

Variant: *NM_030662.3(MAP2K2):c.806C>G (p.Pro269Arg)*

Version: 1.0

CA180914 [↗](#)

177876 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4aa8ed55-4b35-4739-9e18-4c63fd36145a

Approved on: 2020/3/9

Published on: 2020/3/9

HGVS expressions

NM_030662.3:c.806C>G

NM_030662.3(MAP2K2):c.806C>G (p.Pro269Arg)

NC_000019.10:g.4099314G>C

CM000681.2:g.4099314G>C

NC_000019.9:g.4099312G>C

CM000681.1:g.4099312G>C

NC_000019.8:g.4050312G>C

NG_007996.1:g.29815C>G

ENST00000394867.9:n.1245C>G

ENST00000687128.1:n.1245C>G

ENST00000688002.1:n.1100C>G

ENST00000689792.1:n.710C>G

ENST00000262948.10:c.806C>G

ENST00000262948.9:c.806C>G

ENST00000394867.8:c.515C>G

ENST00000593364.5:n.753C>G

ENST00000595715.1:n.621C>G

ENST00000597263.5:n.169+1705C>G

ENST00000599021.1:c.29+1705C>G

ENST00000600584.5:n.1366C>G

ENST00000601786.5:n.1107C>G

NM_030662.4:c.806C>G

Uncertain Significance

Met criteria codes **3**

BP5

PP3

PM2

Not Met criteria codes **1**

BS2

Evidence Links **0**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

RASopathy VCEP

The c.806C>G (p.Pro269Arg) variant in MAP2K2 is absent from gnomAD (PM2). This variant has been observed in an individual with an alternate mechanism of disease (VCV000013329.8; Laboratory for Molecular Medicine internal data; BP5). Four apparently unaffected parental samples involved in whole exome testing were observed with this variant supporting that this variant is likely benign; however, this evidence does not meet current scoring criteria for BS2 at this time. Computational prediction tools and conservation analysis suggest that the p.Pro269Arg variant may impact the protein (PP3). In summary, the clinical significance of the p.Pro269Arg variant is uncertain. RASopathy-specific ACMG/AMP criteria applied (PMID:29493581): PP3, PM2, BP5.

Met criteria codes

BP5			4yo white male het. for this variant. Had diagnostic testing for NS. Also het. for c.184T>G (p.Tyr62Asp) in PTPN11, Path in ClinVar (ID: 13329) by 15 submitters w/o conflict (Laboratory for Molecular Medicine internal data)
PP3			Although the REVEL score is .71, which is above the threshold to apply PP3
PM2			Absent from gnomAD

Not Met criteria codes

BS2			GeneDx: Identified by whole exome in 4 unaffected parents from 4 different trios (SCV000808657.1)
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Curation History



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