



PHENYLKETONURIA (PKU)

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 PKU.

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

PKU affects an enzyme needed to break down proteins from the food we eat. In PKU, the enzyme used to break down proteins is missing or not working properly.

A person who has PKU doesn't have enough enzyme to break down protein containing phenylalanine (PHE). When the body can't break down the PHE, it builds up in the body and causes health problems.

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

What problems can PKU cause?

PKU is different for each child. Some children with PKU have only a few health problems, while other children may have very serious complications.

If PKU is not treated, a child might develop:

- Hyperactivity
- Restlessness
- Seizures
- Behavior problems
- Brain damage

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

What is the treatment for PKU?

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

- Diet low in PHE — a dietitian will help you with the best diet for your child
- Special formula low in PHE

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

Prompt and careful treatment helps children with PKU 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