

Variant: NM_005633.3(SOS1):c.195A>C (p.Arg65=)

CA136085 [↗](#)

40643 (ClinVar) [↗](#)

Gene: SOS1 ([HGNC:6654](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 7c7c5faa-dff9-4352-b563-fea9d0af98a6

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HGVS expressions

NM_005633.3:c.195A>C

NM_005633.3(SOS1):c.195A>C (p.Arg65=)

ENST00000395038.6:c.195A>C

ENST00000402219.6:c.195A>C

ENST00000426016.5:c.195A>C

ENST00000451331.1:c.24A>C

NC_000002.12:g.39067646T>G

CM000664.2:g.39067646T>G

NC_000002.11:g.39294787T>G

CM000664.1:g.39294787T>G

NC_000002.10:g.39148291T>G

NG_007530.1:g.57818A>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.195A>C (p.Arg65=) variant in the SOS1 gene is 10.978% (1192/10346) of African 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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