

Variant: NM_002755.3(MAP2K1):c.1137C>T (p.Ile379=)

CA134592 [↗](#)

40762 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: bb9af7db-7756-46f9-88be-f7ca17304539

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HGVS expressions

NM_002755.3(MAP2K1):c.1137C>T (p.Ile379=)

NM_002755.3:c.1137C>T

NM_006049.2:c.*169G>A

NM_006049.3:c.*169G>A

NR_138061.1:n.688G>A

ENST00000307102.9:c.1137C>T

ENST00000395589.6:c.*169G>A

ENST00000563480.6:c.*169G>A

ENST00000566326.1:c.609C>T

NC_000015.10:g.66490570C>T

CM000677.2:g.66490570C>T

NC_000015.9:g.66782908C>T

CM000677.1:g.66782908C>T

NC_000015.8:g.64569962C>T

NG_008305.1:g.108698C>T

NG_051234.1:g.12246G>A

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.1137C>T (p.Ile379=) variant in the MAP2K1 gene is 0.175% (26/10406) of African 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

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Curation History [↗](#)

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