

Variant: NM_004985.5(KRAS):c.519T>C (p.Asp173=)

CA135582 [↗](#)

41449 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: ecd3287d-e567-4c6a-ad88-0fcb872a517d

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HGVS expressions

NM_004985.5(KRAS):c.519T>C (p.Asp173=)

NM_004985.5:c.519T>C

NC_000012.12:g.25209843A>G

CM000674.2:g.25209843A>G

NC_000012.11:g.25362777A>G

CM000674.1:g.25362777A>G

NC_000012.10:g.25254044A>G

NG_007524.1:g.46078T>C

NG_007524.2:g.46161T>C

ENST00000557334.6:c.180T>C

ENST00000685328.1:c.519T>C

ENST00000686877.1:c.*490T>C

ENST00000687356.1:c.*217T>C

ENST00000688228.1:n.993T>C

ENST00000688940.1:c.519T>C

ENST00000690406.1:c.322T>C

ENST00000690804.1:c.*480T>C

ENST00000692768.1:c.321T>C

ENST00000693229.1:c.444T>C

ENST00000256078.10:c.*73T>C

ENST00000311936.8:c.519T>C

ENST00000256078.8:c.*73T>C

ENST00000311936.7:c.519T>C

ENST00000557334.5:c.180T>C

NM_004985.4:c.519T>C

NM_033360.3:c.*73T>C

NM_001369786.1:c.*73T>C

NM_001369787.1:c.519T>C

NM_033360.4:c.*73T>C

Benign

Met criteria codes 3

BA1 BP7 BP4

Not Met criteria codes 3

PP3 PM2 BS1

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information

Evidence submitted by expert panel

RASopathy VCEP

The c.519T>C variant in the KRAS gene is a synonymous (silent) variant (p.Asp173=) at a nucleotide that is not conserved as shown by UCSC browser, and not predicted by SpliceAI to impact splicing (BP4, BP7). The filtering allele frequency in gnomAD v2.1.1 is 0.2201 (28382/128282 alleles with 3073 homozygotes) in the European (non-Finnish) population, which is higher than the ClinGen RASopathy VCEP threshold (>0.0005) for BA1, and therefore meets this criterion (BA1). In summary, this variant meets criteria to be classified as benign for autosomal dominant RASopathies based on the ACMG/AMP criteria applied, as specified by the ClinGen RASopathy Variant Curation Expert Panel: BA1, BP4, BP7 (Specification Version 2.3, 12/3/2024)

Met criteria codes

BA1			The filtering allele frequency in gnomAD v2.1.1 is 0.2201 (28382/128282 alleles with 3073 homozygotes) in the European (non-Finnish) population, which is higher than the ClinGen RASopathy VCEP threshold (>0.0005) for BA1, and therefore meets this criterion (BA1).
BP7			The c.519T>C variant in the KRAS gene is a synonymous (silent) variant (p.Asp173=) at a nucleotide that is not conserved as shown by UCSC browser, and not predicted by SpliceAI to impact splicing
BP4			The c.519T>C variant in the KRAS gene is a synonymous (silent) variant (p.Asp173=) at a nucleotide that is not conserved as shown by UCSC browser, and not predicted by SpliceAI to impact splicing

Not Met criteria codes

PP3			The c.519T>C variant in the KRAS gene is a synonymous (silent) variant (p.Asp173=) at a nucleotide that is not conserved as shown by UCSC browser, and not predicted by SpliceAI to impact splicing
PM2			The filtering allele frequency in gnomAD v2.1.1 is 0.2201 (28382/128282 alleles with 3073 homozygotes) in the European (non-Finnish) population, which meets BA1
BS1			The filtering allele frequency in gnomAD v2.1.1 is 0.2201 (28382/128282 alleles with 3073 homozygotes) in the European (non-Finnish) population, which meets BA1



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