Newborn Screening and Phenylketonuria
A Parent's Guide

Indiana Department of Health

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What is newborn screening?

Soon after birth, babies have a small amount of blood taken from their heel. The blood is used to test for some rare but treatable conditions. This test is part of the Indiana Newborn Screening Program.

Your baby's newborn screen was positive for a condition called phenylketonuria (also called PKU). This does **not** mean your baby definitely has PKU, but your baby needs more testing to know for sure. While you wait for more test results, this sheet can help you learn more about PKU.

What is phenylketonuria?

Phenylketonuria, or PKU, is a condition present at birth. It affects how babies break down a part of food called protein. Protein is made up of small parts called amino acids. One of these amino acids is called phenylalanine, or Phe. Phe is in most protein foods, including breast milk and infant formula.

Most people have a special helper in their body called an enzyme that breaks down Phe. This enzyme is called phenylalanine hydroxylase, or PAH. Babies with PKU do not have enough of this enzyme. That means that Phe can build up in their blood and brain when they eat food containing Phe. Too much Phe can cause brain damage and other health problems if babies are not treated.

There is a milder type of PKU called hyperphenylalaninemia, or hyperphe. People with hyperphe may not need treatment.

Spot the Signs

Every baby with PKU is different. Most babies with PKU look healthy at birth.

If a baby with PKU does not get treatment, symptoms of PKU can start in the first six months of life.

Symptoms include:

- Learning problems (intellectual disability)
- Seizures
- Eczema
- A mousy or musty body smell
- Light skin and hair

Babies who start treatment early can grow and learn like other children. Some may still need extra help with learning or behavior.

What causes phenylketonuria?

Phenylketonuria is passed from parents to their baby. A baby with PKU gets one changed PAH gene from each parent. These changes stop the body from making enough of the PAH enzyme. Babies who only have one changed PAH gene do **not** have PKU.

What is the treatment for phenylketonuria?

There is no cure for PKU, but starting treatment early – within the first seven to 10 days – can prevent most problems.

Babies with PKU should drink a special medical formula that does not have Phe. This formula gives them the nutrition they need. Since breast milk has some Phe, your baby's doctor can help decide how much breast milk is okay.

Your baby will also have regular doctor visits to check their Phe levels, growth, and development. As they grow, children with PKU eat a low protein diet and may take medicine to help keep Phe levels safe. People with PKU must follow this treatment for life.

What happens next?

For now, follow the doctor's orders for feeding your child. If the test results are positive again for PKU, your baby's doctor will explain what to do next.

Babies with PKU will see a metabolic geneticist (a doctor who helps care for babies with PKU) and may see a dietitian, an expert who can help babies with food and nutrition. Your baby will also keep seeing their regular doctor (pediatrician). These doctors will help with the tests and treatments that your baby needs.

Thanks to newborn screening, babies with PKU can get early care and live a healthier life.

Where are Indiana's metabolic genetics clinics?

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

baby's first test

NORD® National Organization for Rare Disorders

For additional information, visit:
rarediseases.org/rare-diseases/phenylketonuria/

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