

Variant: NM_002755.3(MAP2K1):c.315C>T (p.Pro105=)

CA134604 [↗](#)

44589 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: f18e6c0b-7a6f-4a7e-82d7-5de48e915d3c

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HGVS expressions

NM_002755.3:c.315C>T

NM_002755.3(MAP2K1):c.315C>T (p.Pro105=)

ENST00000307102.9:c.315C>T

ENST00000425818.2:n.826C>T

NC_000015.10:g.66436769C>T

CM000677.2:g.66436769C>T

NC_000015.9:g.66729107C>T

CM000677.1:g.66729107C>T

NC_000015.8:g.64516161C>T

NG_008305.1:g.54897C>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.315C>T (p.Pro105=) variant in the MAP2K1 gene is 0.01% (64/66728) of European 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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