

Variant: NM_005343.3(HRAS):c.520C>T (p.Pro174Ser)

CA135994 [↗](#)

40448 (ClinVar) [↗](#)

Gene: LRRC56 ([HGNC:3265](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d37c8250-6a81-4f7a-b0df-8e5eb3216e8a

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HGVS expressions

NM_005343.3:c.520C>T

NM_005343.3(HRAS):c.520C>T (p.Pro174Ser)

NM_001130442.1:c.520C>T

NM_005343.2:c.520C>T

NM_176795.3:c.*89C>T

NM_001130442.2:c.520C>T

NM_001318054.1:c.283C>T

NM_176795.4:c.*89C>T

NM_005343.4:c.520C>T

ENST00000311189.7:c.520C>T

ENST00000397594.5:c.*89C>T

ENST00000397596.6:c.520C>T

ENST00000417302.5:c.*89C>T

ENST00000451590.5:c.520C>T

ENST00000462734.1:n.295C>T

ENST00000478324.5:n.243-56C>T

ENST00000479482.1:n.441C>T

ENST00000493230.5:c.*89C>T

NC_000011.10:g.532686G>A

CM000673.2:g.532686G>A

NC_000011.9:g.532686G>A

CM000673.1:g.532686G>A

NC_000011.8:g.522686G>A

NG_007666.1:g.7865C>T

Benign

Met criteria codes **1**

BA1

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.520C>T (p.Pro174Ser) variant in the HRAS gene is 0.115% (20/11482) of Latino chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Met criteria codes

BA1			The filtering allele frequency of the c.520C>T (p.Pro174Ser) variant in the HRAS gene is 0.115% (20/11482) of Latino chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)
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Curation History



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