

Variant: NM_030662.3(MAP2K2):c.690G>A (p.Thr230=)

CA291996 [↗](#)

138159 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 76788912-f5e9-4ff4-8343-c7b0c152c0b2

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HGVS expressions

NM_030662.3(MAP2K2):c.690G>A (p.Thr230=)

NM_030662.3:c.690G>A

ENST00000262948.9:c.690G>A

ENST00000394867.8:c.399G>A

ENST00000593364.5:n.637G>A

ENST00000597008.5:n.291G>A

ENST00000597263.5:n.154G>A

ENST00000599021.1:n.14G>A

ENST00000601786.5:n.991G>A

ENST00000602167.5:n.410G>A

NC_000019.10:g.4101034C>T

CM000681.2:g.4101034C>T

NC_000019.9:g.4101032C>T

CM000681.1:g.4101032C>T

NC_000019.8:g.4052032C>T

NG_007996.1:g.28095G>A

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.690G>A (p.Thr230=) variant in the MAP2K2 gene is 0.1273% for Latino chromosomes by the Exome Aggregation Consortium (3/642 with 95% CI), which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert panel for autosomal dominant RASopathy variants (BA1).

Met criteria codes

BA1

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Curation History [↗](#)

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