

The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [\[Disclaimer\]](#)

Variant: *NM_004985.4(KRAS):c.24A>G (p.Val8=)*

[CA135558](#)

[45119 \(ClinVar\)](#)

Gene: KRAS ([HGNC:3845](#))

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

Inheritance Mode: Autosomal dominant inheritance

UID: 56c9c209-4dc1-4b23-8b13-5e4465526b77

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Published on: 2018-12-10

HGVS expressions

NM_004985.4:c.24A>G

NM_004985.4(KRAS):c.24A>G (p.Val8=)

NM_033360.3:c.24A>G

ENST00000256078.8:c.24A>G

ENST00000311936.7:c.24A>G

ENST00000556131.1:c.24A>G

ENST00000557334.5:c.24A>G

NC_000012.12:g.25245361T>C

CM000674.2:g.25245361T>C

NC_000012.11:g.25398295T>C

CM000674.1:g.25398295T>C

NC_000012.10:g.25289562T>C

NG_007524.1:g.10560A>G

Likely Benign

The Expert Panel has overridden the computationally generated classification - "Uncertain Significance - Insufficient Evidence"

Met criteria codes **1**

BS1

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.24A>G (p.Val8=)** variant in the **KRAS** gene is **0.0499% (35/51828)** of European chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BS1; PMID:29493581)

Met criteria codes

BS1



The filtering allele frequency of the **c.69C>T (p.Thr23=)** variant in the **MAP2K1** gene is **0.0484% (4/2820)** of African chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BS1; PMID:29493581)

Curation History [↗](#)

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