

Variant: NM_030662.3(MAP2K2):c.825G>A (p.Leu275=)

CA291999 [↗](#)

138161 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 13f158e3-3e4e-42f5-8828-d2509a51af7d

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HGVS expressions

NM_030662.3(MAP2K2):c.825G>A (p.Leu275=)

NM_030662.3:c.825G>A

NC_000019.10:g.4099295C>T

CM000681.2:g.4099295C>T

NC_000019.9:g.4099293C>T

CM000681.1:g.4099293C>T

NC_000019.8:g.4050293C>T

NG_007996.1:g.29834G>A

ENST00000262948.9:c.825G>A

ENST00000394867.8:c.534G>A

ENST00000593364.5:n.772G>A

ENST00000595715.1:n.640G>A

ENST00000597263.5:n.169+1724G>A

ENST00000599021.1:n.29+1724G>A

ENST00000600584.5:n.1385G>A

ENST00000601786.5:n.1126G>A

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.825G>A (p.Leu275=) variant in the MAP2K2 gene is 0.142% (14/5968) of East Asian 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

Showing 1 to 1 of 1 rows

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