

Women with PKU

Second Edition



California Department of Health Services
Maternal PKU Program

To women with PKU:

If you are a young woman with PKU, this booklet is for you. It contains important information for you and any children that you may have in the future. Please keep this booklet for your records.

To family members and friends of women with PKU:

If you know a woman with PKU, please read this booklet and discuss it with her. Your support will help her follow the advice in this booklet and could make a big difference to her and any children she may have in the future.

Important Telephone Numbers

Name/Address

Telephone

Doctor

PKU Clinic

Nutritionist/Dietitian

Introduction

PKU (phenylketonuria) is an inherited disorder. Inherited means that each parent passed it on to his or her child just like hair, eye and skin color. This is how someone can get PKU. It is **not** contagious. You cannot catch PKU from someone else like you catch a cold.

Most babies born in the United States are tested for PKU through a newborn screening program. In California, about one baby in every 23,000 is born with some form of PKU. When you were a baby, your blood was tested. This is how your family learned you had a certain type of PKU.

Depending on the type of PKU you have, you may have been put on a special diet as treatment for your PKU. This special diet helps prevent mental retardation caused by PKU.

PKU cannot be cured, but it can be treated by a special diet. You cannot outgrow it. If you had PKU as a baby, then you still have it now. PKU can cause problems for people of all ages who are not on treatment. The most harmful problems occur when a pregnant woman with PKU is not on a special diet. Babies born to women who are not on a special diet often have serious birth defects, such as mental retardation and heart defects. However, these problems can be reduced or prevented. The next few pages talk about PKU and Maternal PKU and the special diet.

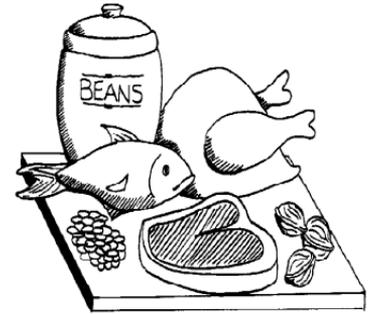


What is Phenylketonuria (PKU)?

PKU is an inherited metabolic disorder. It affects the way your body breaks down the protein in foods you eat into smaller parts called amino acids. Amino acids help to build and repair the body. One amino acid is **phenylalanine**, commonly called “**phe**” (pronounced *fee*).

People with PKU cannot break down phe into tyrosine. They are not able to make tyrosine. **Tyrosine** is another amino acid important for normal growth. The phe cannot be broken down to make tyrosine and builds up to high levels. This is harmful to the body. High phe levels can cause lifelong mental retardation if not treated with a special low phe diet at an early age.

Babies who have high phe levels in their blood and are diagnosed as having PKU are put on a low phe diet. This diet helps insure that their brain develops normally.



High phe levels may cause problems in children, teenagers and adults. These problems include skin rash, seizures, difficulty in paying attention at school, hyperactivity and cranky moods.

Pregnant women with high phe levels are more likely to have a miscarriage or a baby with a serious birth defect than those who have their phe levels under control.

The most harmful problems occur when a pregnant woman with PKU is not on a special diet. Babies born to women who are not on a special diet often have serious birth defects, such as mental retardation and heart defects. However, these problems can be reduced or prevented.

Types of PKU

1) Classical PKU - A complete block in the breakdown of phe to tyrosine. People with this type of PKU are treated with a low phe diet and special formula. Pregnant women with this type of PKU should always be treated with a special low phe diet before and during pregnancy.

2) Variant PKU - An incomplete block in the breakdown of phe to tyrosine. This is sometimes called “Variant Hyperphe.” A low phe diet may or may not be prescribed for variant PKU during childhood.

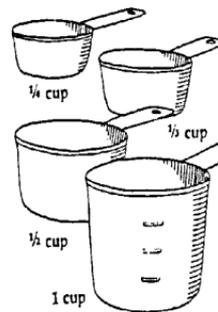
All women with any type of PKU should visit their PKU clinic and discuss the need to be on a special low phe diet **before** planning to become pregnant. The clinic staff will work with you to develop a plan to meet your special needs.

What is Maternal PKU?

Maternal PKU refers to women with PKU who are at the age when they could become pregnant, are pregnant, and/or already have children.

Women with PKU who are not on a special low phe diet before and during pregnancy have high levels of phe in their bodies. These high levels of phe can cause serious problems for the unborn baby.

Babies born to mothers with PKU usually **do not** have PKU themselves. However, many of these babies have mental retardation, heart defects and other problems. These problems can be found in babies born to mothers who have any type of PKU. These problems may not occur or may be reduced if women with PKU stay on the low phe diet.



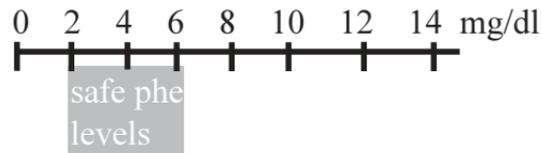
Severe damage to the unborn baby can happen as early as the beginning of the pregnancy. Women with any type of PKU should have controlled phe levels at least **2 months before** becoming pregnant. Controlled phe levels range from 2 to 6 mg/dl* (milligrams per deciliter). **This range is considered safe because the unborn baby has a phe level one and a half (1 1/2) times higher than the mother's phe level.** This means that if a pregnant woman has a phe level of 4 mg/dl then her unborn baby's phe level is 6 mg/dl. The current recommendation for women who are not pregnant is to maintain a phe level range from 2 to 10 mg/dl** but preferably within the 2 to 6 mg/dl* range. Please talk with your PKU clinic to find out what phe level is best for you.

If you are already pregnant and not on a low phe diet, contact your PKU clinic. Starting the diet early improves your chances for having a healthy baby. PKU clinics are listed on pages 12 and 13.

If you use micromoles:

* 2-6 mg/dl = 121 - 363 $\mu\text{m/l}$ (micromoles per liter).

** 2-10 mg/dl = 121 - 605 $\mu\text{m/l}$ (micromoles per liter).



What is the Low Phe Diet?

The low phe diet is the current treatment for PKU and Maternal PKU. The PKU diet provides the right amount of phe for normal growth while preventing a harmful buildup of phe in the body. If you follow the low phe diet, the level of phe in your body will go down. The low phe diet includes a special drink and low protein foods like fruits, juices, most vegetables, and fats. You can buy low phe breads and pasta or you can buy special products to make them at home. Contact your PKU clinic for help in starting the diet.

If you stopped eating a low phe diet, do not try to go back on the same diet that you had as a child. The diet needs to be adjusted to fit your body's changing needs. The diet will be planned just for you.

Most people with PKU are told to stay on a low phe diet for life. Staying on the low phe diet helps you control your phe level. It may not be easy to stay on a low phe diet, but it will help keep you healthy.



Why should I stay on a Low Phe Diet?

There are many benefits to being on the low phe diet. Many women notice when they are on the diet that they feel better, can think more clearly, and have fewer headaches. Some say they do better in school and work. Women who are not pregnant should maintain a phe level range from 2 to 10 mg/dl.

Staying on a low phe diet during your entire life is easier than stopping and then going back on the diet when you decide to have a baby. If you stop the diet, you may have a hard time getting used to the special drink again. It may be hard going without foods high in protein such as milk, eggs, cheese, beans, nuts, meat, fish, chicken, and some breads and grains.

If you are not on the diet and become pregnant, your unborn baby will be more likely to have serious problems caused by your high phe levels. Since most pregnancies are not planned, it makes good sense to stay on the diet whether or not you are planning to have a baby.

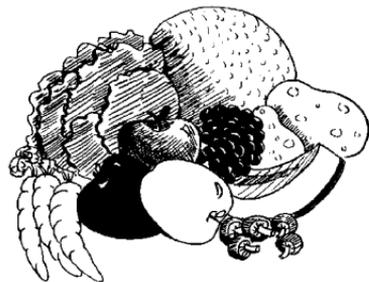


PKU diet for life

The success of your diet depends on you identifying and choosing low phe foods, measuring portions, and planning ahead. How you cope with the diet and self-discipline is also important. To prevent health problems, this diet should be followed for life.

Dietitians play an important role in helping you with the PKU diet. They monitor your phe levels and plan a diet that best meets your needs. You will learn to measure the phe in foods and how to count exchanges of phe on a daily basis. The amount of phe you can eat is determined by the phe level in your blood (measured by blood tests), your body's growth needs, and how well you can tolerate phe.

Parents of children with PKU and dietitians have developed low phe cookbooks and food lists to help you count your daily phe intake. Many of these cookbooks are available only through your metabolic clinic. Ask your dietitian how to get these items.



What if I am not on a Low Phe Diet?

If you have stopped the low phe diet or were never on it, and especially if you plan to become pregnant, contact a PKU clinic for an appointment. (Clinics are listed on pages 12 and 13.) Your doctor and the PKU clinic staff will work with you to plan your care. The dietitian at the clinic will help you plan the low phe diet that is best for you. You will learn how to choose the right foods and how much to eat. Women with PKU who were never on the diet should discuss with the PKU clinic staff the need to be on a low phe diet during pregnancy.

Once you start the low phe diet, it may take several weeks for you to get used to it, and for your body to reach a lower phe level. If you are sexually active during this time, discuss family planning with your doctor or clinic. Only when your doctor or clinic tells you that your phe levels are low enough can you safely begin a pregnancy.

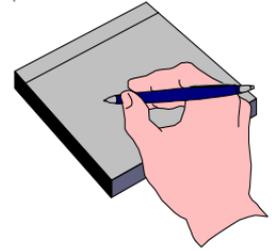
Should I have children?

The decision about when or whether to have children is up to you. You may not be ready to make a choice yet. There may be other things that you want or need to do before thinking of starting a family. You may decide to adopt children or not to have any children at all. Whatever you decide, planning is very important.

Pregnancies are often unplanned. Even with a planned pregnancy, it may take several weeks before you find out that you are pregnant. During those early weeks, the baby's heart, brain and nervous system are forming. Other important growth is also taking place. Getting your phe levels down and in control **at least 2 months before** you get pregnant will provide the best protection for your baby. Once you are pregnant, it is important that you remain on a low phe diet throughout your pregnancy to have the best chance of having a healthy baby.

Family planning services can help you with your plans to have, or not to have, a baby. There are many ways to obtain these services. You can discuss family planning with your doctor, partner or parents. Or look in your phone book, in the yellow pages, under "Family Planning Information Centers." You can also call your local health department or the toll-free number **1-800-942-1054** for information on family planning services in your area.

Steps to take . . .



- 1) Tell your primary care doctor and/or prenatal care provider that you have PKU.
- 2) Call and visit the PKU clinic to develop a low phe diet.
- 3) If you are on a low phe diet, stay on it.
- 4) Call your PKU clinic if you get sick since your diet may need to be adjusted.
- 5) Visit the PKU clinic if you are planning a pregnancy.
- 6) Give your PKU clinic and the California MPKU Program your new address each time you move so that you will continue to get important information.
- 7) Write down any questions you have for the PKU clinic staff on the next page.
- 8) Bring this booklet with you to the PKU clinic.

Your questions for the PKU clinic staff

Metabolic/PKU clinics in California

Northern California

Children's Hospital Medical Center
of Northern California
Oakland, California 94609
(510) 428-3351

U.C. Davis
PKU Metabolic Clinic
Davis, California 95616
(916) 752-7664

Valley Children's Children Hospital
Fresno, California 93726
(209) 243-6400

Stanford Medical Center
Stanford, California 94305
(415) 723-6858

U.C. San Francisco
San Francisco, California 94143
(415) 476-2757

Northern California Kaiser
Permanente Medical Group
Oakland, California 94611
(510) 596-6725

Southern California

Children's Hospital of Los Angeles
Los Angeles, California 90027
(213) 669-2152

Loma Linda University Medical Center
Loma Linda, California 92354
(909) 478-8119

San Diego-Imperial Counties
Developmental Services, Inc. Center
San Diego, California 92123-1648
(619) 576-2975 or 576-2851

U.C. Irvine Medical Center
Orange, California 92668
(714) 456-6878

UCLA Medical Center
Los Angeles, California 90095-1752
(310) 206-6581

Harbor/UCLA Medical Center
Torrance, California 90509
(310) 222-3751

Los Angeles County/USC Medical
Orange, California 92668
(714) 456-6878

Southern California Kaiser
Permanente Medical Group
Los Angeles, California 90027
(213) 667-6970

The California Maternal PKU Program

The Maternal PKU (MPKU) Program began in 1985. This program is in the Newborn Screening Section of the California Department of Health Services, Genetic Disease Branch. The following are the goals of the Program:



- * Provide current information on maternal PKU to young women with PKU.
- * Provide support to young women with PKU in maintaining a healthy diet and lifestyle.
- * Inform health care providers of the special needs of women with PKU before and during pregnancy.

The Program includes a newsletter, educational materials and a week-long camp/conference for young women with PKU. Also, a registry is maintained which contains information on women with PKU. This information is used to evaluate the California MPKU and Newborn Screening Programs.

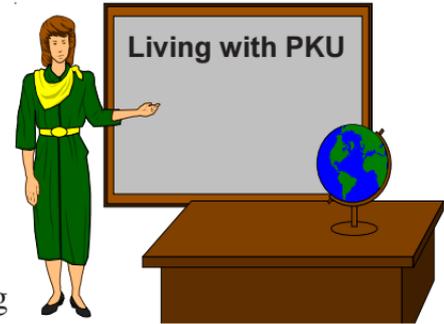
If you are a young woman with PKU, the Program wants to keep you informed about maternal PKU. You may call or write to us directly at the number and address listed on the back page of this booklet. If you know other young women with PKU, please tell them about this Program. Tell them to call their doctor or the Maternal PKU Program for more information.

California's Maternal PKU Camp/Conference

Since 1987 California's Department of Health Services has held an annual week-long educational camp/conference for young women with PKU. The main purpose of the camp is to bring together young women with PKU of childbearing age to learn the importance of being on a low phe diet. This is done through cooking classes, lectures and discussions. Also, these young women get to meet and interact with other young women with PKU.

The Maternal PKU Camp/Conference is held on a college campus during the summer. It is open to young women of childbearing age. Staff includes dietitians, nurses, doctors, health educators, genetic counselors and other health professionals. Workshops focus on nutrition, developing diet self-management skills, coping with PKU, dealing with peer groups, family support, and women's health and self-esteem. Campers learn about PKU and their diet, how to cook low phe foods, and often develop lifelong friendships and have lots of fun in the process. Contact the Maternal PKU Camp Coordinator for more information about the MPKU camp.

There are many people who would be happy to talk to you if you have questions about anything in this booklet. These people include your doctor, the PKU clinic staff and the MPKU Program staff. Feel free to call or write to any of us today. The MPKU Program address and telephone number are located on the back of this booklet.



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- * Parents of children with PKU
- * Young women with PKU
- * Newborn Screening Section of the Genetic Disease Branch

For more information on PKU and Maternal PKU or for additional copies, please contact the California Maternal PKU Program.

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Genetic Disease Branch-Newborn Screening Section
Maternal PKU Program
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