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
UUID: 56c9c209-4dc1-4b23-8b13-5e4465526b77
Approved on: 2017-04-18
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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