

Variant: *NM_006939.4(SOS2):c.674G>A (p.Arg225Gln)*

Version: 1.0

CA7177479 [↗](#)

373121 (ClinVar) [↗](#)

Gene: SOS2 (HGNC:6655)

Condition: RASopathy (MONDO:0021060)

Inheritance Mode: Autosomal dominant inheritance

UID: ca1b8554-fe09-4514-ade4-bd0b60333374

Approved on: 2024-09-17

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HGVS expressions

NM_006939.4:c.674G>A

NM_006939.4(SOS2):c.674G>A (p.Arg225Gln)

NC_000014.9:g.50188537C>T

CM000676.2:g.50188537C>T

NC_000014.8:g.50655255C>T

CM000676.1:g.50655255C>T

NC_000014.7:g.49725005C>T

NG_051073.1:g.48157G>A

ENST00000216373.10:c.674G>A

ENST00000216373.9:c.674G>A

ENST00000543680.5:c.674G>A

ENST00000556469.5:n.482-5931G>A

NM_006939.2:c.674G>A

NM_006939.3:c.674G>A

Uncertain Significance

Not Met criteria codes **2**

BP4 BA1

Evidence Links **0**

Expert Panel

RASopathy VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen RASopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for SOS2 Version 2.1.0

[↗](#) **Criteria Specification Approval History**





[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

RASopathy VCEP

The c.674G>A (NM_006939.4(SOS2):c.674G>A (p.Arg225Gln)) variant in RIT1 is a missense variant predicted to cause substitution of arginine by glutamine at amino acid 225. No ACMG/AMP evidence codes are met. In summary, this variant meets the criteria to be classified as uncertain significance for autosomal dominant RASopathy (Version 2.1; 09/17/2024).

Not Met criteria codes

BP4	 	Does not meet threshold for pathogenic readout, well conserved in UCSC, no predicted splice impact
BA1	 	Present in 0.01885% (3/15918) African/African-American alleles in gnomAD v2.1.1

Curation History [↗](#)

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