

Variant: *NM_000536.4(RAG2):c.1095T>C (p.Ser365=)*

Version: 1.0

CA5950474 [↗](#)

304552 (ClinVar) [↗](#)

Gene: RAG2 ([HGNC:5897](#))

Condition: recombinaase activating gene 2 deficiency ([MONDO:0000573](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 3454b9e8-3665-4835-8fc9-2cc823ee5ebc

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HGVS expressions

NM_000536.4(RAG2):c.1095T>C (p.Ser365=)

NM_000536.4:c.1095T>C

NC_000011.10:g.36593074A>G

CM000673.2:g.36593074A>G

NC_000011.9:g.36614624A>G

CM000673.1:g.36614624A>G

NC_000011.8:g.36571200A>G

NG_007573.1:g.10163T>C

NG_033154.1:g.3582A>G

ENST00000527033.6:c.1095T>C

ENST00000529083.2:c.1095T>C

ENST00000532616.2:c.1095T>C

ENST00000311485.8:c.1095T>C

ENST00000311485.7:c.1095T>C

ENST00000524423.1:n.131+5028T>C

ENST00000534663.1:c.*193A>G

ENST00000618712.4:c.1095T>C

NM_000536.3:c.1095T>C

NM_001243785.1:c.1095T>C

NM_001243786.1:c.1095T>C

NM_001243785.2:c.1095T>C

NM_001243786.2:c.1095T>C

Likely Benign

Met criteria codes **2**

BS1 BP7

Evidence Links **0**

Expert Panel

Severe Combined Immunodeficiency Disease VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Severe Combined Immunodeficiency Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for RAG2 Version 1.0.0

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

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

Severe Combined Immunodeficiency Disease VCEP

The NM_000536.4:c.1095T>C variant is a synonymous (silent) variant (p.Ser365=) with no predicted splice impact (BP7). This variant has not been reported in the literature in individuals with RAG2-related conditions. The highest population minor allele frequency in gnomAD v2.1.1 is 0.00314 (79/25120) in the Finnish population, which is higher than the ClinGen SCID VCEP's threshold for BS1 (>0.00195). In summary, this variant meets the criteria to be classified as Likely Benign for autosomal recessive SCID based on the ACMG criteria applied BS1 and BP7, as specified by the ClinGen SCID VCEP (VCEP specifications 1).

Met criteria codes

BS1			Highest subpopulation frequency (Finnish) in gnomAD is 79/25120 (0.00314) which is greater than the allele frequency expected for RAG2-related disease (>0.00195). There is also 1 homozygote in gnomAD.
BP7			Silent variant, no predicted splice impact. As outlined in SCID VCEP RAG2 specifications, nucleotide conservation is not required in order to apply BP7.

Curation History



 

 

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