

#### **Newborn Screening FACT Sheet**

## T-Cell Related Lymphocyte Deficiencies

#### What is T-Cell Related Lymphocyte Deficiencies?

T-cell related lymphocyte deficiencies are conditions in which the body's immune system is not working properly. A person's immune system is made up of different parts that work together to protect the body from infection. Babies with T-cell deficiencies are born without enough working T-cells. T-cells are a specific type of white blood cell of the immune system that helps to protect the body from certain kinds of illnesses. Individuals with these T-cell related lymphocyte deficiencies may get repeated infections. Early detection through newborn screening and immediate treatment can help prevent these infections.

## What Causes T-Cell Related Lymphocyte Deficiencies?

Our immune system is made up of special cells and proteins that protect us from illnesses and infections. Specifically, T-cell lymphocytes are one type of white blood cell that plays a key role in the functioning and strength of our immune system.

If your baby has a T-cell related lymphocyte deficiency, then his or her body either does not make enough or makes non-working T-cell lymphocytes. Without these working cells, the immune system cannot function correctly. This leaves the body unprotected from serious infections and illnesses.

T-cell related lymphocyte deficiencies usually have a genetic cause. They may be inherited from parents or can be the result of a new genetic change in the child. Most of the T-cell related lymphocyte deficiencies follow either an autosomal recessive or X-linked recessive pattern of inheritance.

When T-cell related lymphocyte deficiencies are autosomal recessive genetic conditions, a child must inherit two copies of the non-working gene for the deficiency, one from each parent, in order to have the condition. The parents of a child with an autosomal recessive condition each carry one copy of the non-working gene, but they typically do not show signs and symptoms of the condition. While having a child with a T-cell related lymphocyte deficiency is rare, when both parents are carriers, they can have more than one child with the condition.

When T-cell related lymphocyte deficiencies are X-linked recessive genetic conditions, a male must inherit one copy of the non-working gene from his mother to have the condition. A female must inherit two copies of the non-working gene, one from each parent, in order to have the condition. In X-linked conditions, the gene is carried on the X sex chromosome, and the condition affects males more than females. While having a child with a T-cell related lymphocyte deficiency is rare, when one or both parents carry the non-working gene for the deficiency, they can have more than one child with the condition.

# What Symptoms or Problems Occur with T-Cell Related Lymphocyte Deficiencies?

From birth, babies with T-cell related lymphocyte deficiencies may not be protected from life-threatening infections. This is why early screening and identification is so important.

Signs of T-cell related lymphocyte deficiencies include:

- infections that do not get better with antibiotic treatment for two or more months
- diarrhea
- poor weight gain or growth (failure to thrive)
- thrush (a fungal infection) in the mouth or throat that does not go away

If your baby shows any of these signs, be sure to contact your doctor immediately.

# What is the Treatment for T-Cell Related Lymphocyte Deficiencies?

**Isolation** – Your baby may need to be kept away from people other than family members, especially from young children. Isolation reduces the risk of exposure to life-threatening illness.

Immunoglobulin Replacement Therapy - Babies with more severe T-cell related lymphocyte deficiencies may need immunoglobulin replacement therapy. This therapy can replace the missing antibodies that help your baby fight illnesses and infections.

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Transplant Surgeries – For severe, life-threatening T-cell related lymphocyte deficiencies, your child may need a bone marrow or thymus transplant. These are parts of the body that help make illness-fighting cells for the immune system, but they may not be working correctly in children with T-cell related lymphocyte deficiencies. In these transplant surgeries, bone marrow cells or a thymus from a person with a working immune system are given to a person with a T-cell related lymphocyte deficiency. These transplants can strengthen the immune system of the person with the T-cell related lymphocyte deficiency.

#### **Things To Remember**

Each person with T-cell related lymphocyte deficiency has a different experience. Some people may have mild cases, while others experience very severe T-cell related lymphocyte deficiencies. Some cases can be life threatening. With early identification and proper treatment, children with T-cell related lymphocyte deficiencies can avoid many life threatening illnesses and infections. These children can lead healthy lives.

Babies who do not receive treatment for T-cell related lymphocyte deficiency are at risk of catching a life-threatening illness.