

Variant: NM_002755.3(MAP2K1):c.848C>T (p.Ala283Val)

CA134622 [↗](#)

40756 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d798c554-02fa-4c7b-96d8-4fda7463f9de

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HGVS expressions

NM_002755.3(MAP2K1):c.848C>T (p.Ala283Val)

NM_002755.3:c.848C>T

NC_000015.10:g.66485144C>T

CM000677.2:g.66485144C>T

NC_000015.9:g.66777482C>T

CM000677.1:g.66777482C>T

NC_000015.8:g.64564536C>T

NG_008305.1:g.103272C>T

ENST00000307102.9:c.848C>T

ENST00000566326.1:c.320C>T

Benign

Met criteria codes **2**

BA1

BP5

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.848C>T (p.Ala283Val) variant in the MAP2K1 gene is 0.052% for African chromosomes by the Exome Aggregation Consortium (10/10366 with 95% CI), which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert panel for autosomal dominant RASopathy variants (BA1). This variant has been identified in a patient with an alternate molecular basis for disease (BP5; Partners LMM, GeneDx internal data; GTR ID: 21766, 26957; ClinVar SCV000207935.12; SCV000061262.5). In summary, this variant meets criteria to be classified as benign. RASopathy-specific ACMG/AMP criteria applied (PMID:29493581): BA1, BP5.

Met criteria codes

BA1



0.096% (10/10366) in Africans (ExAC)

BP5

SCV000207935; SCV000061262

Curation History [↗](#)

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