A>G (p.Lys273Arg) variant in the MAP2K2 gene is 0.0368% (6/7088) 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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Curation History [↗](#)

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