



## IL2RG gene

interleukin 2 receptor subunit gamma

### Normal Function

The *IL2RG* gene provides instructions for making a protein called the common gamma chain. This protein is a component of several different receptors that are involved in immune system function. The receptors span the cell membrane, with one end outside the cell like an antenna and the other end inside to transmit signals to the nucleus. Other proteins attach to these receptors, like a key in a lock, to trigger a series of chemical reactions inside the cell.

Receptors containing the common gamma chain are located on the surface of immature blood-forming cells in bone marrow. They partner with other proteins to direct blood-forming cells to form lymphocytes (a type of white blood cell). The receptors also regulate the growth and maturation of several subtypes of lymphocytes: T cells, B cells, and natural killer cells. These cells kill viruses, make antibodies, and help regulate the entire immune system.

### Health Conditions Related to Genetic Changes

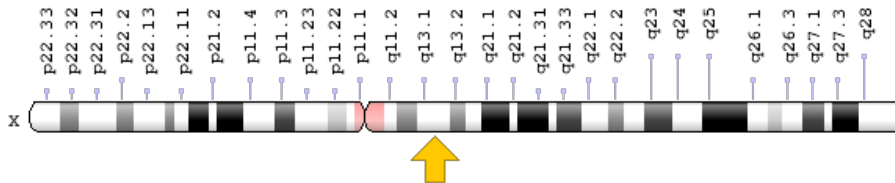
#### X-linked severe combined immunodeficiency

More than 300 mutations in the *IL2RG* gene have been identified in people with X-linked severe combined immunodeficiency (SCID). Most of these mutations involve changes in one or a few DNA building blocks (nucleotides) in the gene. These changes lead to the production of a nonfunctional version of the common gamma chain or prevent any protein from being produced. Without the common gamma chain, important chemical signals are not relayed to the nucleus and lymphocytes cannot develop normally. A lack of functional mature lymphocytes prevents the immune system from fighting off infections.

## Chromosomal Location

Cytogenetic Location: Xq13.1, which is the long (q) arm of the X chromosome at position 13.1

Molecular Location: base pairs 71,107,404 to 71,111,577 on the X chromosome (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CD132
- common cytokine receptor gamma chain
- Gamma-C
- IL2RG\_HUMAN
- IMD4
- interleukin 2 receptor, gamma
- interleukin 2 receptor, gamma (severe combined immunodeficiency)
- SCIDX
- SCIDX1
- $\gamma_c$

## Additional Information & Resources

### Educational Resources

- Immunobiology (fifth edition, 2001): Defects in T-cell function result in severe combined immunodeficiencies  
<https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1509>

### Clinical Information from GeneReviews

- X-Linked Severe Combined Immunodeficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK1410>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28IL2RG%5BTIAB%5D%29+OR+%28%28CD132%5BTIAB%5D%29+OR+%28common+cytokine+receptor+gamma+chain%5BTI%5D%29+OR+%28IMD4%5BTIAB%5D%29+OR+%28SCIDX%5BTIAB%5D%29+OR+%28common+gamma+chain%5BTI%5D%29+OR+%28IL-2+receptor%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- INTERLEUKIN 2 RECEPTOR, GAMMA  
<http://omim.org/entry/308380>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_IL2RG.html](http://atlasgeneticsoncology.org/Genes/GC_IL2RG.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=IL2RG%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:6010](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6010)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:3561>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3561>
- UniProt  
<https://www.uniprot.org/uniprot/P31785>

### **Sources for This Summary**

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