

Variant: NM_002755.3(MAP2K1):c.927A>T (p.Ala309=)

CA180933 [↗](#)

40758 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: a895f2eb-4350-485d-b678-a8f94a4cc002

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HGVS expressions

NM_002755.3(MAP2K1):c.927A>T (p.Ala309=)

NM_002755.3:c.927A>T

ENST00000307102.9:c.927A>T

ENST00000566326.1:c.399A>T

NC_000015.10:g.66487259A>T

CM000677.2:g.66487259A>T

NC_000015.9:g.66779597A>T

CM000677.1:g.66779597A>T

NC_000015.8:g.64566651A>T

NG_008305.1:g.105387A>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.927A>T (p.Ala309=) variant in the MAP2K1 gene is 1.307% (156/10406) 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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