JAK3-deficient severe combined immunodeficiency

Description

JAK3-deficient severe combined immunodeficiency (SCID) is an inherited disorder of the immune system. Individuals with *JAK3*-deficient SCID lack the necessary immune cells to fight off certain bacteria, viruses, and fungi. They are prone to repeated and persistent infections that can be very serious or life-threatening. Often the organisms that cause infection in people with *JAK3*-deficient SCID are described as opportunistic because they ordinarily do not cause illness in healthy people. Affected infants typically develop chronic diarrhea, a fungal infection in the mouth called oral thrush, pneumonia, and skin rashes. Persistent illness also causes affected individuals to grow more slowly than other children. Without treatment, people with *JAK3*-deficient SCID usually live only into early childhood.

Frequency

JAK3-deficient SCID accounts for an estimated 7 to 14 percent of cases of SCID. The prevalence of SCID from all genetic causes combined is approximately 1 in 50,000, although it may be higher in certain regions.

Causes

JAK3-deficient SCID is caused by mutations in the *JAK3* gene. The protein produced from this gene helps regulate the growth and maturation of certain types of white blood cells (lymphocytes) called T cells and natural killer cells. In addition, the JAK3 protein 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.

Mutations in the *JAK3* gene prevent the production of JAK3 protein or lead to production of a nonfunctional protein. A loss of functional JAK3 protein results in the absence of T cells and natural killer cells and a normal number of poorly functioning B cells. This shortage of functional lymphocytes causes people with *JAK3*-deficient SCID to be susceptible to infections.

Learn more about the gene associated with JAK3-deficient severe combined immunodeficiency

- JAK3

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Autosomal recessive T cell-negative, B cell-positive, NK cell-negative severe combined immunodeficiency
- Autosomal recessive T-B+NK- SCID
- JAK3 SCID
- T cell-negative, B cell-positive, NK cell-negative SCID
- T-B+ severe combined immunodeficiency due to JAK3 deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: T-B+ severe combined immunodeficiency due to JAK3 deficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1833275/)

Genetic and Rare Diseases Information Center

- Severe combined immunodeficiency (https://rarediseases.info.nih.gov/diseases/7628/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-NEGATIVE (https://omim.org/entry/600802)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28JAK3%5BTIAB%5D%29+AND+%28severe+combined+immunodeficiency%5BTIAB%5D%29%29+OR+%28%28JAK3%5BTIAB%5D%29+AND+%28SCID%5BTIAB%5D%29%29+OR+%28JA

K3+severe+combined+immunodeficiency%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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