

Variant: NM_030662.3(MAP2K2):c.141C>T (p.Asp47=)

CA9091086 [↗](#)

448953 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 39270fbb-deac-4d6c-b2ab-d39741932713

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HGVS expressions

NM_030662.3:c.141C>T

NM_030662.3(MAP2K2):c.141C>T (p.Asp47=)

NC_000019.10:g.4117581G>A

CM000681.2:g.4117581G>A

NC_000019.9:g.4117579G>A

CM000681.1:g.4117579G>A

NC_000019.8:g.4068579G>A

NG_007996.1:g.11548C>T

ENST00000262948.9:c.141C>T

ENST00000394867.8:c.-151C>T

ENST00000599345.1:n.338C>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.141C>T (p.Asp47=) variant in the MAP2K2 gene is 0.054% (9/8646) of East 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			The filtering allele frequency of the c.141C>T (p.Asp47=) variant in the MAP2K2 gene is 0.054% (9/8646) of East 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)
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Curation History



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