

## Overview of Newborn Screening for Homocystinuria – For Parents

### What is newborn screening?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to test for a group of conditions. One of these conditions is **homocystinuria**. Babies who screen positive for homocystinuria need follow-up tests done to confirm they have homocystinuria. **Not all babies with a positive newborn screen will have homocystinuria.**

### What is homocystinuria?

When a person has homocystinuria, his/her body is not able to break down **methionine**. Methionine is an **amino acid** (one of the “building blocks” used to make proteins).

Most people with homocystinuria are missing an **enzyme** (a protein that helps our bodies function) called **cystathionine beta-synthase** (also called CBS). When the CBS enzyme is missing, a person cannot break down methionine. People with homocystinuria have high levels of methionine and other proteins in their bodies.

There are other types of homocystinuria that occur when a person is missing other enzymes that help our bodies break down or use methionine. These types of homocystinuria are very rare.

### What causes homocystinuria?

Homocystinuria is an **inherited** (passed from parent to child) condition. Everyone inherits two copies of the gene for cystathionine beta-synthase. We inherit one copy of the CBS gene from our fathers and one copy from our mothers. Sometimes these genes have changes (also called mutations) that prevent the gene from working correctly. In order for a person to have homocystinuria, he or she must have two CBS gene changes. People with one CBS gene change do not have homocystinuria.

### What are the symptoms of homocystinuria?

Every child with homocystinuria is different. Most babies with homocystinuria will look normal at birth. Symptoms of homocystinuria can appear during childhood if a baby with homocystinuria does not receive treatment.

Some of the symptoms of untreated homocystinuria include:

- Dislocation of the lens of the eye
- Mental retardation or psychiatric disorders
- A higher risk of blood clots
- Vision problems
- A tall, thin body with long arms & legs
- Arachnodactyly (long, “spidery” fingers)
- Other skeletal (bone) changes, such as scoliosis (a curve of the spine)

### What is the treatment for homocystinuria?

There is no cure for homocystinuria. However, there are treatments that can help with the symptoms. People with homocystinuria may be given **vitamin B6** or a medicine called **betaine** to help their bodies break down and use methionine. People with homocystinuria may also need to drink a special formula and low protein diet that contains low levels of methionine. A person with homocystinuria will need treatment for his/her entire life.

### What happens next?

Although there is no cure for homocystinuria, good medical care makes a difference. Children with homocystinuria should see a metabolic geneticist (a doctor who specializes in homocystinuria and other related conditions) as well as their pediatrician. Your child’s doctor will work with the metabolic geneticist to coordinate any treatment, tests, or appointments that your child needs.

### Where are Indiana’s metabolic genetics clinics?

Indiana’s metabolic genetics clinics are located at Riley Hospital for Children at IU Health, Indianapolis, IN, (317) 274-3966 and The Community Health Clinic, Topeka, IN, (260) 593-0108.

### Where can I get more information about homocystinuria?

- **STAR-G** - <http://www.newbornscreening.info/Parents/aminoaciddisorders/CBS.html>
- **Region 4 Genetics Collaborative** – <https://www.region4genetics.org/education/families/>