



## PHENYLKETONURIA - (PKU) INFORMATION FOR PARENTS/CARERS

### What is Phenylketonuria (PKU)?

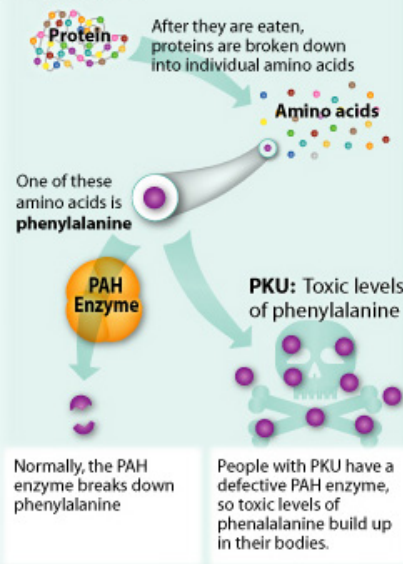
PKU is an inherited disorder which prevents the normal use of protein food, and causes changes in the body chemistry which without treatment can lead to severe mental retardation.

### How is PKU detected?

All newborn babies are screened for several treatable disorders by a blood test taken at about three days of age. Phenylketonuria (PKU) is one of them. About 1 in 10,000 – 14,000 has PKU.

### How does PKU cause problems?

#### People with PKU Have a Defective PAH Enzyme



PKU is a condition in which the body is unable to break down one of the protein building blocks from food. These building blocks are called amino acids and one of them is phenylalanine. In PKU the phenylalanine cannot be processed (metabolised) normally and builds up in the blood and tissues. The high phenylalanine level can prevent the brain from developing as it should. Progressive mental retardation results if the condition is not treated in early infancy.

### What are the symptoms of PKU?

Babies born with PKU usually have no symptoms at first. But if the disease is left untreated, babies experience severe brain damage. This damage can cause epilepsy, behavioral problems, and stunt the growth of the baby. Other symptoms include: eczema (skin rash), a musty body odor (from too much phenylalanine), a small head (microcephaly), and fair skin (because phenylalanine is necessary for skin pigmentation).

### How do doctors diagnose PKU?

Because PKU must be treated early, babies in must be routinely tested for the disease. A small blood sample is taken from the baby's heel or arm and checked in a laboratory for high levels of phenylalanine.

### How is PKU treated?



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People who have PKU must eat a protein-free diet, because nearly all proteins contain phenylalanine. Infants are given a special formula without phenylalanine. Older children and adults have to avoid protein-rich foods such as meat, eggs, cheese, and nuts. They must also avoid artificial sweeteners with aspartame, which contains phenylalanine.

## Why did my child get PKU?

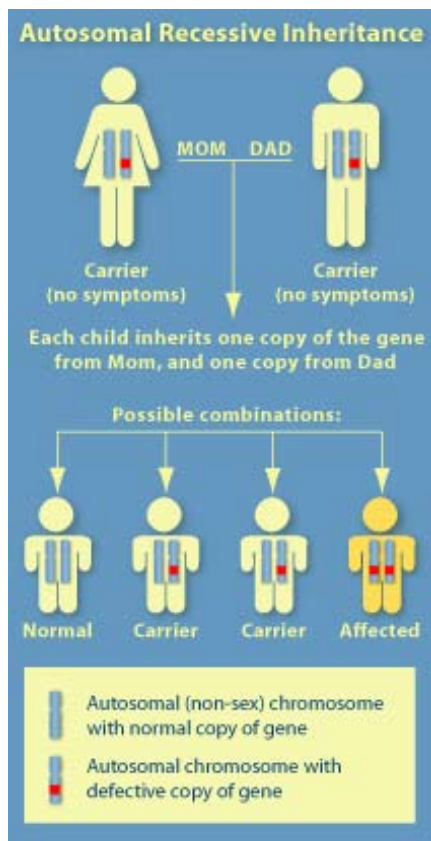
PKU is an inherited disorder, which occurs when both parents have a mutation (mistake) copy of the PKU gene and pass it on to their baby. Genes are the particles of heredity which occur in all cells in the body, and form the "blue-prints" for all the bodily processes and traits such as hair colour, blood group and so on.

A parent who has one copy of a 'PKU' gene and one copy of a normal gene is a carrier of PKU but is perfectly healthy. When both parents are carriers there is a one in four chance in each pregnancy that their baby will inherit a 'PKU' gene from each parent, and so be born with PKU.

This type of inheritance is called autosomal recessive inheritance, and is explained more fully in the PKU handbook you will receive when you come to the hospital. Most families cannot 'trace' PKU in any of their relatives.

There is nothing the parents of a child with PKU could have done to prevent their child **having PKU**.

## How do people get PKU?



PKU is an autosomal recessive disorder, meaning that you need to inherit mutations in both copies of the gene to develop the symptoms of the disorder. A carrier does not have symptoms of the disease, but can pass on the defective gene to his or her children. If both parents carry one copy of the faulty gene, each of their children have a 25 percent chance of being born with the disease.

## Is my child already damaged (before the diet was started)?

Your child is not damaged at all as the diagnosis of PKU is made early in the newborn period. Your baby would have had normal phenylalanine levels while in utero. This is because the mother's metabolism can clear phenylalanine before it accumulates. Only after birth does the phenylalanine level rise.





### **Will my child grow normally?**

Children with PKU treated from early infancy grow and develop quite normally providing the treatment is followed. A special low phenylalanine diet is given, and regular blood tests to measure the phenylalanine level are sent to the laboratory.

The diet for PKU is essential in childhood to prevent damage to the growing brain.

Continuing the diet during adolescence and adult life gives the best outcome in school performance, concentration and ability to think clearly. Apart from needing a diet, children with PKU should be treated exactly as other children. They are neither more nor less likely to get colds and coughs or other illness. Routine immunisations should be given at the usual times. Most medicines can be given safely, but check with your PKU clinic doctor.

When your baby is small, the diet is relatively easy, and allows you to breast or bottle feed plus give the special protein supplement the dietician will tell you about when you come to the hospital.

If your child is diagnosed with PKU, the family may be admitted to hospital for two nights so that confirmatory tests can be carried out and the PKU team can meet with you and discuss the disorder carefully so you fully understand it. The team consists of your clinician, dietician, social worker, clinical nurse consultant for metabolic disorders and the laboratory staff.

### **What about other children in the family**

Both the mother and father of a child with PKU 'carry' the disorder, even though they are perfectly healthy. In each pregnancy there is a 1 in 4 chance that the baby will be affected.

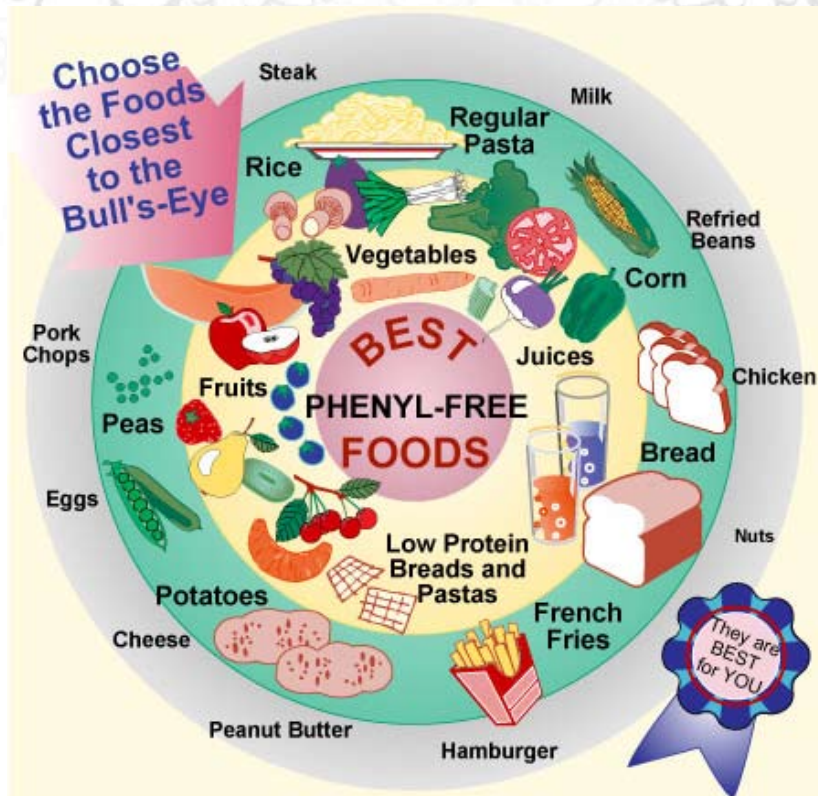
### **What is the Diet for PKU?**

The diet for PKU consists of a milk substitute or formula such as Phenyl-Free 2\* and measured amounts of fruits, vegetables, bread, pasta and cereals.

Many foods must be eliminated from a low phenylalanine diet. These foods are high protein foods such as milk and dairy products, meat, fish, chicken, eggs, beans and nuts which contain large amounts of phenylalanine. Eating these foods will cause high blood phenylalanine levels.

The target is an easy way to visualize the foods allowed on the diet for PKU. The phenylalanine-free formula is the center of the target diet. As the foods get farther away from the bull's-eye they are higher in phenylalanine. The foods outside the target are not allowed on the low-phenylalanine meal plan at all.





It is not unusual for someone on a phenylalanine restricted diet to have two kinds of vegetables and a baked potato for dinner. However, if these foods were all a person on a phenylalanine restricted diet consumed, their diet would be lacking protein, vitamins and minerals. That is where the special formula comes in.

A special formula, such as Phenyl-Free 2\*, contains protein, vitamins, minerals and calories with no phenylalanine. With formula, a person with PKU gets plenty of protein and doesn't get the side effects of the high phenylalanine content of most foods. The phenylalanine-free formula is the most important part of the diet for PKU. Another important part of the diet is low protein breads and pastas. They are nearly free of phenylalanine, allow greater freedom in food choices, and provide energy and variety in the diet.

