

Variant: NM_005343.3(HRAS):c.309G>A (p.Val103=)

CA5779334 [↗](#)

40440 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b7f7a482-5a60-4da9-8e99-433b8dea636b

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HGVS expressions

NM_005343.3(HRAS):c.309G>A (p.Val103=)

NM_005343.3:c.309G>A

NC_000011.10:g.533594C>T

CM000673.2:g.533594C>T

NC_000011.9:g.533594C>T

CM000673.1:g.533594C>T

NC_000011.8:g.523594C>T

NG_007666.1:g.6957G>A

NM_001130442.1:c.309G>A

NM_005343.2:c.309G>A

NM_176795.3:c.309G>A

NM_001130442.2:c.309G>A

NM_001318054.1:c.-11G>A

NM_176795.4:c.309G>A

NM_005343.4:c.309G>A

ENST00000311189.7:c.309G>A

ENST00000397594.5:c.309G>A

ENST00000397596.6:c.309G>A

ENST00000417302.5:c.309G>A

ENST00000451590.5:c.309G>A

ENST00000462734.1:n.2G>A

ENST00000478324.5:n.19G>A

ENST00000479482.1:n.230G>A

ENST00000493230.5:c.309G>A

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.309G>A (p.Val103=) variant in the HRAS gene is 0.063% (10/8640) of East 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



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Curation History

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Showing 1 to 1 of 1 rows

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