# Severe Combined Immune Deficiency (SCID)

## An overview for the APHL

Bethesda North Marriott Hotel & Conference Center
In-Person Meeting
July 31, 2015


Barb Ballard
- Parent
- Patient Advocate
- The Immune Deficiency Foundation, Board of Trustees
- SCID Angels for Life, Executive Board
- SCID Family Network (SCID.net), Administrator

# Severe Combined Immune Deficiency (SCID)

SCID is often called "bubble boy disease"

# Why Early Detection is Important

To make precautions and treatment available before exposure to infection

# Complications from late diagnosis

- 4 months in PICU / 11 months in the hospital
- PCP pneumonia scarred lungs
- GI track damaged by enteral virus and GvH – TPN and g-tube feeds
- Surgeries for chest tubes, IV central lines / ports, g-tube, trach, endoscopies and a bowel resection
- Deafness from antibiotics
- Short Stature / poor growth

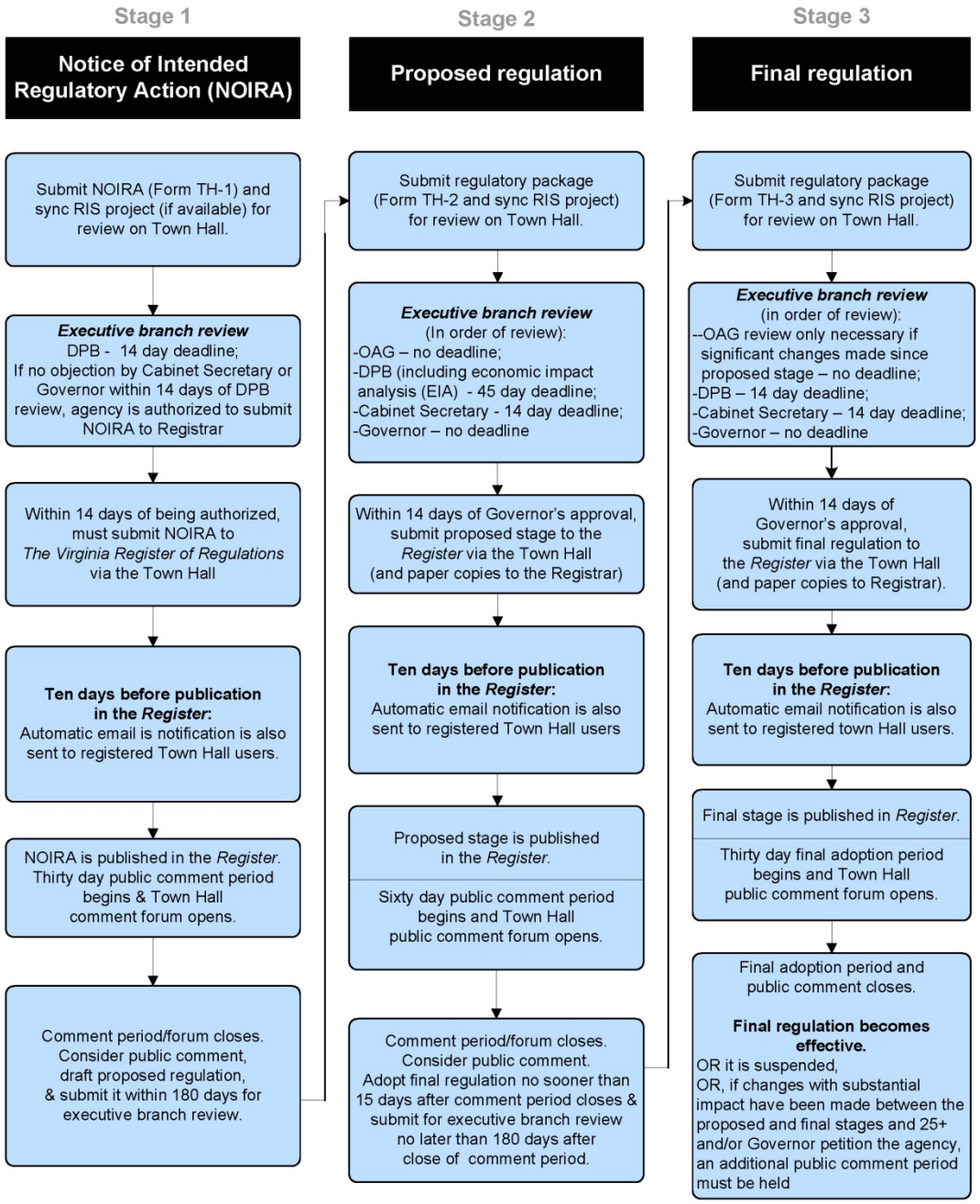
# Costs of Late Diagnosis

- Medical costs maxed out a $2million insurance policy before he was 5 years old.
- Though he survived where many have not, many costly challenges and disabilities could have been prevented
- Benefits of early diagnosis would have been a life without these ongoing costs and challenges

# Newborn Screening Virginia

- Example of a state with little opposition, but which required a lengthy regulatory process
- SCID screening was recommended in 2011 by the VA Genetic Advisory Committee
- Screening began July 2015

# **Standard regulatory process:** Guide for state agencies



# Newborn Screening Louisiana

- In October, 2010 Louisiana began a pilot program to start screening
- Just over 30,000 babies were screened before the state decided to end the pilot in April, 2011
- Since then, little has been done to reinstate a process that saves lives and reduces healthcare costs.

# Louisiana

- 70% of babies born in Louisiana are on Medicaid. If a Medicaid baby is born with SCID, the state is required to pay 50% of the medical costs for that child
- Babies born with SCID, a treatable illness, deserve a chance at a healthy life and implementing testing in Louisiana will save lives AND health care costs

# Resources

- Immune Deficiency Foundation www.primaryimmune.org
  - IDF SCID Initiative primaryimmune.org/programs/idf-scid-initiative/
  - IDF SCID Newborn Screening idfscidnewbornscreening.org/
  - PUBLICATIONS
    - What does an abnormal screen for SCID mean? – A quick guide for parents
    - SCID: A Parents Guide


Newborn Screening for Severe Combined Immunodeficiency (SCID) and Conditions Associated with T Cell Lymphopenia

Your baby had routine newborn screening tests done in the hospital before discharge. A few drops of blood were taken from your baby's heel and tested for a number of disorders that can appear at birth. One of the tests performed detects potential immune disorders. Your baby had a positive (abnormal) result on this test and additional testing is necessary to determine whether your baby has a serious primary immune disorder.

What Does An Abnormal Screening Test Mean?

The screening test shows that your baby has a low number of T cells (T cell lymphopenia). These cells, sometimes called lymphocytes, are a type of white blood cell. Low numbers of T cells can be associated with a genetic condition called Severe Combined Immunodeficiency or SCID (pronounced 'skid') which would place your baby at extreme risk for serious, life-threatening infections. The screening test alone cannot be used to make a diagnosis of SCID, which is why another blood test is needed right away to determine if your baby has a life-threatening immune disorder. Note that babies who are born prematurely can have low T cells at birth. If this is the case, the next blood test will be able to tell if that is the reason. If your baby is premature, it may be recommended that the test be repeated once your baby is full term to determine if a primary immune deficiency is present.

What is SCID?

SCID is actually a set of more than a dozen different disorders, all of which result in a failure of affected infants to develop T cells. It is a syndrome that results from a variety of genetic causes. Although most newborns with SCID appear healthy, the disorder is present at birth. Typically SCID includes problems with both T cell and B cell function, which can result in the onset of one or more serious infections in the first few months of life. T cells originate in the bone marrow, mature in the thymus gland, and then enter into the blood stream, where they help B cells kill infections and regulate the immune system. B cells, another type of white blood cell, are produced in the bone marrow. After they mature into plasma cells they produce immunoglobulins, or antibodies, that keep us well by killing germs such as viruses, fungi, and bacteria.

What Other Diseases or Conditions are Detected by this Test?

In addition to SCID, the screening test also picks up other conditions associated with T cell lymphopenia. This includes Complete DiGeorge Syndrome and other T cell deficiencies, some of which are not well defined. These other genetic disorders can also result in a baby having low T cells at birth. Babies with these disorders can also develop serious infections so additional testing and treatment is necessary.

How Common is SCID?

In past years, it was believed that the incidence for babies born each year with SCID was 1 in 100,000. New data available from states that have initiated newborn screening programs for SCID suggest that the incidence may be closer to 1 in every 30,000 births.

How are SCID and Other Conditions Associated with T cell Lymphopenia Treated?

The most effective treatment for SCID is a bone marrow transplant. This treatment can be done soon after birth and has a high success rate when done in the first few months of life. Some of the other conditions associated with T cell lymphopenia will also be treated with bone marrow transplant, while others may be most appropriately treated with other therapies or a thymus transplant. This is why it is important to follow up with diagnostic evaluation to determine what kind of treatment your baby needs.

SCID INITIATIVE

IMMUNE DEFICIENCY FOUNDATION


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 that parent might also be at risk to carry that 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 video 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)
www.primaryimmune.org
idf@primaryimmune.org

This brochure was developed with support from the New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services, the IDF SCID Initiative and SCID, Angels for Life Foundation.

NYMAC

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A Guide for Parents Following a Diagnosis

THE SCID INITIATIVE

# Additional Resources for Families

- SCID, Angels for Life www.scidangelsforlife.com
- SCID website and listserv as well as private Facebook groups www.scid.net

# Thank You

Thank you for the opportunity to share my family's story