

Variant: *NM_002755.3(MAP2K1):c.648C>T (p.Ile216=)*

CA134607 [↗](#)

44590 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4e4166b8-f343-4e54-a3e5-aab6290a5355

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HGVS expressions

NM_002755.3(MAP2K1):c.648C>T (p.Ile216=)

NM_002755.3:c.648C>T

ENST00000307102.9:c.648C>T

ENST00000566326.1:c.120C>T

NC_000015.10:g.66481834C>T

CM000677.2:g.66481834C>T

NC_000015.9:g.66774172C>T

CM000677.1:g.66774172C>T

NC_000015.8:g.64561226C>T

NG_008305.1:g.99962C>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.648C>T (p.Ile216=) variant in the MAP2K1 gene is 0.274% (38/10402) 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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