



Newborn Screening FACT Sheet

Citrullinemia (CIT)

What is Citrullinemia?

Citrullinemia is a type of amino acid disorder. People with citrullinemia can't rid the body of ammonia. It is made when the body breaks down protein and amino acids.

What Causes Citrullinemia?

Enzymes help start chemical reactions in the body. Citrullinemia is a condition called "urea cycle disorder." It happens when an enzyme called "argininosuccinic acid synthetase" (ASAS) is either missing or doesn't work right. ASAS helps break down amino acids. It also removes ammonia from the body. The amino acid citrulline builds up in the blood when ASAS doesn't work. Ammonia also builds up. Too much ammonia can cause brain damage. It can cause death if untreated.

What Symptoms or Problems Occur with Citrullinemia?

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

Some of the first symptoms of high ammonia are:

- poor appetite
- too much sleepiness or lack of energy
- irritable mood
- vomiting

If untreated, high ammonia can cause:

- muscle weakness
- breathing problems
- problems staying warm
- seizures
- swelling of the brain
- coma, sometimes leading to death

What is the Treatment for Citrullinemia?

1. Low-protein diet and/or special medical foods and formula – The best treatment for citrullinemia is a very low-protein diet (avoid meat, fish, eggs, milk products, nuts and beans). There are medical foods such as special low-protein flours, noodles, and rice available. A dietitian will make a food plan for your child. Dietitians know what the right foods to eat are. The doctor or dietitian may give your baby a special formula with the right nutrients and amino acids. People with citrullinemia should follow their food plan for life.

2. Medication – Medications can also rid the body of ammonia. Children with citrullinemia take these by mouth or feeding tube.

3. Blood tests – Regular blood tests will check your child's amino acid and ammonia levels.

Things to Remember

Call your doctor right away if your child has any of the following:

- loss of appetite
- low energy or too much sleepiness
- vomiting
- fever
- infection or illness
- behavior or personality changes (such as crying for no reason)
- problems walking or balancing
- bad headache

Children with high ammonia often need to be treated in the hospital.