

Variant: NM_030662.3(MAP2K2):c.453C>T (p.Asp151=)

CA137943 [↗](#)

46237 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b79aa141-9bf2-4854-99cb-72cca42926e8

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HGVS expressions

NM_030662.3(MAP2K2):c.453C>T (p.Asp151=)

NM_030662.3:c.453C>T

NC_000019.10:g.4102451G>A

CM000681.2:g.4102451G>A

NC_000019.9:g.4102449G>A

CM000681.1:g.4102449G>A

NC_000019.8:g.4053449G>A

NG_007996.1:g.26678C>T

ENST00000262948.9:c.453C>T

ENST00000394867.8:c.162C>T

ENST00000593364.5:n.400C>T

ENST00000597008.5:n.54C>T

ENST00000599345.1:n.723C>T

ENST00000601786.5:n.754C>T

ENST00000602167.5:n.173C>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.453C>T (p.Asp151=) variant in the MAP2K2 gene is 27.128% (10569/38338) 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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