

Variant: NM_030662.3(MAP2K2):c.1162C>T (p.Arg388Trp)

CA180895 [↗](#)

40843 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: a8067cbf-1dd7-4aab-bdb7-dfb3edfdaedf

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HGVS expressions

NM_030662.3:c.1162C>T

NM_030662.3(MAP2K2):c.1162C>T (p.Arg388Trp)

NC_000019.10:g.4090639G>A

CM000681.2:g.4090639G>A

NC_000019.9:g.4090637G>A

CM000681.1:g.4090637G>A

NC_000019.8:g.4041637G>A

NG_007996.1:g.38490C>T

ENST00000262948.9:c.1162C>T

ENST00000394867.8:c.871C>T

ENST00000597263.5:n.347C>T

ENST00000599021.1:n.272C>T

ENST00000600584.5:n.2611C>T

ENST00000601786.5:n.1463C>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.1162C>T (p.Arg388Trp) variant in the MAP2K2 gene is 0.125% for African chromosomes by the Exome Aggregation Consortium (25/2788 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). Additional case-level data available: SCV000204180.4; SCV000252871.4.

Met criteria codes

BA1





The filtering allele frequency of the c.1162C>T (p.Arg388Trp) variant in the MAP2K2 gene is 0.125% for ETHNICITY chromosomes by the Exome Aggregation Consortium (25/2788 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).

Curation History [↗](#)

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