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CARRIER SCREEN

X-linked severe combined immunodeficiency

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.[4] 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.[1]

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:[2]

- 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.[2] Prophylactic antibiotics and immunoglobulin infusions may be helpful.[2] Without treatment, the disease usually leads to death in infancy.[2]

SCID is included in newborn screening panels in some states in the United States.[3]

How is X-linked severe combined immunodeficiency inherited?

X-linked SCID is an X-linked recessive disease caused by mutations in the *IL2RG* gene.[4] 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.[2] More than 50% of affected male patients do not have a family history, in part due to *de novo* mutations.[5]

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?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/x-linked-severe-combined-immunodeficiency

Severe Combined Immunodeficiency: http://www.scid.net/the-scid-homepage/about-scid/

References

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5. Puck JM, *et al.* Mutation analysis of IL2RG in human X-linked severe combined immunodeficiency. *Blood* 1997;89(6):1968-1977.