

Variant: NM_002755.3(MAP2K1):c.804C>G (p.Ala268=)

CA134619 [↗](#)

44592 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 240b9c1e-16ca-4644-9dfa-ec5011367f68

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HGVS expressions

NM_002755.3(MAP2K1):c.804C>G (p.Ala268=)

NM_002755.3:c.804C>G

ENST00000307102.9:c.804C>G

ENST00000566326.1:c.276C>G

NC_000015.10:g.66485100C>G

CM000677.2:g.66485100C>G

NC_000015.9:g.66777438C>G

CM000677.1:g.66777438C>G

NC_000015.8:g.64564492C>G

NG_008305.1:g.103228C>G

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.804C>G (p.Ala268=) variant in the MAP2K1 gene is 1.234% (228/16512) of South Asian 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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