

Gene — phenotype browser

Query disease gene to access evidence sources

Accurately report your finding

- ⚡ **ACMG standards and guidelines:** for the interpretation of variants.
- ⚡ **Use accurate HGVS variant nomenclature:** for precise clinical genetics.
- ⚡ **Use accurate HGNC gene names:** for precise clinical genetics.
- ⚡ **Find other genes:** outside of IUIS IEI genes.
- ⚡ **Coming soon: summarise all diagnostic evidence sources.**

IUIS IEI table

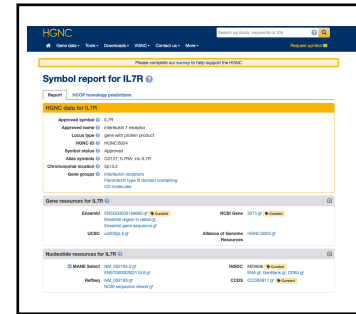
Disease	Gene symbol	Inheritance	Inheritance detail	T cell count	B cell count	Immunoglobulin levels	Neutrophil count
CD3d deficiency	CD3D	AR		Very low	Normal	Low	Normal
CD3e deficiency	CD3E	AR		Very low	Normal	Low	Normal
CD3z deficiency	CD3Z	AR		Very low	Normal	Low	Normal
Coronin-1A deficiency	CORO1A	AR		Very low	Normal	Low	Normal
gc deficiency (common gamma chain SCID, CD132 deficiency)	IL2RG	XL		Very low	Normal to high	Low	Normal
IL7Ra deficiency	IL7R	AR		Very low	Normal to high	Low	Normal

New: GWAS Catalog

Variant and risk allele	P-value	P-value annotation	RAF	OR	Beta	CI	Mapped gene	Reported trait	Trait(s)
rs6897932-C	2 x 10 ⁻⁶		0.75	1.12	-	[1.02-1.23]	IL7R	Multiple sclerosis	multiple sclerosis
rs6897932-G	8 x 10 ⁻⁶		0.71	1.12	-	[1.06-1.19]	IL7R	Type 1 diabetes	type 1 diabetes mellitus
rs6897932-G	2 x 10 ⁻⁸		NR	1.11	-	[1.09-1.13]	IL7R	Multiple sclerosis	multiple sclerosis
rs11567701-T	1 x 10 ⁻⁶³		0.260269	-	0.04246806 unit increase	[0.038-0.047]	IL7R	Lymphocyte percentage of white cells	lymphocyte percentage of leukocytes
rs987107-A	3 x 10 ⁻²²		0.261079	-	0.020353 SD unit increase	[0.016-0.024]	IL7R	White blood cell count	myeloid white cell count

Showing 1 to 5 of 101 rows 5 rows per page

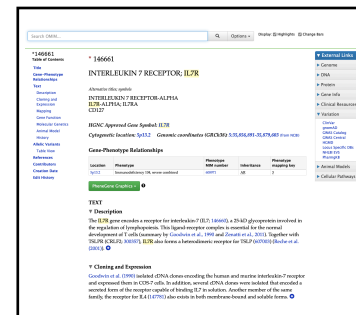
HGNC



Symbol report for IL7R

Approved symbol: IL7R
Approved name: IL7R
Locus type: gene with protein product
HGNC ID: HGNC:00438
Symbol status: Approved
Gene symbol: IL7R
Chromosome location: 12p12
Gene product: Interleukin-7 receptor
Gene function: Interleukin-7 receptor, a heterodimeric receptor consisting of an alpha chain and a beta chain.

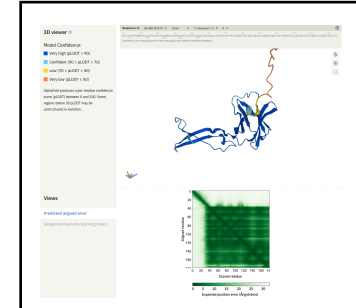
OMIM



316661
INTERLEUKIN-7 RECEPTOR, IL7R
INTERLEUKIN-7 RECEPTOR-ALPHA
IL7R-1, IL7R-1A, IL7R-1B
CD127

OMIM Approved Gene Symbol: IL7R
OMIM Approved Gene Name: IL7R
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OMIM Approved Gene Name: IL7R

Alpha fold



3D viewer

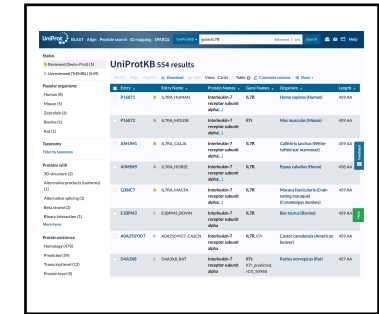
Model Confidence

High (0.9-1.0)
Medium (0.5-0.9)
Low (0.1-0.5)
Disordered (0.0-0.1)

Visualise

Protein structure visualization showing the IL7R receptor structure.

UniProt



UniProtKB S54 results

Accession	Protein	Organism	Length	Mass	PI
P18711	IL7R1_HUMAN	Homo sapiens	454	49.5 kDa	5.15
A01451	IL7R1_MOUSE	Mus musculus	454	49.5 kDa	5.15
A01451	IL7R1_MOUSE	Mus musculus	454	49.5 kDa	5.15
A01451	IL7R1_MOUSE	Mus musculus	454	49.5 kDa	5.15

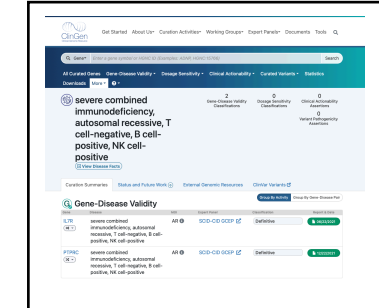
GnomAD



gnomAD genome browser view for IL7R

Genome browser view showing the IL7R gene structure and associated variants.

ClinGen



ClinGen Gene-Disease Validity

Gene: IL7R
Disease: Multiple sclerosis
Evidence: 1
Clinical Significance: Pathogenic
Clinical Validity: 1

and more