

## JAK3 gene

Janus kinase 3

### Normal Function

The *JAK3* gene provides instructions for making a protein that is critical for the normal development and function of the immune system. The JAK3 protein is part of a signaling pathway called the JAK/STAT pathway, which transmits chemical signals from outside the cell to the cell's nucleus. Signals relayed by the JAK3 protein regulate the growth and maturation of certain types of white blood cells (lymphocytes) called T cells and natural killer cells. In addition, JAK3 is important for the normal maturation of another type of lymphocyte called B cells. T cells, B cells, and natural killer cells attack bacteria, viruses, and fungi and help regulate the entire immune system.

### Health Conditions Related to Genetic Changes

#### JAK3-deficient severe combined immunodeficiency

More than 50 *JAK3* gene mutations have been identified in people with *JAK3*-deficient severe combined immunodeficiency (SCID), an impairment of the immune system that leads to repeated and persistent infections that can be life-threatening.

Most mutations in the *JAK3* gene prevent the production of JAK3 protein or result in a protein that is unstable and quickly broken down. Others lead to production of a protein that cannot perform its function. Without functional JAK3 protein, certain chemical signals are not relayed to the nucleus, impairing lymphocyte growth and development. As a result, individuals with *JAK3*-deficient SCID have few or no T cells and natural killer cells and a normal number of poorly functioning B cells. The shortage of functional lymphocytes causes people with this condition to be susceptible to infections.

### Other Names for This Gene

- JAK-3
- JAK3\_HUMAN
- JAKL
- Janus kinase 3 (a protein tyrosine kinase, leukocyte)
- L-JAK
- leukocyte Janus kinase

- LJAK
- tyrosine-protein kinase JAK3

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of JAK3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3718\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3718[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28JAK3%5BTIAB%5D%29+OR+%28Janus+kinase+3%5BTIAB%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+720+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- JANUS KINASE 3; JAK3 (<https://omim.org/entry/600173>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3718>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=JAK3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=JAK3[gene]))

## References

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## **Genomic Location**

The *JAK3* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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