

Variant: NM_030662.3(MAP2K2):c.619G>A (p.Glu207Lys)

CA180944 [↗](#)

40813 (ClinVar) [↗](#)

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

Condition: cardiofaciocutaneous syndrome ([MONDO:0015280](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 93491eab-4df5-480a-a62b-7022611f18ee

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HGVS expressions

NM_030662.3:c.619G>A

NM_030662.3(MAP2K2):c.619G>A (p.Glu207Lys)

NM_030662.4:c.619G>A

ENST00000262948.9:c.619G>A

ENST00000394867.8:c.328G>A

ENST00000593364.5:n.566G>A

ENST00000597008.5:n.220G>A

ENST00000597263.5:n.83G>A

ENST00000599345.1:n.889G>A

ENST00000601786.5:n.920G>A

ENST00000602167.5:n.339G>A

NC_000019.10:g.4101105C>T

CM000681.2:g.4101105C>T

NC_000019.9:g.4101103C>T

CM000681.1:g.4101103C>T

NC_000019.8:g.4052103C>T

NG_007996.1:g.28024G>A

Pathogenic

Met criteria codes **5**

PP2 PM6 PM2 PS2 PS4

Evidence Links **0**

Expert Panel

[RASopathy VCEP](#) [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

RASopathy VCEP

The c.619G>A (p.Glu207Lys) variant in MAP2K2 was absent from large population studies (PM2; [gnomAD.broadinstitute.org](#)). It has been identified in 6 individuals with clinical features of a RASopathy (PS4; SCV000204213.4, SCV000207959.10, SCV000815593.1, Otto von Guericke University Magdeburg internal data). One of these cases was a confirmed de novo occurrence and another was an unconfirmed de novo occurrence (PS2; PM6; SCV000815593.1, Otto von Guericke University Magdeburg internal data). Additionally, the c.619G>A (p.Glu207Lys) is located in MAP2K2, which has been defined by the ClinGen RASopathy Expert Panel as a gene with a low rate of benign missense variants and pathogenic missense variants are common (PP2; PMID: 29493581). In summary, this variant meets criteria to be

classified as pathogenic for RASopathies in an autosomal dominant manner. Rasopathy-specific ACMG/AMP criteria applied (PMID:29493581): PS4, PS2, PM6, PM2, PP2.

Met criteria codes			
PP2			The variant is located in the MAP2K2 gene, which has been defined by the ClinGen RASopathy Expert Panel as a gene with a low rate of benign missense variants and pathogenic missense variants are common (PP2; PMID: 29493581).
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



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