

Severe combined immunodeficiency (SCID)

What is SCID?

SCID is a rare genetic disorder that causes problems with the immune system's ability to fight infections. The immune system does not develop or work well. Therefore it cannot fight infections like it should and children with SCID have a higher risk to develop serious infections.

SCID is commonly known as bubble boy disease. It affects 1 baby in every 50,000 – 100,000 babies born in the United States. It can be found in all ethnic groups.

How does SCID affect a child?

Babies born with SCID often look normal at birth, but within the first few months of life, they develop serious and sometimes fatal infections. Their immune system is unable to work normally to fight infection because they do not produce the proper number and type of cells and because the cells do not work correctly. The common cold or other common childhood viruses can be very serious infections for babies with SCID.

Babies with SCID also frequently have trouble gaining weight or failure to thrive, and may have chronic diarrhea. Sometimes they have problems with skin rashes.

What causes SCID?

SCID is a genetic condition that can be caused by several different genetic mutations. Sometimes these mutations are carried in the family and sometimes they are new in the baby. These mutations affect T cells and block their normal development. Depending on the type of mutation, B and NK cells, two other types of immune cells, can also be affected.

SCID can be X-linked, meaning it is carried on the X chromosome and primarily affects boys. In this case, the mother carries the mutated gene but is not affected.

SCID can also be autosomal recessive, which means two copies of the gene must be changed for a person to be affected with SCID. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "carriers", with one copy of the changed gene and one copy of the normal gene.

When both parents are carriers, there is a one-in-four (or 25 percent) chance that both will pass the changed gene on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal gene, and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed gene from one parent and a normal gene from the other, making it a carrier like its parents. These chances are the same in each pregnancy with the same parents.

Is there a test for SCID?

Yes. Babies are tested (newborn screening) for SCID before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to test for TREC levels. T cell receptor excision circles (TRECs) are usually found in every newborn's blood. TRECs are made by T cells, which are an important part of the immune system. Babies with SCID have little to no TRECs in their blood.

A positive newborn screen does not mean that your baby definitely has SCID. However, it means that additional blood tests are needed. Low levels of TRECs in the blood can be caused by SCID, prematurity or other, less serious immune disorders. It is also possible for a baby to have a positive newborn screen for SCID, but have a normal immune system. It is also possible that even though your baby has a normal newborn screen for SCID, they may still have or develop problems with their immune system.

What is the treatment for SCID?

The long-term treatment for SCID is restoration of the immune system through bone marrow or stem cell transplant or enzyme replacement. Before the transplant, babies need to be in a protective environment that limits the chance of infections, and they should never be exposed to large crowds of people. These babies should not receive live viral vaccines, like rotavirus, MMR or varicella. They will also need to be treated with immunoglobulin replacement and antibiotics while they are waiting for a transplant

Please consult with your immunologist and/or regional genetic center to determine what further tests and/or treatment will be needed for your baby.

DISCLAIMER: The information contained on this page is not intended to replace the advice of an immunologist medical professional.

Resources:

Immune Deficiency Foundation
110 West Road, Suite 300
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Phone: 800-296-4433
Fax: 410-321-9165
<http://primaryimmune.org/>