

Variant: *NM_002755.3(MAP2K1):c.69C>T (p.Thr23=)*

CA181309 [↗](#)

138155 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3eacc550-bf0c-432b-b902-eee2cb07e8c4

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HGVS expressions

NM_002755.3(MAP2K1):c.69C>T (p.Thr23=)

NM_002755.3:c.69C>T

ENST00000307102.9:c.69C>T

ENST00000425818.2:n.580C>T

NC_000015.10:g.66387416C>T

CM000677.2:g.66387416C>T

NC_000015.9:g.66679754C>T

CM000677.1:g.66679754C>T

NC_000015.8:g.64466808C>T

NG_008305.1:g.5544C>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.69C>T (p.Thr23=) variant in the MAP2K1 gene is 0.0484% (4/2820) of African 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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