

Variant: NM_005633.3(SOS1):c.3709C>A (p.Pro1237Thr)

CA136149 [↗](#)

45367 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 95b1a6fb-b4b5-4f40-b593-6235a54a0739

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HGVS expressions

NM_005633.3(SOS1):c.3709C>A (p.Pro1237Thr)

NM_005633.3:c.3709C>A

ENST00000395038.6:c.3664C>A

ENST00000402219.6:c.3709C>A

ENST00000426016.5:c.3709C>A

ENST00000469581.1:n.452C>A

NC_000002.12:g.38986117G>T

CM000664.2:g.38986117G>T

NC_000002.11:g.39213258G>T

CM000664.1:g.39213258G>T

NC_000002.10:g.39066762G>T

NG_007530.1:g.139347C>A

Likely Benign

The Expert Panel has overridden the computationally generated classification - "Uncertain Significance - Insufficient Evidence"

Met criteria codes **1**

BS1

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.3709C>A (p.Pro1237Thr) variant in the SOS1 gene is 0.0373% (11/16510) of South Asian chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BS1; PMID:29493581)

Met criteria codes

BS1			The filtering allele frequency of the c.3709C>A (p.Pro1237Thr) variant in the SOS1 gene is 0.0373% (11/16510) of South Asian chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BS1; PMID:29493581)
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Curation History



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