

*Variant: NM\_030662.3(MAP2K2):c.420C>T (p.Asp140=)*

CA137938 [↗](#)

46235 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** dc9fdbbf-4e28-41d1-96de-532d635a2b43

**Approved on:** 2017-05-09

**Published on:** 2018-12-10

### *HGVS expressions*

**NM\_030662.3:c.420C>T**

**NM\_030662.3(MAP2K2):c.420C>T (p.Asp140=)**

NC\_000019.10:g.4110539G>A

CM000681.2:g.4110539G>A

NC\_000019.9:g.4110537G>A

CM000681.1:g.4110537G>A

NC\_000019.8:g.4061537G>A

NG\_007996.1:g.18590C>T

ENST00000262948.9:c.420C>T

ENST00000394867.8:c.129C>T

ENST00000599345.1:n.617C>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.420C>T (p.Asp140=) variant in the MAP2K2 gene is 0.271% (32/8604) 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**

|            |                                                                                   |                                                                                   |                                                                                                                                                                                                                                                                                                                             |
|------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>BA1</b> |  |  | The filtering allele frequency of the c.420C>T (p.Asp140=) variant in the MAP2K2 gene is 0.271% (32/8604) 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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