

Variant: NM_030662.3(MAP2K2):c.1140C>T (p.Ala380=)

CA143886 [↗](#)

50938 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b23e5835-5f83-4a46-b686-0e354d53e944

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HGVS expressions

NM_030662.3(MAP2K2):c.1140C>T (p.Ala380=)

NM_030662.3:c.1140C>T

ENST00000262948.9:c.1140C>T

ENST00000394867.8:c.849C>T

ENST00000597263.5:n.325C>T

ENST00000599021.1:n.250C>T

ENST00000600584.5:n.2589C>T

ENST00000601786.5:n.1441C>T

NC_000019.10:g.4090661G>A

CM000681.2:g.4090661G>A

NC_000019.9:g.4090659G>A

CM000681.1:g.4090659G>A

NC_000019.8:g.4041659G>A

NG_007996.1:g.38468C>T

Likely Benign

The Expert Panel has overridden the computationally generated classification - "Uncertain Significance - Insufficient Evidence"

Met criteria codes 1

BS1

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.1140C>T (p.Ala380=) variant in the MAP2K2 gene is 0.003% (2/796) of East Asian chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BS1; PMID:29493581)

Met criteria codes

BS1



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Curation History



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