

Variant: NM_005343.3(HRAS):c.357C>T (p.Asp119=)

CA135978 [↗](#)

40441 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4da936bb-53cd-4fad-8d47-40c90dc60a73

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HGVS expressions

NM_005343.3(HRAS):c.357C>T (p.Asp119=)

NM_005343.3:c.357C>T

NM_001130442.1:c.357C>T

NM_005343.2:c.357C>T

NM_176795.3:c.357C>T

NM_001130442.2:c.357C>T

NM_001318054.1:c.38C>T

NM_176795.4:c.357C>T

NM_005343.4:c.357C>T

ENST00000311189.7:c.357C>T

ENST00000397594.5:c.357C>T

ENST00000397596.6:c.357C>T

ENST00000417302.5:c.357C>T

ENST00000451590.5:c.357C>T

ENST00000462734.1:n.50C>T

ENST00000478324.5:n.67C>T

ENST00000479482.1:n.278C>T

ENST00000493230.5:c.357C>T

NC_000011.10:g.533546G>A

CM000673.2:g.533546G>A

NC_000011.9:g.533546G>A

CM000673.1:g.533546G>A

NC_000011.8:g.523546G>A

NG_007666.1:g.7005C>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.357C>T (p.Asp119=) variant in the HRAS gene is 0.129% (102/66416) of European 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.357C>T (p.Asp119=) variant in the HRAS gene is 0.129% (102/66416) of European 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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