

Variant: NM_030662.3(MAP2K2):c.603C>T (p.Leu201=)

CA137956 [↗](#)

40811 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: a290ee11-587b-4c61-861c-80b328f3af80

Approved on: 2017-05-09

Published on: 2018-12-10

HGVS expressions

NM_030662.3:c.603C>T

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NC_000019.10:g.4101121G>A

CM000681.2:g.4101121G>A

NC_000019.9:g.4101119G>A

CM000681.1:g.4101119G>A

NC_000019.8:g.4052119G>A

NG_007996.1:g.28008C>T

ENST00000262948.9:c.603C>T

ENST00000394867.8:c.312C>T

ENST00000593364.5:n.550C>T

ENST00000597008.5:n.204C>T

ENST00000597263.5:n.67C>T

ENST00000599345.1:n.873C>T

ENST00000601786.5:n.904C>T

ENST00000602167.5:n.323C>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.603C>T (p.Leu201=) variant in the MAP2K2 gene is 0.191% (18/6102) 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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