



Newborn Screening FACT Sheet

Citrullinemia Type II/Citrin Deficiency (CIT II)

What is Citrullinemia II?

Citrullinemia, type II is a condition in which the body is unable to make citrin, a protein that helps move substances within the cells. These substances are important for breaking down sugars, producing proteins and nucleotides, and allowing for the normal function of the liver. Citrullinemia II is considered an amino acid condition because people with this condition are unable to transport certain amino acids into mitochondria, the energy production centers of the cell. You may also hear citrullinemia II called a urea cycle condition. This name is used to describe conditions that cause ammonia to accumulate in the bloodstream.

Citrullinemia II is not the same condition as citrullinemia, type I. Even though these conditions have similar names, they have different signs, outcomes, and treatments.

If you are looking for information about citrullinemia, type I, go to <http://www.babysfirsttest.org/>.

What Causes Citrullinemia II?

When we eat food, our bodies use a series of enzymes to break it down. Some of these enzymes help move nutrients within the cell. Citrin is one such enzyme. Citrin helps break down carbohydrates and transport certain amino acids, which are the building blocks of proteins. If your baby has citrullinemia II, then his or her body either does not make enough or makes non-working citrin. When citrin does not work properly, your baby's body cannot break down carbohydrates correctly. It causes low sugar levels, high amino acid levels, and high amounts of ammonia in the blood.

Everyone has some amino acids and ammonia in their blood, but high levels can be toxic.

The imbalances affect other enzymes that help break down amino acids and get rid of ammonia through the urine. In citrullinemia II, an amino acid transporter that helps bring certain compounds across mitochondrial membrane no longer works correctly. This enzyme helps break down ammonia so the body can get rid of it through the urine. Without it, ammonia levels build up in the body.

What Symptoms or Problems Occur with Citrullinemia II?

[Symptoms are something out of the ordinary that a parent notices.]

Signs of citrullinemia II can begin in infancy, adolescence, and adulthood. In babies, the signs of citrullinemia II usually begin between one and five months of age.

- yellowish skin and eyes (known as jaundice)
- low birth weight
- delayed growth
- low blood sugar (called hypoglycemia)

Some of these signs may be seen especially when babies with citrullinemia II eat foods that their bodies cannot break down. They can be triggered by long periods of time without eating, illnesses, and infections.

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

What is the Treatment for Citrullinemia II?

1. Dietary Treatments - Babies with citrullinemia II are often treated with a change to a low carbohydrate diet. A nutritionist or dietician familiar with citrullinemia II can help you plan an appropriate diet for your child.

2. Supplements and Medication - Your baby's doctor may prescribe arginine supplements. Arginine is a substance naturally found in proteins, which can help lower ammonia levels in the blood. Everyone has some ammonia in their blood, but citrullinemia II can cause dangerously high levels, and high ammonia levels can be toxic. Your baby's doctor can write you a prescription for these supplements.

Things to Remember

In babies, citrullinemia II is often treated with a change to a low carbohydrate diet. A nutritionist or dietician familiar with citrullinemia II can help you plan an appropriate diet for your child.