The Journal of the American Medical Association

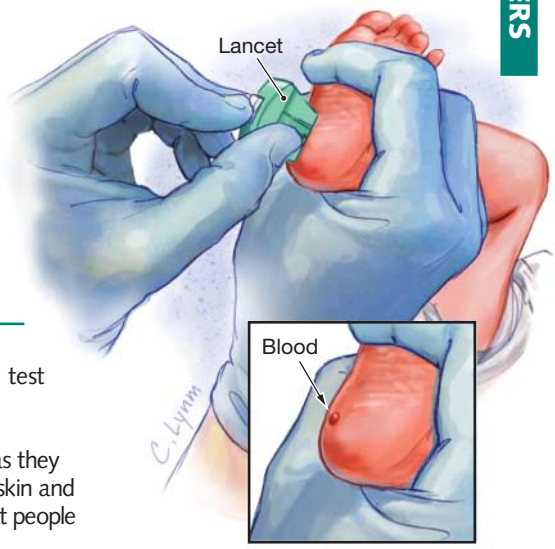
Severe Combined Immunodeficiency

The immune system is made up of the different tissues and cells in the body that fight infections. This system includes the bone marrow, in which the different kinds of white blood cells that fight infection are formed. There are several types of **immunodeficiencies**, or diseases in which the immune system does not work normally. Some immunodeficiencies are **congenital** (present at birth) while others are **acquired** (develop later in life; for example, as a result of infections or medications).

Severe combined immunodeficiency (**SCID**) results from **genetic mutations** (changes in genetic material that can be passed on to children) that result in very small numbers of **T cells** or **B cells** (types of cells required for a normally functioning immune system). Because mutations in any 1 of at least 13 different genes can lead to SCID, there are several genetic types of SCID. The most common form is **X-linked**, which means that the gene is passed from mothers to their sons.

Drawing blood sample for newborn screening

A small puncture of the heel (heelstick) is performed to obtain a sample of blood for testing.



DIAGNOSIS

- In several states, infants are tested for SCID on the **newborn screen**, a blood test performed at birth to check for several diseases that would not otherwise be identified.
- Children with SCID can appear healthy at birth (so SCID is not diagnosed), but as they grow older they experience persistent diarrhea, failure to grow normally, fungal skin and mouth infections, and severe pneumonia from microorganisms that do not affect people with normal immune systems.
- Affected children may also develop severe infections such as **meningitis** (infection of the membranes surrounding the brain and spinal cord) and **sepsis** (bloodstream infections). When young infants develop these complicated infections, an immunodeficiency is likely to be the underlying cause.
- Specialized blood tests in infants with SCID will show an absence of T cells as well as a lack of response to things that stimulate T cells and a lack of antibody **titers** (measures of response) to any vaccines the infant has received.

TREATMENT

- Protection from common infections that affect children with normal immune systems by keeping the infant away from other children
- **Intravenous immunoglobulin** (**IVIG**) is a product derived from human blood that contains **antibodies** (proteins) normally made in the body to fight infections. It can temporarily protect against infections.
- A **bone marrow transplant** should be done as soon as possible to prevent death from severe infections. In this procedure, a donor's healthy bone marrow containing **stem cells** is given to an infant or a child with SCID. These stem cells produce normally functioning T cells and B cells.

FOLLOW-UP CARE

- Children with SCID are cared for by an **immunologist**, a physician who specializes in diseases of the immune system.
- After bone marrow transplantation, children with SCID require vaccinations on a specially designed schedule.

FOR MORE INFORMATION

- Genetics Home Reference
  ghr.nlm.nih.gov/condition/x-linked-severe-combined-immunodeficiency
- National Human Genome Research Institute
  www.genome.gov/13014325
- American Academy of Allergy, Asthma, and Immunology
  www.aaaai.org/conditions-and-treatments/primary-immunodeficiency-disease/severe-combined-immunodeficiency.aspx

INFORM YOURSELF

To find this and previous JAMA Patient Pages, go to the Patient Page link on *JAMA*'s website at www.jama.com. Many are available in English and Spanish.

Sources: Immune Deficiency Foundation; National Human Genome Research Institute; American Academy of Allergy, Asthma, and Immunology

Ann R. Punnoose, MD, Writer
Cassio Lynm, MA, Illustrator
Robert M. Golub, MD, Editor