

## What is urea cycle disorder?

A urea cycle disorder is a metabolic problem that occurs when too much ammonia accumulates in the blood. Too much ammonia in the blood can lead to behavioral problems, mental retardation, coma or even death. Babies with these disorders need a special diet and/or medication with monitoring. There are different types of urea cycle disorders including citrullinemia and argininosuccinic acidemia; both of these disorders result in excess ammonia in the blood.

## What causes a Urea Cycle Disorder?

It is inherited from both parents and cannot be “caught” like a cold or “given” to another person

## How are urea cycle disorders found?

Shortly after birth, several drops of blood are taken from a baby’s heel. The dried blood sample is sent to the State Department of Health and Senior Services’ Inborn Errors of Metabolism Laboratory, where it is tested for urea cycle disorders and several other conditions. These tests are all part of the State’s Newborn Screening Program. If there is any abnormality, the baby’s doctor and parents are notified.

## Does an abnormal screening test mean that my baby has a urea cycle disorder??

Screening tests always need to be confirmed by additional testing and medical evaluation.

**IF YOU ARE ASKED TO HAVE YOUR BABY RE-TESTED, ACT QUICKLY AND FOLLOW YOUR DOCTOR’S ADVICE**

## What will my baby’s doctor do?

Your doctor may refer you and your baby to a metabolic/genetic specialist. Your doctor and specialist will determine whether your baby is affected by a urea cycle disorder by doing one or more of the following:

- Get a complete medical history and do a physical exam
- Look for symptoms such as poor feeding, vomiting, unusual sleepiness or crying, or seizures
- Take a blood sample to confirm the screening results.

## How are urea cycle disorders treated?

Treatment should be provided by health care professionals who have experience with urea cycle disorders. A special diet and medication may be prescribed and special precautions may need to be taken if the baby is sick. Urea cycle disorders are permanent and lifetime treatment is necessary.

For more information, contact:

The New Jersey Department of Health  
and Senior Services

Newborn Screening and Genetic  
Services at (609) 292-1582

or

The Inborn Errors of Metabolism  
Laboratory at (609) 292-3090

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## Urea Cycle Disorders

Citrullinemia  
Argininosuccinic acidemia

Important Information  
For Parents



