

Citrullinemia (CIT)

(sit-roo-lin-ee-me-ah)

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen shows an increase in a component of protein called citrulline. This result suggests your baby might have a disorder called citrullinemia.

There is another form of citrullinemia that can also cause an increase in citrulline on newborn screening. This form is very rare and is called citrullinemia, type II.

A positive newborn screen does not mean your baby has citrullinemia, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like citrullinemia.

What is citrullinemia?

Citrullinemia affects an enzyme needed to break down certain proteins and remove waste ammonia from the body so it doesn't build-up and cause health problems.

A person who has citrullinemia doesn't have enough enzyme to break down protein containing citrulline or remove ammonia from the body. Both citrulline and ammonia are very harmful to the body and can cause health problems if they build up.

Citrullinemia is a disorder that is passed on, or inherited, from a child's mother and father. Because citrullinemia is a genetic disease, family members are at risk of having citrullinemia too, even if no one in the family has had it before.

What problems can citrullinemia cause?

Citrullinemia is different for each child. Some children have a mild form of citrullinemia with fewer health problems, while other children may have a severe form of citrullinemia with serious complications.

If citrullinemia is not treated, a child might develop:

- Feeding problems
- Sleepiness
- Vomiting
- Muscle weakness
- Seizures
- Swelling of the brain
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for citrullinemia?

Citrullinemia can be treated. The treatment is life-long. Treatment for children with citrullinemia can include:

- Medications to help prevent high ammonia
- Special formula low in protein
- Low protein diet - a dietician helps families set up the best diet for their child

Children with citrullinemia should see their regular doctor, a doctor who specializes in citrullinemia, and a dietician.

Children with citrullinemia can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders
www.rarediseases.org

National Urea Cycle Disorders Foundation
www.nucdf.org

MN Children & Youth with Special Health Needs
(651) 201-3650 or (800) 728-5420

MN Newborn Screening Program
www.health.state.mn.us/newbornscreening

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155, Phone (800) 664-7772, Fax (651) 201-5471