

Variant: NM_005633.3(SOS1):c.570C>T (p.Asp190=)

CA136171 [↗](#)

40655 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b4afa727-6000-4b36-b685-4bcd8c3df504

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HGVS expressions

NM_005633.3(SOS1):c.570C>T (p.Asp190=)

NM_005633.3:c.570C>T

ENST00000395038.6:c.570C>T

ENST00000402219.6:c.570C>T

ENST00000426016.5:c.570C>T

NC_000002.12:g.39054764G>A

CM000664.2:g.39054764G>A

NC_000002.11:g.39281905G>A

CM000664.1:g.39281905G>A

NC_000002.10:g.39135409G>A

NG_007530.1:g.70700C>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.570C>T (p.Asp190=) variant in the SOS1 gene is 0.314% (234/66628) 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.570C>T (p.Asp190=) variant in the SOS1 gene is 0.314% (234/66628) 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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