

Variant: NM_030662.3(MAP2K2):c.192C>T (p.Val64=)

CA137916 [↗](#)

40770 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: cf89340c-b64c-46a6-aef4-094850f57f25

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HGVS expressions

NM_030662.3(MAP2K2):c.192C>T (p.Val64=)

NM_030662.3:c.192C>T

NC_000019.10:g.4117530G>A

CM000681.2:g.4117530G>A

NC_000019.9:g.4117528G>A

CM000681.1:g.4117528G>A

NC_000019.8:g.4068528G>A

NG_007996.1:g.11599C>T

ENST00000262948.9:c.192C>T

ENST00000394867.8:c.-100C>T

ENST00000599345.1:n.389C>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.192C>T (p.Val64=) variant in the MAP2K2 gene is 7.417% (1283/16512) of South 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			The filtering allele frequency of the c.192C>T (p.Val64=) variant in the MAP2K2 gene is 7.417% (1283/16512) of South 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)
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Curation History



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