

Variant: NM_030662.3(MAP2K2):c.525C>T (p.Ile175=)

CA137952 [↗](#)

46239 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 5a6be788-3cc6-4e54-b10f-96366d0767d5

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HGVS expressions

NM_030662.3(MAP2K2):c.525C>T (p.Ile175=)

NM_030662.3:c.525C>T

NC_000019.10:g.4102379G>A

CM000681.2:g.4102379G>A

NC_000019.9:g.4102377G>A

CM000681.1:g.4102377G>A

NC_000019.8:g.4053377G>A

NG_007996.1:g.26750C>T

ENST00000262948.9:c.525C>T

ENST00000394867.8:c.234C>T

ENST00000593364.5:n.472C>T

ENST00000597008.5:n.126C>T

ENST00000599345.1:n.795C>T

ENST00000601786.5:n.826C>T

ENST00000602167.5:n.245C>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.525C>T (p.Ile175=) variant in the MAP2K2 gene is 0.255% (11/2418) of Latino 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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