X-linked severe combined immunodeficiency

Description

X-linked severe combined immunodeficiency (SCID) is an inherited disorder of the immune system that occurs almost exclusively in males. Children with X-linked SCID are prone to recurrent and persistent infections because they lack the necessary immune cells to fight off certain bacteria, viruses, and fungi.

If untreated, infants with X-linked SCID can develop poor growth, chronic diarrhea, a fungal infection called thrush, skin rashes, and life-threatening infections. X-linked SCID can be detected shortly after birth by newborn screening, which allows for prompt treatment.

Frequency

X-linked SCID is the most common form of a group of severe combined immunodeficiency disorders. This group of disorders can be caused by variants in more than 20 genes. The incidence of all severe combined immunodeficiency disorders is 1 in 60,000 newborns and it is estimated that one-quarter to one-third of these cases are X-linked SCID.

Causes

Variants (also known as mutations) in the *IL2RG* gene cause X-linked SCID. The *IL2RG* gene provides instructions for making a protein that is critical for normal immune system function. This protein is necessary for the growth and maturation of developing immune system cells called lymphocytes. Lymphocytes defend the body against potentially harmful invaders, make antibodies, and help regulate the entire immune system. Variants in the *IL2RG* gene prevent these cells from developing and functioning normally. Without functional lymphocytes, the body is unable to fight off infections.

Learn more about the gene associated with X-linked severe combined immunodeficiency

- IL2RG

Inheritance

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a variant would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- IL2RG SCID, T- B+ NK-
- SCIDX1
- X-linked SCID
- X-SCID
- XSCID

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: X-linked severe combined immunodeficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1279481/)

Genetic and Rare Diseases Information Center

- T-B+ severe combined immunodeficiency due to gamma chain deficiency (https://rarediseases.info.nih.gov/diseases/5618/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22X-linked severe combined immunodeficiency%22)

Catalog of Genes and Diseases from OMIM

- SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED; SCIDX1 (https://omim.org/entry/300400)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Severe+Combined+Immunodeficiency%5BMAJR%5D%29+AND+%28%28x-linked+severe+combined+immunodeficiency%5BTIAB%5D%29+OR+%28scidx1%5BTIAB%5D%29+OR+%28x-scid%5BTIAB%5D%29+OR+%28xscid%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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