

Variant: *NM_004985.4(KRAS):c.90C>T (p.Asp30=)*

CA291858 [↗](#)

138061 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 68557210-74f7-4a1b-90d6-38f877f1a54a

Approved on: 2017-04-18

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HGVS expressions

NM_004985.4(KRAS):c.90C>T (p.Asp30=)

NM_004985.4:c.90C>T

NM_033360.3:c.90C>T

ENST00000256078.8:c.90C>T

ENST00000311936.7:c.90C>T

ENST00000556131.1:c.90C>T

ENST00000557334.5:c.90C>T

NC_000012.12:g.25245295G>A

CM000674.2:g.25245295G>A

NC_000012.11:g.25398229G>A

CM000674.1:g.25398229G>A

NC_000012.10:g.25289496G>A

NG_007524.1:g.10626C>T

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.90C>T (p.Asp30=) variant in the KRAS gene is 0.0292% (5/6746) of East Asian 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



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Curation History [↗](#)

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