

Variant: *NM_007373.4(SHOC2):c.170C>T (p.Ser57Phe)*

Version: 1.2

CA185080 [↗](#)

179760 (ClinVar) [↗](#)

Gene: SHOC2 ([HGNC:8036](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 6b2642de-1576-44fc-ad42-04b45dae029d

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HGVS expressions

NM_007373.4(SHOC2):c.170C>T (p.Ser57Phe)

NM_007373.4:c.170C>T

NC_000010.11:g.110964528C>T

CM000672.2:g.110964528C>T

NC_000010.10:g.112724286C>T

CM000672.1:g.112724286C>T

NC_000010.9:g.112714276C>T

NG_028922.1:g.49986C>T

ENST00000265277.10:c.170C>T

ENST00000451838.2:c.-242-35887C>T

ENST00000480155.2:n.406C>T

ENST00000685059.1:c.170C>T

ENST00000685613.1:c.170C>T

ENST00000687592.1:n.469C>T

ENST00000688928.1:c.170C>T

ENST00000689118.1:c.170C>T

ENST00000689300.1:c.170C>T

ENST00000689997.1:c.-380-21100C>T

ENST00000691151.1:n.462C>T

ENST00000691369.1:c.170C>T

ENST00000691441.1:c.170C>T

ENST00000691903.1:c.170C>T

ENST00000692776.1:c.170C>T

ENST00000369452.9:c.170C>T

ENST00000265277.9:c.170C>T

ENST00000369452.8:c.170C>T

ENST00000489390.1:n.56-35887C>T

ENST00000489783.1:n.548C>T

NM_001269039.1:c.170C>T

NM_007373.3:c.170C>T

NM_001269039.2:c.170C>T

NM_001324336.1:c.170C>T

NM_001324337.1:c.170C>T

NR_136749.1:n.116-21100C>T

NM_001269039.3:c.170C>T

NM_001324336.2:c.170C>T

NM_001324337.2:c.170C>T

Uncertain Significance

Met criteria codes 1

PP2

Evidence Links 0

Expert Panel

RASopathy VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

RASopathy VCEP

The c.170C>T (p.Ser57Phe) variant in SHOC2 was present in 0.0055% (5/35376 CI 95%) of Latino alleles in gnomAD v2.1.1 (BS1 not met; PMID: 29493581). It has been identified in healthy individuals without clinical features of a RASopathy, but this information does not meet the current criteria to apply BS2 (Ambry Genetics internal data, Baylor Genetics internal data). The variant is located in SHOC2, which has been defined by the ClinGen RASopathy Expert Panel as a gene with a low rate of benign missense variants and pathogenic missense variants are common (PP2; PMID: 29493581). In summary, the clinical significance of the p.Ser57Phe variant is uncertain. RASopathy-specific ACMG/AMP criteria applied (PMID:29493581): PP2.

Met criteria codes

PP2



No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



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