

Variant: NM_005343.3(HRAS):c.510G>A (p.Lys170=)

CA135992 [↗](#)

45305 (ClinVar) [↗](#)

Gene: HRAS ([HGNC:3265](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9a40f720-b52f-4f73-b00a-f4480714a568

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HGVS expressions

NM_005343.3(HRAS):c.510G>A (p.Lys170=)

NM_005343.3:c.510G>A

NC_000011.10:g.532696C>T

CM000673.2:g.532696C>T

NC_000011.9:g.532696C>T

CM000673.1:g.532696C>T

NC_000011.8:g.522696C>T

NG_007666.1:g.7855G>A

ENST00000397594.7:c.*20-66G>A

ENST00000417302.7:c.*79G>A

ENST00000397594.6:c.251-66G>A

ENST00000417302.6:c.*79G>A

ENST00000462734.2:c.*122G>A

ENST00000311189.8:c.510G>A

ENST00000311189.7:c.510G>A

ENST00000397594.5:c.*79G>A

ENST00000397596.6:c.510G>A

ENST00000417302.5:c.*79G>A

ENST00000451590.5:c.510G>A

ENST00000462734.1:n.285G>A

ENST00000478324.5:n.243-66G>A

ENST00000479482.1:n.431G>A

ENST00000493230.5:c.*79G>A

NM_001130442.1:c.510G>A

NM_005343.2:c.510G>A

NM_176795.3:c.*79G>A

NM_001130442.2:c.510G>A

NM_001318054.1:c.273G>A

NM_176795.4:c.*79G>A

NM_005343.4:c.510G>A

NM_001318054.2:c.273G>A

NM_001130442.3:c.510G>A

NM_176795.5:c.*79G>A

Met criteria codes **4**

BS1

BP4

BP7

BP5

Expert Panel

Likely Benign

Not Met criteria codes 3

RASopathy VCEP

BA1

PP3

PM2

Evidence Links 0

Criteria Specification Information

[Criteria Specification:](#) ClinGen RASopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for HRAS Version 2.1.0

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

RASopathy VCEP

The c.510G>A variant in the HRAS gene is a synonymous (silent) variant (p.Lys170=) at a nucleotide that is not predicted by SpliceAI to impact splicing (BP4, BP7). The filtering allele frequency in gnomAD v4.1.0 is 0.02525% (17/44900 alleles) in the East Asian population meeting BS1. This variant has been identified in patients with an alternate molecular basis for disease (BP5; LMM and GeneDx internal data; GTR ID's: 21766, 26957; ClinVar SCV000062144.5; SCV000168832.9). In summary, this variant meets criteria to be classified as likely benign for autosomal dominant RASopathies based on the ACMG/AMP criteria applied, as specified by the ClinGen RASopathy Variant Curation Expert Panel: BS1, BP4, BP5, BP7 (Specification Version 2.1, 09/17/2024)

Met criteria codes

BS1			The filtering allele frequency in gnomAD v4.1.0 is 0.02525% (17/44900 alleles) in the East Asian population meeting BS1.
BP4			Computational prediction tool SpliceAI predicts no impact on splicing (BP4).
BP7			The c.510G>A (p.Lys170=) variant is a synonymous (silent) variant at a nucleotide that is not highly conserved and is not predicted to impact splicing (BP7).
BP5			This variant has been identified in a patient with an alternate molecular basis for disease (BP5; LMM and GeneDx internal data; GTR ID's: 21766, 26957; ClinVar SCV000062144.5; SCV000168832.9).

Not Met criteria codes

BA1			The filtering allele frequency in gnomAD v4.1.0 is 0.02525% (17/44900 alleles) in the East Asian population meeting BS1.
PP3			Computational prediction tools and conservation analysis suggest that the p.Lys170= variant does not impact the protein (BP4).
PM2			The filtering allele frequency in gnomAD v4.1.0 is 0.02525% (17/44900 alleles) in the East Asian population meeting BS1.

[illegible]

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.