

For more information, please see:  
<http://nj.gov/health/fhs/nbs/index.shtml>

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**DISCLAIMER:**

THIS INFORMATION DOES NOT PROVIDE  
MEDICAL ADVICE.

After reading this information sheet, you are encouraged to review the information carefully with your doctor or other healthcare provider. The content is not intended to be a substitute for professional medical advice, diagnosis or treatment. NEVER DISREGARD PROFESSIONAL MEDICAL ADVICE OR DELAY IN SEEKING IT BECAUSE OF SOMETHING YOU HAVE READ ON THIS INFORMATION SHEET.



**PHENYLKETONURIA  
( PKU)**

**Important Information  
For Parents**

## What is PKU?

Phenylketonuria (PKU) is an inherited metabolic disorder. A baby with PKU cannot use phenylalanine (“phe” for short). “Phe” is an essential amino acid that is a part of all food proteins. When “phe” builds up in the body, it can prevent the brain from developing as it should. This can result in mental retardation.

Babies born with PKU may appear normal for the first few months. If untreated, in three to five months, they may begin to show signs of the disorder. The severe mental retardation and other problems due to PKU can be prevented by early diagnosis and proper diet control.

## What causes PKU?

A child inherits PKU from both parents. It cannot be “caught” like a cold and cannot be “given” to another child.

## How is PKU found?

Shortly after birth, several drops of blood are taken from a baby’s heel. The dried blood sample is sent to the State Department of Health and Senior Services’ Newborn Screening Laboratory, where it is tested for PKU and several other conditions. These tests are all part of the State’s Newborn Screening Program. If there is any abnormality, the baby’s doctor and parents are notified.

## Does an abnormal screening test mean that my baby has PKU?

Screening tests always need to be confirmed by additional testing and medical evaluation.

**IF YOU ARE ASKED TO HAVE YOUR BABY RE-TESTED, ACT QUICKLY AND FOLLOW YOUR DOCTOR’S ADVICE**

## What will my baby’s doctor do?

Your baby’s doctor will work with a metabolic/genetic specialist. In addition to other things they may do the following:

- Get a complete history and do a physical exam.
- Look for signs and symptoms of vomiting, irritability or convulsions.
- Obtain other blood and/or urine tests to confirm the screening results and to help to make a diagnosis.

## How is PKU treated?

Prompt treatment is necessary. Treatment should be provided by a health care professional who has special expertise caring for individuals with PKU. At the present time, a diet low in phenylalanine is the only treatment for PKU. If the diet is started early enough and closely followed, the severe problems associated with PKU can be prevented.

Your baby’s doctor is the best source of information regarding your baby’s health. If you have any questions about the test results or about PKU, contact your baby’s doctor for more information.