ADVANTAGES OF GENETIC TESTING

During the evaluation process, it is possible that the altered SCID gene responsible for the baby's illness can be identified. If the gene is the one that causes XSCID, carried by mothers, this finding would give the female relatives of the mother (primarily her sisters) an opportunity to be tested for their own XSCID carrier status. Knowing if a woman carries the XSCID gene could have important implications for their own child-bearing decisions and options.

If it is learned that each parent carries one copy of an altered SCID gene, the brothers and sisters of those parents might also be at risk to carry an alternation in the same SCID gene and choose to be tested. During the genetic testing process, parents may want to take advantage of genetic counseling services so that they can be well informed about future reproductive risks and options.

HOPEFUL OUTCOMES

Parents are always anxious and distressed to learn that their newborn has a serious medical problem. However, they should be assured that the early diagnosis, made possible by newborn screening, provides the best chance for successful treatment of SCID through early intervention.

SUPPORT

As with any baby born with a serious medical condition, there will be frequent medical visits, which can involve many tests and procedures. The process can be very stressful to the entire family. Parents must remember that maintaining their own physical and emotional health is very important, especially while their child is going through treatment. It will be important to identify sources of support and strength, possibly from other family members, close friends, professionals and spiritual associations. The demands on the time and energies of the parents can be overwhelming. We know one of the best sources of support comes from other parents who have traveled this path and can offer first hand encouragement and support gained by their own experience.

FOR PARENTS

Your newborn baby underwent your state's newborn screening for many conditions that could impact your baby's health. As you are aware, the results of the newborn screening test indicated a possible problem with your infant's immune system. After your doctor completed the follow up testing, it was discovered that your baby does in fact have a primary immunodeficiency disease. The primary immunodeficiency diseases are a group of disorders that are caused by a defect in the cells and tissues of the immune system. The condition recently identified in your newborn is known as Severe Combined Immune Deficiency or SCID.

BOTTOM LINE

It is important to remember that the early diagnosis of SCID made possible by newborn screening has provided the best opportunity for successful treatment for your baby.

RESOURCES

Additional information and resources available from:

- **Immune Deficiency Foundation** – offers both information and support for patients and family members including videos from other families impacted by SCID and the IDF Patient & Family Handbook for Primary Immunodeficiency Diseases which contains more detailed medical information about SCID **www.primaryimmune.org**
- **SCID Listserv** – a valuable online support group **www.SCID.net**
- **SCID, Angels for Life Foundation** – offers information and videos from families sharing their own experiences with SCID **www.SCIDangelsforlife.com**
- **American Academy of Allergy, Asthma and Immunology** – offers more detailed medical information about SCID **www.aaaai.org**

IMMUNE DEFICIENCY FOUNDATION

For additional information and references please contact IDF

40 West Chesapeake Avenue, Suite 308
Towson, MD 21204
800-296-4433
410-321-6647
410-321-9165 (Fax)
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NYMAC
NEW YORK-MID-ATLANTIC CONSORTIUM FOR GENETIC AND NEWBORN SCREENING SERVICES

SCID, Angels for Life Foundation

Severe Combined Immune Deficiency (SCID)

A Guide for Parents Following a Diagnosis

THE SCID INITIATIVE

Severe Combined Immune Deficiency (SCID)

A GUIDE FOR PARENTS FOLLOWING A DIAGNOSIS

WHAT IS SCID?

SCID is a rare, serious disorder involving T and B lymphocytes, or T and B cells, the white blood cells that are responsible for fighting infections caused by viruses, bacteria and fungi. Babies with SCID are born with little or no immune system and are at risk for recurrent infections such as pneumonia, meningitis and chickenpox. Without treatment even common infections can be life threatening. Years ago SCID was commonly known as the "Bubble Boy" disease but we know that girls can also be born with SCID. While SCID is a disorder of the immune system it is not related to HIV or AIDS. SCID is not a condition that can be spread from one person to another.

WHAT DO I DO NOW?

Since the mother's antibodies protect a newborn in the first few months of life, a baby born with an immune disorder may not appear ill right away. Even so, while the details of your baby's treatment are being worked out, there are important measures to follow to keep the baby safe from infections.

SAFETY PRECAUTIONS

- Your baby should be isolated from anyone with a cold or infection.
- Avoid public places where there are crowds or children who are not members of the immediate family.
- Everyone should follow strict hand washing or hand sanitizing before touching your baby.

Avoid live virus vaccines

Vaccines such as rotavirus, chickenpox, mumps, measles and live poliovirus should not be given to any baby suspected of having a poorly functioning immune system. In addition, household members should not receive any live vaccines.

Blood or platelet transfusions must be irradiated

If a baby diagnosed with SCID needs a blood or platelet transfusion, the blood product must be irradiated, meaning it must be exposed to an x-ray. This is to prevent a life threatening reaction between any live T cells from the transfusion against the baby's tissues. The blood product also needs to be tested to make sure it is free of the virus called cytomegalovirus (CMV).

Other supporting treatments

Preventive antibiotics may be needed to ward off infections the baby's immune system is unable to handle. Most of these antibiotics can be given by mouth. Babies with SCID are missing an important protective antibody, called immunoglobulin, because their B cells do not make it. However, replacement immunoglobulin can be given through the vein or under the skin to give your baby more protection against infections.

HOW IS SCID TREATED?

The most common treatment for SCID is to correct or replace the baby's immune system through transplantation of blood-forming cells from a healthy donor. Cells in the bone marrow called blood-forming stem cells grow into all the types of blood cells, including lymphocytes (B and T cells). These cells are obtained from a healthy, tissue-matched, person and given to the baby through a vein. The new cells produce functioning lymphocytes that correct the SCID defect. This is called a bone marrow or stem cell transplant.

The ideal donor for an infant with SCID is a perfectly matched sibling. When a perfect match is not available, it is also possible to achieve excellent results using a half-matched related donor, such as a parent. A matched bone marrow or matched cord blood transplantation from an unrelated donor is another successful way to treat SCID.

In some instances, depending on the altered gene that caused SCID in your baby, other treatments are possible and immunology specialists will discuss these with you.

All of these treatment options need to be done in a specialized medical center where there are doctors called pediatric immunologists who are experienced with SCID

WHAT CAUSES SCID?

There are several different forms of SCID. An altered gene, passed to the baby from one or both parents, is associated with many forms of SCID. However, sometimes the SCID gene shows up for the first time in the newborn without being passed down from a parent (spontaneous occurrence). In fact, in most instances of infants born with SCID there are no other known cases of SCID in the family. Unless there has already been a baby born with SCID in the family, there is no way for a couple to know who is at risk to carry a gene that causes SCID.

A faulty gene passed from the mother to her son causes most cases of SCID in males. These boys have X-Linked Severe Combined Immune Deficiency (XSCID), referred to previously as the "Bubble Boy" disease. Other types of SCID are caused by 2 copies of a faulty gene, 1 copy of the faulty gene being passed down from each parent to the baby. Each parent who carries only one copy of the altered SCID gene is healthy, but when the baby inherits 2 altered copies, one from each parent, the function of T, B and Natural Killer (NK) cells is seriously impaired. NK cells are other important cells in the immune system that are so named because they easily kill cells infected with viruses. No matter which gene or genes cause an infant to have SCID, intervention is essential and the sooner it is started the better things will be.