

Variant: NM_030662.3(MAP2K2):c.846C>T (p.Pro282=)

CA137975 [↗](#)

40827 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3a3c6e6a-a7bb-47bf-afb2-aacf5127c6fd

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HGVS expressions

NM_030662.3:c.846C>T

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NC_000019.10:g.4099274G>A

CM000681.2:g.4099274G>A

NC_000019.9:g.4099272G>A

CM000681.1:g.4099272G>A

NC_000019.8:g.4050272G>A

NG_007996.1:g.29855C>T

ENST00000262948.9:c.846C>T

ENST00000394867.8:c.555C>T

ENST00000593364.5:n.793C>T

ENST00000595715.1:n.661C>T

ENST00000597263.5:n.169+1745C>T

ENST00000599021.1:n.29+1745C>T

ENST00000600584.5:n.1406C>T

ENST00000601786.5:n.1147C>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.846C>T (p.Pro282=) variant in the MAP2K2 gene is 0.515% (46/6886) 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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