MICHIGAN Newborn Screening

Family Fact Sheet

Homocystinuria *(HCY)*

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 homocystinuria.

A positive newborn screen does not mean your baby has homocystinuria, 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 Homocystinuria?

Homocystinuria affects an enzyme needed to break down proteins in the food we eat, so they can be used for energy and growth. In homocystinuria, the enzyme used to break down proteins is missing or not working properly.

A person with homocystinuria doesn't have enough enzyme to break down protein containing homocysteine. When the body can't break down the homocysteine, it builds up in the body and causes health problems.

Homocystinuria 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 homocystinuria. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can Homocystinuria cause?

Homocystinuria is different for each child. Some children with homocystinuria have few health problems, while other children may have a more severe form of homocystinuria with serious complications.

If homocystinuria is not treated, a child might develop:

- Dislocated eye lens
- Increased risk of blood clots
- Brain damage
- Bone problems
- Osteoporosis

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

What is the treatment for Homocystinuria?

Homocystinuria can be treated. Treatment is life-long and can include:

- Low protein diet - a dietician will help you set up the best diet for your child.
- Medications (Vitamin B6) to help break down proteins.
- Surgery for dislocated eye lenses or bone problems.

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

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

Michigan Resources and Support

Michigan Newborn Screening
Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Children's Hospital of Michigan Metabolic Clinic
Toll-free: 1-866-442-4662

Children's Special Health Care Services
Toll-free: 1-800-359-3722

MDHHS Michigan Department of Health & Human Services
RICK SNYDER, GOVERNOR | NICK LYON, DIRECTOR

Michigan Newborn Screening Follow-up
Phone 1-866-673-9939
www.michigan.gov/newbornscreening
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