

Variant: NM_030662.4(MAP2K2):c.844C>T (p.Pro282Ser)

CA137972 [↗](#)

40826 (ClinVar) [↗](#)

Gene: MAP2K2 ([HGNC:5605](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 413325d3-6867-4b66-a6a6-655f337675bc

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HGVS expressions

NM_030662.4:c.844C>T

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NC_000019.10:g.4099276G>A

CM000681.2:g.4099276G>A

NC_000019.9:g.4099274G>A

CM000681.1:g.4099274G>A

NC_000019.8:g.4050274G>A

NG_007996.1:g.29853C>T

ENST00000394867.9:n.1283C>T

ENST00000687128.1:n.1283C>T

ENST00000688002.1:n.1138C>T

ENST00000689792.1:n.748C>T

ENST00000262948.10:c.844C>T

ENST00000262948.9:c.844C>T

ENST00000394867.8:c.553C>T

ENST00000593364.5:n.791C>T

ENST00000595715.1:n.659C>T

ENST00000597263.5:n.169+1743C>T

ENST00000599021.1:c.29+1743C>T

ENST00000600584.5:n.1404C>T

ENST00000601786.5:n.1145C>T

NM_030662.3:c.844C>T

Benign

Met criteria codes **3**

BA1 **BP4** **BP5**

Not Met criteria codes **3**

BS1 **PP3** **PM2**

Evidence Links **0**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen RASopathy Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines for MAP2K2 Version 2.3.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

RASopathy VCEP

The c.844C>T variant in the MAP2K2 gene is a missense variant predicted to cause substitution of proline by serine at amino acid 282 (p.Pro282Ser). The filtering allele frequency in gnomAD v2.1.1 is 0.001415 (42/22756 alleles) in the African/African American population, meeting the criterion for BA1. The computational predictor REVEL gives a score of 0.072, which predicts no impact on protein function (BP4). This variant has been identified in a patient with an alternate molecular basis for disease (BP5; GeneDx internal data; GTR ID's 26957; SCV000207963.8). In summary, this variant meets criteria to be classified as benign for autosomal dominant RASopathies based on the ACMG/AMP criteria applied, as specified by the ClinGen RASopathy Variant Curation Expert Panel: BA1, BP4, BP5 (Specification Version 2.3, 12/3/2024)

Met criteria codes

BA1			The filtering allele frequency in gnomAD v2.1.1 is 0.001415 (42/22756 alleles) in the African/African American population, 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).
BP4			The computational predictor REVEL gives a score of 0.072, which predicts no impact on protein function
BP5			This variant has been identified in a patient with an alternate molecular basis for disease (BP5; GeneDx internal data; GTR ID's 26957; SCV000207963.8).

Not Met criteria codes

BS1			The filtering allele frequency in gnomAD v2.1.1 is 0.001415 (42/22756 alleles) in the African/African American population, meeting BA1
PP3			The computational predictor REVEL gives a score of 0.072, which predicts no impact on protein function
PM2			The filtering allele frequency in gnomAD v2.1.1 is 0.001415 (42/22756 alleles) in the African/African American population, meeting BA1



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