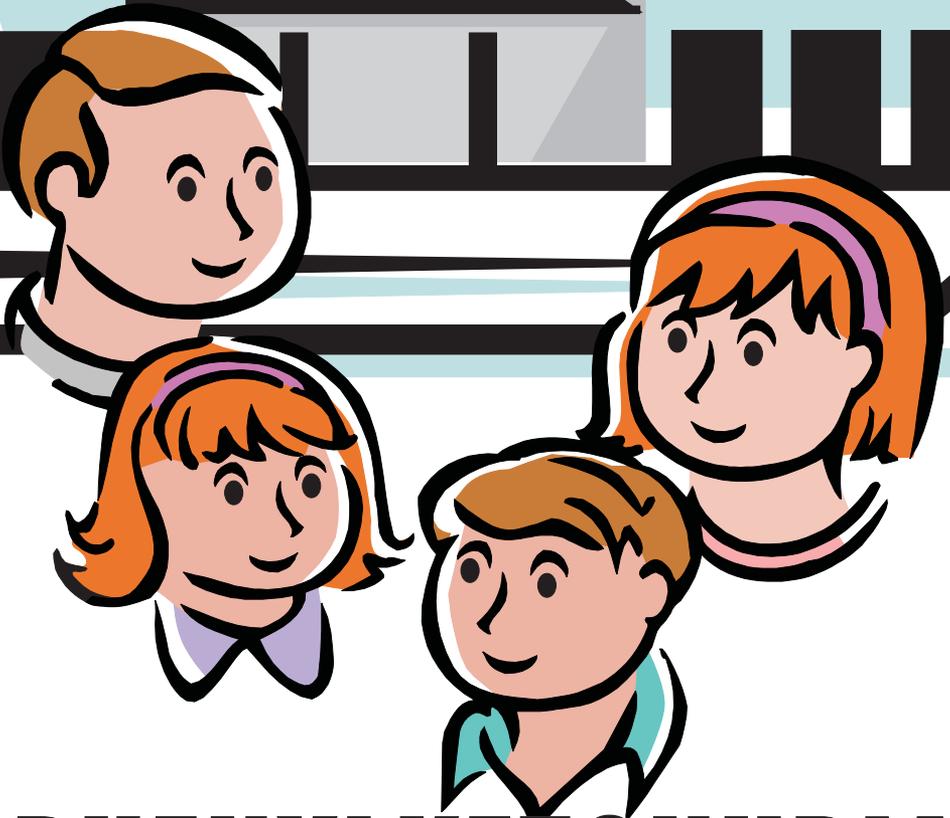


TREATMENT CENTER



**PHENYLKETONURIA**  
**P K U**



# INTRODUCTION

This booklet is written for individuals with phenylketonuria (PKU) and their families and friends. This booklet is to help people learn more about PKU; the cause; and the treatment.

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# PHENYLKETONURIA (PKU) TERMS

## 1. PHENYLKETONURIA

- Also known as PKU,
- Means phenylketones in the urine,
- Phenylketones are chemicals, which build up in the bodies of people with PKU. The phenylketones come from phenylalanine.
- People with PKU have hyperphenylalanemia.
- HYPER - too much
- PHENYLALANINE (PHE) - a building block
- EMIA - in the blood
- Means too much PHE in the blood,
- All people have some PHE in their blood. Most people have a small amount, others have more than normal.
- Anyone who has more than the usual amount of PHE in the blood is said to have PKU.

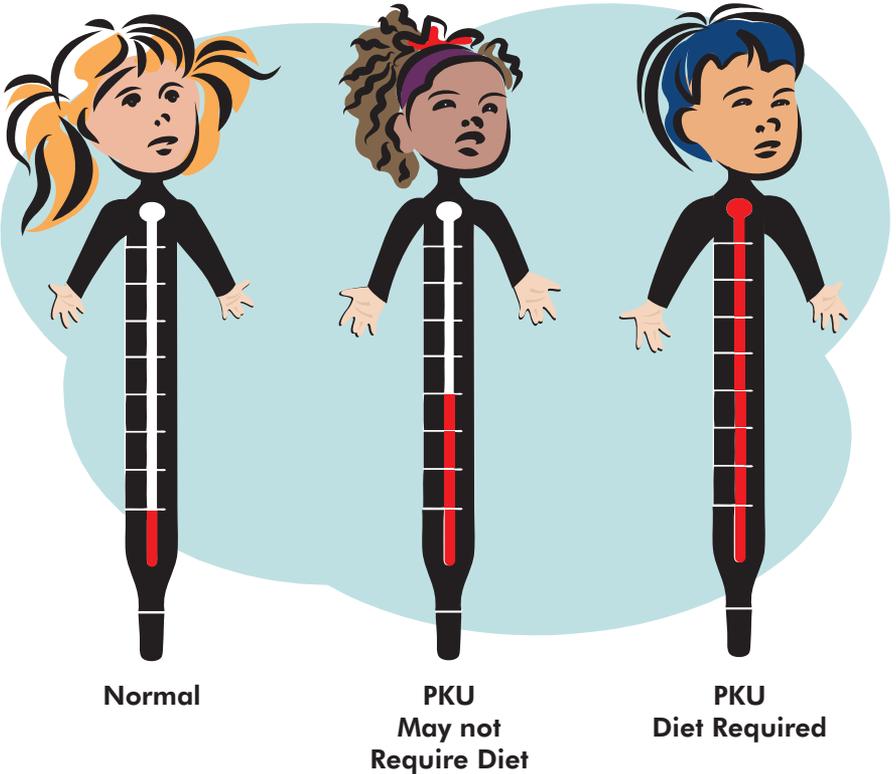
## 2. PHENYLALANINE

- Also known as PHE,
- Is a building block of protein. The protein in our bodies have 20 kinds of these building blocks, called amino acids, which can be put together in different ways to make all of the body proteins - hair protein, muscle protein, brain protein, and many others. The building blocks for proteins are obtained from foods we eat. PHE is in all our body proteins.

## TYPES OF PKU

There are different levels of hyperphe in PKU patients. They differ from one another according to how much extra PHE is present after eating high protein food.

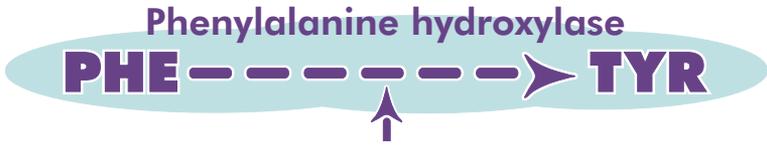
- Some people with PKU have a lot of PHE in their blood and require a PKU diet.
- Some people with PKU have less PHE in their blood and may or may not require a PKU diet.





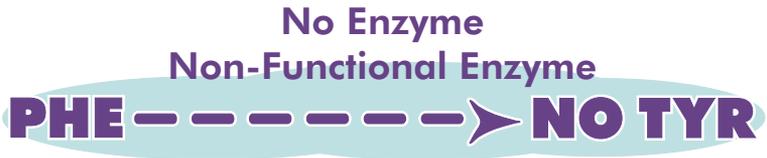
Chemical reactions usually require an enzyme for the reaction to work.

Above the arrow we write the name of the enzyme.



How are people with PKU different from most other people? Their livers are otherwise healthy, but they cannot make PHE into TYR. The chemical reaction will not work because people with PKU are missing the needed enzyme:

- Since PHE is not made into TYR, it builds up in the body. Some of it is changed into other substances, such as phenylketones, but most of it remains as PHE in the body.



- In rare individuals, a co-factor deficiency could lead to elevated PHE levels. Your child can be screened for co-factor deficiencies by a blood test and a urine test.
- Some people have a small amount of enzyme present. The serum PHE would be elevated in these people, but would not be as elevated as people with complete absence of the enzyme.

## People with PKU have too much PHE in their bodies, and that extra PHE can be a problem.

- This problem is most serious for babies and young children, because extra PHE can hurt their growing brains.

If babies and young children with PKU eat foods very high in protein (foods such as milk, chicken, fish, eggs, nuts, meat), their brains will be damaged by the extra PHE. They will become mentally retarded and may have seizures.

- Brain damage can be prevented with a special diet. This diet limits the amount of PHE the baby or child eats and keeps the level in the body low enough so that the brain is not injured. The special diet is the only way to prevent brain damage in babies and young children with PKU.

The special diet will provide protein, which is necessary for growth.

## Extra PHE in the body may also be a problem for older children with PKU.

- The elevated PHE levels may have effects on their mood and their ability to pay attention and learn in school.
- Doctors now know that using the special diet in older children and adults with PKU is necessary.
- It is now recommended that children with PKU stay on their diet for the rest of their lives.

Too much PHE in the body is a serious problem if a woman with PKU becomes pregnant.

- The extra PHE can harm the baby growing inside the mother.
- The chances for damage can be greatly reduced if the mother goes on a special diet **before** she becomes pregnant. For more information, please read/ask for the *Maternal Hyperphenylalaninemia* booklet.

## PKU AND GENES

### Genetic Disorders:

- Health problems caused by genes are called genetic disorders.
- They are different from health problems caused by other things like germs, accidents, or not getting enough good food.
- You cannot "catch" genes from other people like you can catch colds.
- **PKU is a genetic disorder.**

### Genes:

- Tiny particles made of DNA found in all cells of our bodies.
- Every person has about 100,000 different pairs of genes.
- Each gene pair has its own job to do in the complicated process of growth and development before and after birth.
- Some genes control the color, shape, or size of body parts.

Other genes give directions for how the body works. Sometimes genes give directions, which make the body look or work differently enough to cause problems.

Each of us is special and unique.



### Children inherit their genes from their parents:

- One of each kind of gene comes in the mother's egg; the matching gene of the pair comes in the father's sperm.
- Exact copies of every gene are put into every new cell of the growing baby's body.
- Parents have no control over which genes they give their children.
- Every time eggs and sperm are made, the genes going into them are mixed together by chance into new combinations. This means that even brothers and sisters (except identical twins) are genetically different from each other. **This is why some children in a family may have a genetic disorder, while their brothers and sisters may not.**

## Usually parents of children with PKU do not have PKU themselves:

- Parents of children with PKU are "carriers" of the PKU gene,
- Carriers only have one PKU gene in each cell. It is paired with the usual form of gene which does make the liver enzyme,
- Thus, carriers' bodies make enough enzyme to keep low levels of PHE. In fact, most carriers of PKU genes do not know they are carriers.
- Usually the way people learn they are PKU carriers is by having a baby with PKU.
- Parents who are carriers of a PKU gene may be the first in their generation because of spontaneous mutation of their genes.

## The only way people can get PKU:

- Is to inherit a pair of PKU genes, one from each parent.
- When women make eggs and men make sperm, one of every kind of gene, including the gene for the liver enzyme, which changes PHE to TYR, goes into each egg and sperm.
- When both parents are PKU carriers, half of their eggs or sperm get the usual gene which makes the liver enzyme, and the other half of the eggs and sperm get the PKU gene. When a sperm from a carrier fertilizes an egg from a carrier, there is a 1 in 4 chance in each pregnancy that both the egg and sperm have PKU genes.
- A baby growing from the fertilized egg will have two PKU genes and will be unable to make the liver enzyme, which changes PHE into TYR. The baby will have PKU.

# WHAT IS THE CHANCE THAT PKU-CARRIER PARENTS WILL HAVE A CHILD WITH PKU?

## Parents

Carrier  
Father



Carrier  
Mother



Father's Sperm  
without PKU gene  
**N** & with PKU gene  
**X**

Mother's Eggs  
without PKU gene  
**N** & with PKU gene  
**X**

Possible combinations  
of Eggs and Sperm

Outcome  
for Child



Chance of Occurance  
at each  
Pregnancy

no PKU genes  
**NN**

PKU carrier baby  
**XN**

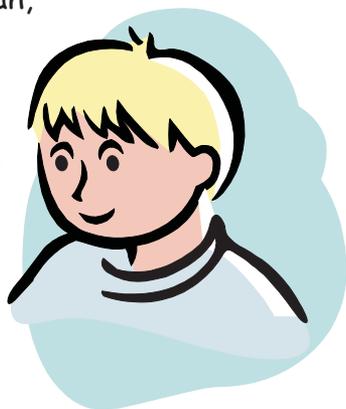
PKU carrier baby  
**NX**

PKU baby  
**XX**

# HOW MANY PEOPLE HAVE PKU?

- Not very many.
- In the United States, about one of every 60 Caucasians is a PKU carrier.
- One of every 3,600 Caucasian couples are PKU carriers.
- About one of every 10,000 - 14,000 Caucasian babies born in the United States has PKU.
- In Pennsylvania, 15 to 20 babies per year are born with one of the types of PKU.
- PKU is rare in African-American, Hispanic and Asian children.

Most people with PKU do not know anyone else outside their family with PKU. Most parents who learn that their baby has PKU are extremely surprised, because they have never heard of PKU before. PKU is rare enough that most people know very little or nothing about it, and it is hard to learn much about PKU except from a PKU Treatment Center.



Parents find out their baby has PKU when they receive the report from a blood test done on all babies a couple of days after birth. Then the family goes to a PKU Treatment Center. The parents and the center staff plan treatment to meet the special needs of the baby and the family.

# HOW PKU IS TREATED

Babies and children with PKU must be treated to protect their brains from being damaged by the extra PHE in their blood. There are three parts to the treatment - special diet, blood tests, and PKU Treatment Center visits.

## 1. The Special PKU diet

- Includes foods that are low in protein and therefore have very little PHE.
- These foods are mainly fruits, most vegetables, some cereal and grains, and low protein food products.
- Parents and children learn how much PHE is in certain foods and how to measure the food so that they know how much PHE the child with PKU is eating.

- Please discuss breastfeeding with your healthcare team.

Mothers with PKU babies may partially breastfeed. Mother's milk has low levels of phenylalanine. The amount of PKU medical formula the baby needs combined with the amount of mother's milk is determined by the baby's serum PHE levels. Your health care team will monitor the baby's PHE levels carefully.



- The diet also includes a special milk or metabolic formula. There are several brands of medical food formulas, for example: Maxamaid, Maxamum, Periflex, Phenex. The medical formula is a powder, which is mixed with water and is consumed like milk. It can also be mixed with flavoring and eaten as "paste," like a pudding. The medical formula contains most of the food substances our bodies need - vitamins, minerals, fats, sugars, and starches. It also contains a lot of the building blocks of protein (amino acids), especially TYR (which people with PKU cannot make). Most importantly, the formula has little or no PHE.
- The specific formula will need to be changed as the child grows and nutritional requirements change.

### Without the special medical formula:

- Children with PKU do not get enough of the building blocks (amino acids) they need to make body proteins.
- Protein-rich foods cannot be eaten to give them these building blocks because they contain too much PHE. Foods with a lot of protein (milk, nuts, meats, fish and eggs), should **never** be eaten by a person on the special PKU diet. The medical formula takes the place of these foods.

### Not all children with extra PHE in their blood have to follow the special diet.

Children with elevated serum PHE levels may need to follow the diet, because without it, the PHE in their blood would be too high and could lead to brain damage.

Children with lower levels of PHE, may not need the special diet - their brains will grow normally because their PHE levels are in a safe range. Children with mild hyperphe still need blood tests to make sure that the serum PHE levels do not rise.

## 2. Blood tests

Blood tests are used to tell how much PHE is in a person's body. The test shows a level of PHE in the blood. This level should not be too high because high levels can damage the growing brain.

The level should not be too low because people need PHE to make body proteins. Children may not grow well if they do not have enough PHE.

It is important for people on the special diet to have blood levels checked.

- Blood tests are done weekly on babies and toddlers with PKU because they grow so fast.
- Blood tests are done at least once a month on older children and teenagers on the special diet.
- Doctors, nurses and dietitians use the blood levels to help them determine how much PHE from food a child may consume.
- Parents and children can then measure the amount of foods by the amount of PHE they can have. Blood tests may be done by the community health nurse in your area. Parents can also learn how to do the blood tests by pricking the baby's heel or their child's finger for a blood sample.



### 3. Medication

- Some PKU patients may respond to an oral medication called Kuvan.
- Kuvan helps the enzyme lower plasma PHE levels.
- Speak to your treatment team specialist for further information regarding Kuvan.

### 4. PKU Treatment Center

Most children with PKU go to special PKU Treatment Centers.

During a treatment center visit, children and their families may see several people.

- The doctor examines the children and measures their growth.
- The doctor, nurse and dietitian talk about blood tests and diet.
- The social worker helps families with such things, as family relationships, school planning, and finding insurance coverage and other needed services in their communities.
- Families may also meet with the genetic counselor to learn how PKU came to be in the family.
- Older children and teenagers can have questions answered about what it means to have PKU.
- Older girls learn about the special problem of maternal hyperphe.\*

Most of all, PKU Treatment Center visits are a time to talk about questions and concerns about PKU with professionals who can help with answers.

\*In addition, some children and families have their own public health nurse in their own community. These nurses help parents learn about the diet, obtain blood tests, and answer questions about PKU in between visits to the treatment center. Also, anything about working with your child's primary care doctor regarding immunization and illness, can be discussed.

# SUMMARY

1. PKU is an inherited disorder in which genes, inherited from carrier parents, cause their child to have too much PHE in their bodies after eating protein-rich foods.
2. Some PHE is necessary for good health, but too much PHE can harm a young child's growing brain.
3. Harm to the child's brain can be prevented with a diet of special medical food formula and foods low in PHE.
4. PKU is treated in three parts - a special diet plus formula, regular blood testing for PHE levels, and regular visits to a PKU Treatment Center.
5. As children with PKU grow older, they learn about PKU and begin to take more responsibility for their own health care.



# **PENNSYLVANIA**

## **PKU TREATMENT CENTERS**

**St. Christopher's Hospital for Children**  
Erie Avenue at Front Street  
Philadelphia, PA 19134  
(215) 427-5485

**Milton S. Hershey Medical Center**  
Department of Pediatrics  
500 University Drive  
Hershey, PA 17033  
(717) 531-8006

**Children's Hospital of Pittsburgh**  
45th St & Penn Ave  
Pittsburg, PA 15201  
(412) 692-8631

**Children's Hospital of Philadelphia**  
Section of Metabolic Disease  
34th Street and Civic Center Boulevard  
Philadelphia, PA 19104  
(215) 590-3376



**pennsylvania**  
DEPARTMENT OF HEALTH

717-783-8143

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and Pennsylvania Department of Health

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To connect children with special needs and their families  
with recreation and leisure opportunities, call the Special  
Kids Network at 1-800-986-4550.