What is X-linked severe combined immunodeficiency?

X-linked severe combined immunodeficiency (SCID) is an inherited disease of the immune system and the most common form of SCID. Individuals with X-linked SCID have a deficiency of a protein required for proper development and functioning of the immune system. Signs and symptoms of X-linked SCID are due to the body’s ability to fight infection.

What are the symptoms of X-linked severe combined immunodeficiency and what treatment is available?

Signs and symptoms of X-linked SCID usually appear in the first few months and may include:

- Failure to thrive
- Absent lymph nodes and tonsils
- Recurrent and persistent infections
- Skin rashes
- Chronic diarrhea
- Cough and congestion
- Fevers
- Pneumonia
- Sepsis (a complication of infection)

Treatment includes bone marrow transplantation or gene replacement therapy. Prophylactic antibiotics and immunoglobulin infusions may be helpful. Without treatment, the disease usually leads to death in infancy.

SCID is included in newborn screening panels in some states in the United States.

How is X-linked severe combined immunodeficiency inherited?

X-linked SCID is an X-linked recessive disease caused by mutations in the IL2RG gene. A male who inherits one copy of an IL2RG gene mutation is affected with X-linked SCID. A female who inherits one copy of an IL2RG gene mutation is a carrier and is not expected to have related health problems. A female who inherits two mutations in the IL2RG gene, one from each parent, is affected with X-linked SCID, although this is an uncommon occurrence.

If a female is a carrier, the risk for each son to be affected is 50% and the risk for each daughter to be a carrier is 50%. If a male is affected, each son is unaffected and each daughter is an obligate carrier.

Who is at risk for X-linked severe combined immunodeficiency?

X-linked severe combined immunodeficiency can occur in individuals of all races and ethnicities and has an estimated worldwide incidence of at least 1 in 50,000 to 1 in 100,000. More than 50% of affected male patients do not have a family history, in part due to de novo mutations.

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?
A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

**Where can I get more information?**


**References**