

Variant: NM_030662.3(MAP2K2):c.1020C>T (p.Pro340=)

CA137906 [↗](#)

46226 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: ce8d4a0e-fe41-460b-91e2-0b5c99377b0a

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HGVS expressions

NM_030662.3(MAP2K2):c.1020C>T (p.Pro340=)

NM_030662.3:c.1020C>T

ENST00000262948.9:c.1020C>T

ENST00000394867.8:c.729C>T

ENST00000595715.1:n.835C>T

ENST00000597263.5:n.205C>T

ENST00000599021.1:n.130C>T

ENST00000600584.5:n.1580C>T

ENST00000601786.5:n.1321C>T

NC_000019.10:g.4095414G>A

CM000681.2:g.4095414G>A

NC_000019.9:g.4095412G>A

CM000681.1:g.4095412G>A

NC_000019.8:g.4046412G>A

NG_007996.1:g.33715C>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.1020C>T (p.Pro340=) variant in the MAP2K2 gene is 0.262% (29/7910) 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





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Curation History [↗](#)

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