



CITRULLINEMIA (CIT)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called citrullinemia (CIT).

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

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What is citrullinemia?

Citrullinemia affects an enzyme needed to break down certain proteins and remove waste ammonia from the body.

A person with citrullinemia doesn't have enough enzyme to break down protein and remove ammonia from the body. Ammonia is very harmful to the body and can cause health problems if not removed.

Citrullinemia is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause citrullinemia. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can citrullinemia cause?

Citrullinemia is different for each child. Some children have a mild form of citrullinemia with few 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. Treatment is life-long and can include:

- Low protein diet - a dietitian will help you set up the best diet for your child.
- Special formula low in protein.
- Medications to help prevent high ammonia.

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

Prompt and careful treatment helps children with citrullinemia live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening

Nurse Consultant

Toll-free: 1-866-673-9939

newbornscreening@michigan.gov

Michigan Metabolic Coordinating Center

C.S. Mott Children's Hospital, Michigan Medicine:
1-734-764-0579

Children's Special Health Care Services

Toll-free: 1-800-359-3722