

Phenylketonuria (PKU)

(fee-null-kee-tone-yer-ee-ah)

FAMILY FACT SHEET POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen shows an increase in a component of protein called phenylalanine (PHE). This result suggests your baby might have a disorder called PKU. There are forms of PKU that can also cause an increase in PHE on newborn screening. These other forms are very rare.

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

Your baby's doctor will help arrange for more testing by specialists in disorders like PKU.

What is PKU?

PKU affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. In PKU, an 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), so harmful toxins can build up in the body.

PKU is a disorder that is passed on, or inherited, from a child's mother and father. Because PKU is a genetic disease, family members are at risk of having PKU too, even if no one in the family has had it before.

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 serious complications.

An infant with PKU might be late in learning to sit, crawl, and stand. Older children may need extra help in school. If PKU is not treated, a child might also develop:

- Hyperactivity
- Learning disabilities
- Seizures
- Mental retardation

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

What is the treatment for PKU?

There are treatments for children with PKU, which are life-long. Treatments for children with PKU can include:

- Special formula low in PHE
- Diet low in PHE - a dietician helps families set up the best diet for their child

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

Children with PKU can benefit from prompt and careful treatment.

Resources for parents

National PKU News
www.pkunews.org

Children's PKU Network
www.pkunetwork.org

MN Children & Youth with Special Health Needs
(651) 201-3650 or (800) 728-5420

MN Newborn Screening Program
www.health.state.mn.us/newbornscreening

