Additional Resources

Immune Deficiency Foundation

The Immune Deficiency Foundation (IDF), founded in 1980, is the national non-profit patient organization dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases (PI) through advocacy, education and research. IDF has a wealth of resources and groundbreaking information developed by a legion of dedicated professionals – healthcare, insurance, education and lifestyle advocates. Because of the generosity of donors and sponsors, most IDF services and resources are provided at no cost. For complete information on all IDF has to offer, please visit www.primaryimmune.org or call 800-296-4433.

IDF Resources for Severe Combined Immune Deficiency (SCID)

- Website: www.primaryimmune.org/SCID
- Blog: www.idfscidnewbornscreening.org
- Materials:
  - *IDF Patient & Family Handbook for Primary Immunodeficiency Diseases*, Chapters Dedicated to SCID and Newborn Screening
  - *SCID: A Parent's Guide* (Brochure), Available in English & Spanish
  - *What does an abnormal screen for SCID mean?* A quick guide for parents (Flier), Available in English & Spanish
- Videos: Multiple IDF videos featuring families living with SCID, including one family's journey through isolation after diagnosis: http://bit.ly/SCIDisolation

Ask IDF – Individualized Assistance for all Living with PI

IDF offers help with the unique aspects of living with PI. Patients can use Ask IDF to answer their questions, receive peer support, help them locate a specialist in their area, and assist them with insurance issues. Go to: www.primaryimmune.org/ask-idf

Join the PI Community – Learn and Share with Others

- IDF Social Networks – IDF Friends, www.idffriends.org, and IDF Common Ground, www.idfcommonground.org, are two exclusive social networks for patients living with PI.
- IDF Get Connected Groups – Individuals and families can meet others living with PI in their local area. To find an upcoming meeting, visit www.primaryimmune.org/events-calendar
- IDF Advocacy Center – Monitor public policy issues that are critical to patients at national and state levels. Learn more at www.primaryimmune.org/idf-advocacy-center

United States Immunodeficiency Network (USIDNET) – Patient Registry and Research Consortium*

USIDNET, a program of the Immune Deficiency Foundation (IDF) funded in part by the National Institute of Allergy and Infectious Diseases (NIAID) and the National Institutes of Health (NIH), is a research consortium established to advance scientific research in the field of PI. The current focus of this initiative is on the patient-consented registry, and education and mentoring for young investigators. Learn more at: www.usidnet.org

Valuable Tools – Improving Health, Powering Research

IDF ePHR, www.idfephr.org, is the electronic personal health record for people with PI to track their health and it gives them the opportunity to consent into PI CONNECT, the IDF Patient-Powered Research Network, www.idfpiconnect.org, which transforms research by bringing together patient data with clinical data in USIDNET.

FILL – Following Infants with Low Lymphocytes

Because your baby has been identified with low lymphocytes through newborn screening, please consider taking part in a research study that will track their diagnoses and outcomes. FILL (Following Infants with Low Lymphocytes) is a program of the Clinical Immunology Society (CIS) and the United States Immunodeficiency Network* (USIDNET) sponsored by the Jeffrey Modell Foundation. Infants with low lymphocytes are being identified, often by newborn screening for Severe Combined Immune Deficiency (SCID). This research study will track their diagnoses and outcomes.

To learn more, visit www.usidnet.org/fill. For more information about how to include your child's information in the FILL program, contact USIDNET: contact@USIDNET.org

SCID, Angels for Life Foundation

Heather and John Smith started this charity in 2008 as a memorial to Heather's first child, Brandon, and his short battle with Severe Combined Immune Deficiency (SCID), and as a tribute to Heather's other son, Taylor, and his long life with SCID. This foundation offers emotional support to affected families while also providing limited financial assistance to families currently going through treatment for SCID.

Interested families can log onto the website at SCIDAngelsforlife.com to learn more about the SCID Angels Family Scholarship Fund and other available resources. Want more information? Contact Heather directly: Heather@SCIDAngelsforlife.com

The SCID Group

The SCID Group is designed to help families dealing with Severe Combined Immune Deficiency (SCID) find a support network of similar families. Go to www.scid.net, and select the "SCID Email Listserv Support Group" to sign up.

** The United States Immunodeficiency Network (USIDNET) is funded by a cooperative agreement, U24AI086037, from the National Institute of Allergy and Infectious Diseases (NIAID) and the National Institutes of Health (NIH), awarded to the Immune Deficiency Foundation (IDF). NIAID supports basic and applied research to better understand, treat and ultimately prevent infectious, immunologic, and allergic diseases.*

Glossary

Combined immunodeficiency – Immunodeficiency when both T- and B-lymphocytes cells are inadequate or lacking.

Cytomegalovirus – Cytomegalovirus (CMV) is a common virus that can infect almost anyone. Most people do not know they have CMV because it rarely causes symptoms. However, if you are pregnant or have a weakened immune system, CMV is cause for concern. Once infected with CMV, your body retains the virus for life. However, CMV usually remains dormant if you are healthy. CMV spreads from person to person through body fluids, such as blood, saliva, urine, semen and breast milk.

DiGeorge Syndrome – DiGeorge Syndrome is a primary immunodeficiency disease caused by abnormal migration and development of certain cells and tissues during fetal development. As part of the developmental defect, the thymus gland may be affected and T cell production may be impaired, resulting in low T cell numbers and frequent infections. DiGeorge syndrome is often, but not exclusively, caused by deletions in chromosome 22 (hence it's other name 22q11.2 deletion syndrome). However, other chromosomal abnormalities, such as deletions on chromosome 10, have also been shown to cause DiGeorge syndrome.

DNA – Deoxyribonucleic acid (DNA), which contains the biological instructions that make each species unique. DNA, along with the instructions it contains, is passed from adult organisms to their offspring during reproduction.

Primary immunodeficiency diseases (PI) – Primary immunodeficiency diseases (PI) are a group of more than 300 rare, chronic disorders in which part of the body's immune system is missing or functions improperly. While not contagious, these diseases are caused by hereditary or genetic defects, and, although some disorders present at birth or in early childhood, the disorders can affect anyone, regardless of age or gender. Some affect a single part of the immune system; others may affect one or more components of the system.

Severe Combined Immune Deficiency (SCID) – Severe Combined Immune Deficiency (SCID, pronounced "skid") is a potentially fatal primary immunodeficiency in which there is combined absence of T cell and B cell function. There are at least 13 different genetic defects that can cause SCID. These defects lead to extreme susceptibility to very serious infections. This condition is generally considered to be the most serious of the primary immunodeficiencies.

T cell receptor excision circle (TREC) – TRECs are made as a type of immune or white blood cells called T cells develop in the thymus gland.

Thymus Gland – The thymus is a lymphoid gland comprised of two identically sized lobes, located behind the sternum (breastbone) but in front of the heart. It derives its name from a resemblance it bears to the bud of the thyme plant (thymus in Latin). At puberty, the thymus reaches the height of its use, becoming its largest.

This brochure was made possible through funding from the Association of Public Health Laboratories.

Immune Deficiency Foundation

Understanding the Low T Cell Results from Your Baby's Newborn Screening

A Guide from the Immune Deficiency Foundation

You just received low T cell results from your baby's newborn screening. ***What does this mean?***

You just received low T cell results from your baby's newborn screening. What does this mean?

Babies can appear healthy at birth and still have health problems that need to be identified and treated. Your baby had routine newborn screening tests done in the hospital before discharge, which included taking a few drops of blood from your baby's heel and testing for a number of disorders. One of the tests can detect problems with the immune system, called the TREC test, and your baby had a result on this test that was either abnormal or did not give a clear result. Therefore, additional testing needs to be done as soon as possible.

What is TREC testing?

TREC testing screens babies for Severe Combined Immune Deficiency (SCID), commonly known as the bubble boy disease, and other low T cell disorders. This test shows that your baby may have a low number of a type of white blood cells, called T cells, sometimes called T lymphocytes. When T cells develop in the thymus gland, TRECs (T cell receptor excision circles) are made.

If the TREC is low in a baby, this raises concern that the baby might have no or very low T cells. These white blood cells are vital components of the immune system to prevent life threatening infections.

It is important to remember that the TREC testing is very useful for screening for low T cells, but it doesn't confirm a diagnosis—it only identifies something is abnormal. As we will describe later, additional testing and advice from your doctors will help to determine if your child does in fact have low or no T cells.

To read more about how TREC testing works, go to www.primaryimmune.org/low-tcell

Why is evaluation and continued follow up by a specialist important?

Primary immunodeficiency diseases are serious genetic medical conditions, and if not properly diagnosed and treated, they can be life threatening.

Severe Combined Immune Deficiency (SCID) is generally considered the most serious of the primary immunodeficiency diseases. Babies with SCID look healthy at birth, but without early treatment, most often by bone marrow transplant from a healthy donor, these infants cannot survive. Affected infants lack T cells that work normally. Both before and after the corrective treatment, babies with SCID are placed in isolation to protect them, or isolate them, from germs that could make them sick—without taking such precautions, a family risks creating irreversible damage decreasing the chances for a successful outcome after transplant. (For more information, see the IDF brochure "SCID: A Parent's Guide," which covers next steps, safety precautions, treatment, causes and more).

In addition to SCID, other disorders associated with low T cells require continued care by a specialist, like a clinical immunologist, who has the knowledge and experience to recognize which treatments are most likely to address each patient's unique needs. Clinical immunologists have undergone specialty training in recognizing, diagnosing and treating disorders associated with low T cells.

Infants definitively diagnosed with SCID are immediately sent for treatment appropriate for their diagnosis. Children without such a definitive diagnosis also require ongoing evaluation and follow up with a specialist to ensure their reason for low T cells is understood and appropriately treated.

What conditions or disorders can be seen in babies with no or low T cells?

The main reason TRECs are tested in newborns is to detect SCID, but other conditions can also be identified. The test can identify:

- Combined immunodeficiency (leaky SCID)
- Congenital abnormalities associated with low T cells
- Prematurity
- DiGeorge Syndrome
- Genetic syndromes associated with low T cells
- Lymphopenia (low lymphocyte counts) of infancy
- Other primary immunodeficiencies
- Secondary T cell deficiency from loss of T cells
- Severe Combined Immune Deficiency (SCID)
- Transient immunosuppression from maternal medication or certain viral exposures during pregnancy

All babies with low T cells should be evaluated by a clinical immunologist to help families and pediatricians decide the best treatment and monitoring plan for each baby's individual needs.

Are low T cells common in babies?

Approximately, 1 in 20,000 babies have low T cells based on the data from newborn screening.

How many babies with an abnormal TREC test have low T cells not associated with SCID?

40-50% of babies with an abnormal TREC test have low T cells. Of these, about two thirds will have low T cells not associated with SCID.

How are the tests completed?

Each state's Department of Health will determine the types and the order of other tests to be completed, so the exact process may vary from state to state. Regardless of the specific details, more definitive testing is needed to establish a diagnosis. You will need to bring your baby in so that a person trained in drawing blood from small babies will collect a small amount of your baby's blood for testing. In general the basic testing will include a complete blood count with differential (CBC with diff) that shows the different types of white blood cells of the immune system. Usually, infants with abnormal TRECs will also have a low Absolute Lymphocyte Count (also known as ALC). Lymphocyte studies will count the number of lymphocytes [this includes T cells and other types of lymphocytes called B cells and natural killer (NK) cells].

To read more about additional testing that your doctor may recommend, go to www.primaryimmune.org/low-tcell

Could your baby eventually be diagnosed with SCID?

It is important to note that some infants who have abnormal TRECs and low T cells at birth eventually normalize over time, without suffering from serious infections or poor health outcomes. However, many infants with abnormal TRECs tests are eventually diagnosed with some form of SCID or other primary immunodeficiency after an exhaustive evaluation.

What should you do for your baby while waiting for a diagnosis? How do I keep my baby from getting sick?

It takes time to perform a thorough evaluation of a baby found to have abnormal TRECs. Some laboratory tests come back quickly, while others, like genetic tests, may take weeks or months. Sometimes doctors want to repeat tests over time, in order to try to find any trends in a baby's cell counts, especially if the numbers are borderline low and/or the baby is not too ill.

As difficult as it might seem, try to stay calm and remember that you are not alone. Your clinical immunologist can help you at every step of the way during this sometimes confusing and challenging process. They are there to provide you with accurate, scientifically sound information so that you can make well-informed decisions for your child.

During that time, the safest course of action is to assume that the child may have an immune problem until proven otherwise. If the child truly has a primary immunodeficiency, then the child is at higher risk for developing infections, which could be life threatening if not recognized and treated appropriately. Thus, it is a good idea to take precautions:

- Ask your immunologist what you should and shouldn't do while your child is still being evaluated.
  - Check with your doctors whenever the child develops signs or symptoms of an infection (for example, fever or cough or diarrhea).
  - Discuss with your immunologist whether or not regular immunizations should be given to your baby. In infants with low lymphocytes immunizations are often delayed until more is known concerning your baby's immune system.
- Ask your immunologist if breastfeeding is okay or if formula feeding is better for the time being, since breastfeeding can expose the baby to an infection called cytomegalovirus (also known as CMV). CMV can be very harmful, especially if your child needs a transplant. If your baby is hospitalized for any reason, be sure that the hospital physicians consult with your immunologist before beginning standard treatments.
- Maintain good hygiene, including frequent hand washing or using hand sanitizer.
- Avoid contact with sick people when you have a newborn. Make sure to ask your immunologist what to do if your baby has an accidental exposure to a sick person, like an older sibling or parent.
- Overall, make sure you have good lines of communication with your clinical immunologist and pediatrician. Don't hesitate to ask questions, and make sure you keep your appointments with your doctors.

Also, don't forget to contact the Immune Deficiency Foundation (IDF) for additional resources and support. Primary immunodeficiency diseases affect many families, and IDF can help you build a support network of folks who understand what you are going through.