

Variant: *NM_002185.5(IL7R):c.132C>T (p.Ser44=)*

Version: 1.0

CA3231851 [↗](#)

377976 (ClinVar) [↗](#)

Gene: IL7R ([HGNC:3575](#))

Condition: severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive ([MONDO:0012163](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: b873b5bd-5641-4da9-8c71-cd000bf4dba6

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HGVS expressions

NM_002185.5(IL7R):c.132C>T (p.Ser44=)

NM_002185.5:c.132C>T

NC_000005.10:g.35860901C>T

CM000667.2:g.35860901C>T

NC_000005.9:g.35861003C>T

CM000667.1:g.35861003C>T

NC_000005.8:g.35896760C>T

NG_009567.1:g.9013C>T

ENST00000303115.8:c.132C>T

ENST00000303115.7:c.132C>T

ENST00000506850.5:c.132C>T

ENST00000508941.5:c.132C>T

ENST00000511031.1:n.266C>T

ENST00000511982.1:c.132C>T

ENST00000514217.5:c.132C>T

ENST00000515665.1:c.132C>T

NM_002185.3:c.132C>T

NR_120485.1:n.235C>T

NM_002185.4:c.132C>T

NR_120485.2:n.261C>T

NR_120485.3:n.219C>T

Likely Benign

Met criteria codes **2**

BS1 BP7

Not Met criteria codes **1**

BS2

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 IL7R Version 1.0.0

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

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

Severe Combined Immunodeficiency Disease VCEP

The c.132C>T (p.Ser44=) variant (NM_002185.5) variant in IL7R is a synonymous (silent) variant that is not predicted to impact splicing (BP7). The filtering allele frequency (the lower threshold of the 95% CI of 342/24962) of the c.132C>T variant in IL7R is 0.01271 in exomes and 0.01082 in genomes for African/African American chromosomes by gnomAD v2.1.1. Both values are higher than the ClinGen SCID VCEP threshold (0.00126) for BS1 and therefore meet this criterion (BS1). In summary, this variant meets the criteria to be classified as likely benign for SCID. ACMG/AMP criteria applied, as specified by the ClinGen SCID VCEP: BP7 and BS1. (VCEP specifications version 1).

Met criteria codes

BS1			The filtering allele frequency (the lower threshold of the 95% CI of 342/24962) of the c.132C>T variant in IL7R is 0.01271 in exomes and 0.01082 in genomes for African/African American chromosomes by gnomAD v2.1.1. Both values are higher than the ClinGen SCID VCEP threshold (0.00126) for BS1 and therefore meet this criterion (BS1).
BP7			The c.132C>T (p.Ser44=) variant (NM_002185.5) is a synonymous (silent) variant that varSEAK, NNSplice, and SpliceAI do not predict impact splicing. (BP7 is met).

Not Met criteria codes

BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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Curation History



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