

Variant: NM_030662.3(MAP2K2):c.240G>A (p.Ala80=)

CA9091070 [↗](#)

378913 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 63dda72c-31c5-4e8f-b694-d44b4e9c8d51

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HGVS expressions

NM_030662.3:c.240G>A

NM_030662.3(MAP2K2):c.240G>A (p.Ala80=)

NC_000019.10:g.4117482C>T

CM000681.2:g.4117482C>T

NC_000019.9:g.4117480C>T

CM000681.1:g.4117480C>T

NC_000019.8:g.4068480C>T

NG_007996.1:g.11647G>A

ENST00000262948.9:c.240G>A

ENST00000394867.8:c.-52G>A

ENST00000599345.1:n.437G>A

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.240G>A (p.Ala80=) variant in the MAP2K2 gene is 0.0284% (9/16506) of South Asian 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			The filtering allele frequency of the c.240G>A (p.Ala80=) variant in the MAP2K2 gene is 0.0284% (9/16506) of South Asian 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)
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Curation History



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