

Variant: NM_005343.3(HRAS):c.81T>C (p.His27=)

CA135996 [↗](#)

40431 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: c23db7a4-fb09-4dd7-9ab1-e7f4dfc7dbe9

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HGVS expressions

NM_005343.3(HRAS):c.81T>C (p.His27=)

NM_005343.3:c.81T>C

NC_000011.10:g.534242A>G

CM000673.2:g.534242A>G

NC_000011.9:g.534242A>G

CM000673.1:g.534242A>G

NC_000011.8:g.524242A>G

NG_007666.1:g.6309T>C

NM_001130442.1:c.81T>C

NM_005343.2:c.81T>C

NM_176795.3:c.81T>C

NM_001130442.2:c.81T>C

NM_001318054.1:c.-239T>C

NM_176795.4:c.81T>C

NM_005343.4:c.81T>C

ENST00000311189.7:c.81T>C

ENST00000397594.5:c.81T>C

ENST00000397596.6:c.81T>C

ENST00000417302.5:c.81T>C

ENST00000451590.5:c.81T>C

ENST00000468682.2:n.569T>C

ENST00000493230.5:c.81T>C

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.81T>C (p.His27=) variant in the HRAS gene is 36.7% for African chromosomes by the Exome Aggregation Consortium (3874/10274 with 95% CI), which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert panel for autosomal dominant RASopathy variants (BA1). Additional case-level data provided by: SCV000058315; SCV000062146; SCV000196686.

Met criteria codes

BA1	
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