

WHAT IS PHENYLKETONURIA?
(*fennel-key-ton-uria*)

Phenylketonuria is called PKU for short. It is an inherited condition that makes a child unable to use protein from foods in the right way. The problem concerns one part of protein. It is an amino acid called phenylalanine (Phe). Amino acids are building blocks for body growth and repair.

When a person eats foods with protein, enzymes break down the protein into amino acids. Enzymes help start chemical reactions in the body. PKU happens when an enzyme that breaks down Phe does not work properly. As a result, Phe, builds up in the blood and causes problems. Phe is found in almost every food, except pure fat and sugar.

WHAT CAN PKU CAUSE?

A child with PKU who doesn't get treatment will become mentally retarded. Other problems include:

- restlessness or fussiness
- hyperactivity (overactive)
- seizures
- eczema (itchy areas of skin that become flaky or hard)
- a "musty" or "mousy" body odor
- light hair and skin

HOW DO CHILDREN GET PKU?

Children inherit PKU from their parents. Inherited characteristics such as eye color, hair color, and PKU are carried on special parts of the cell called genes. Everyone carries four or five different

abnormal genes without showing signs of any of the disorders. PKU occurs in a child who has two genes for PKU, one inherited from the father and the other from the mother. It is estimated that one in every 70 persons is a carrier for PKU and that the disorder affects one in every 15,000 to 20,000 infants born in the United States.

WHAT IS THE TREATMENT FOR PKU?

A Special Diet: A diet low in Phe is the only treatment for PKU. Some Phe is needed for growth. Too much Phe is harmful. Each child needs different amounts of Phe in his or her diet. The diet for each child is specially planned to make sure there is the right amount of Phe. Special low-Phe medical formulas help give babies and children with PKU the nutrients and protein they need. Children with PKU can also have foods low in protein and Phe. The amounts of these foods have to be carefully measured. These foods include most fruits and vegetables, some cereals and candies, and special breads, cookies, and pastas. All high-protein foods such as milk, meat, eggs, and cheese must not be eaten at all. Regular blood tests measure the amount

of Phe in the blood. The Phe level shows if the diet needs to be changed. The diet may be changed as needed by your doctor and dietitian. Dietitians know the right foods to eat. A diet record needs to be kept so the dietitian knows what to change in the diet if needed.

THINGS TO REMEMBER

- Children with PKU who start treatment soon after birth usually have normal growth and intelligence.
- It is very important that the special PKU diet is followed at all times. Everyone who comes in contact with your child should know about the diet and help your child follow it.
- This diet should be followed at least through adolescence and possibly for life. A young woman with PKU will need to be very strict about her diet both prior to and during pregnancy.
- PKU cannot be cured but can be treated.
- PKU is an inherited condition.
- Regular blood tests are necessary.

WHERE TO GET HELP

The Texas Department of State Health Services provides certain services free of charge to residents of Texas who have PKU. Upon written request by your child's doctor, a public health dietitian may provide help with the diet. Would you like more information? Call the Texas Department of State Health Services Newborn Screening Program free of charge at 1-800-252-8023. Or visit our website at www.dshs.state.tx.us/newborn/pku.htm



Texas Department of State Health Services
Newborn Screening Program
1100 W. 49th Street
Austin, Texas 78756
1-800-252-8023 ext. 2129
www.dshs.state.tx.us/newborn

