NORTH CAROLINA NEWBORN SCREENING PROGRAM

Phenylalanine hydroxylase (PAH) deficiency

Includes phenylketonuria (PKU)

Fact Sheet for Parents

All babies born in North Carolina are screened at birth to look for certain diseases or other health problems that can be treated if caught early. The newborn screening result showed that your baby might have phenylalanine hydroxylase (PAH) deficiency. Your baby will be referred to a specialist for more testing to know for sure.

There are usually no signs of PAH deficiency at birth.

What is PAH deficiency?

PAH deficiency is a rare genetic condition that prevents the body from breaking down an amino acid called phenylalanine. PAH deficiency is a spectrum ranging from mild to severe. Phenylketonuria (PKU) is the most common type of PAH deficiency. Hyperphenylalaninemia is a milder form of PAH deficiency, and pterin deficiencies are related disorders. In individuals with these disorders, phenylalanine builds up in the blood and can cause health problems. Early diagnosis and treatment of PAH deficiency will prevent developmental or intellectual delays and other serious health problems.

What are the symptoms of PAH deficiency?

Symptoms of PAH deficiency may not be visible at birth but can develop within a few months of life if not treated. Symptoms can include:

- A musty odor in the urine and breath
- Skin rashes
- Development delays
- Intellectual delays
- Seizures

What happens next?

Your baby's pediatrician will refer your baby to a pediatric metabolic specialist familiar with PAH deficiency at UNC Chapel Hill Genetics and Metabolism PKU clinic. Your baby will receive follow-up testing to confirm a diagnosis.

How is PAH deficiency treated?

Individuals with PAH deficiency may need to follow a diet low in phenylalanine. Phenylalanine is found in many foods, especially those that are high in protein such as eggs, milk, cheese, nuts, soy, beans, meat, and fish. Most individuals with PKU also take a PAH/PKU formula to make sure they get enough essential proteins and nutrients to grow and develop. Once seen in clinic, babies with PAH deficiency may start a low-protein diet and should follow this diet for their lifetime. Babies and children who follow the diet advised by their medical team typically grow and develop normally. Your baby's health care provider may recommend additional medications as your child develops and grows into adulthood.

Where do I go for more information?

Use your phone's camera to scan the QR codes below.

Newborn Screening Information Center

Baby's First Test

National PKU Alliance

MedlinePlus

UNC Pediatric Genetics & Metabolism

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services Division of Public Health

www.ncdhhs.gov

https://slph.dph.ncdhhs.gov/newborn/default.asp

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This fact sheet was supported by Health Resources and Services Administration (HRSA), Department of Health and Human Services (HHS) as part of a financial assistance award totaling $344,348, with 100 percent funded by HRSA/HHS. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA/HHS or the U.S. Government.