

Variant: NM_005343.3(HRAS):c.378A>G (p.Glu126=)

CA135984 [↗](#)

40442 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 33df4a63-9662-4572-ab6f-22dc7b693309

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HGVS expressions

NM_005343.3(HRAS):c.378A>G (p.Glu126=)

NM_005343.3:c.378A>G

NM_001130442.1:c.378A>G

NM_005343.2:c.378A>G

NM_176795.3:c.378A>G

NM_001130442.2:c.378A>G

NM_001318054.1:c.59A>G

NM_176795.4:c.378A>G

NM_005343.4:c.378A>G

ENST00000311189.7:c.378A>G

ENST00000397594.5:c.378A>G

ENST00000397596.6:c.378A>G

ENST00000417302.5:c.378A>G

ENST00000451590.5:c.378A>G

ENST00000462734.1:n.71A>G

ENST00000478324.5:n.88A>G

ENST00000479482.1:n.299A>G

ENST00000493230.5:c.378A>G

NC_000011.10:g.533525T>C

CM000673.2:g.533525T>C

NC_000011.9:g.533525T>C

CM000673.1:g.533525T>C

NC_000011.8:g.523525T>C

NG_007666.1:g.7026A>G

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.378A>G (p.Glu126=) variant in the HRAS gene is 0.095% (23/16510) of South Asian 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.378A>G (p.Glu126=) variant in the HRAS gene is 0.095% (23/16510) of South Asian 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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