

Variant: NM_002755.3(MAP2K1):c.1098T>C (p.Ala366=)

CA7624163 [↗](#)

448949 (ClinVar) [↗](#)

Gene: MAP2K1 ([HGNC:5604](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9581cc43-8005-4a02-8e31-2c7cb18c67df

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HGVS expressions

NM_002755.3:c.1098T>C

NM_002755.3(MAP2K1):c.1098T>C (p.Ala366=)

NC_000015.10:g.66490531T>C

CM000677.2:g.66490531T>C

NC_000015.9:g.66782869T>C

CM000677.1:g.66782869T>C

NC_000015.8:g.64569923T>C

NG_008305.1:g.108659T>C

NG_051234.1:g.12285A>G

NM_006049.2:c.*208A>G

NM_006049.3:c.*208A>G

NR_138061.1:n.727A>G

ENST00000307102.9:c.1098T>C

ENST00000395589.6:c.*208A>G

ENST00000563480.6:c.*208A>G

ENST00000566326.1:c.570T>C

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.1098T>C (p.Ala366=) variant in the MAP2K1 gene is 0.0459% (8/8654) 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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