New Parents'

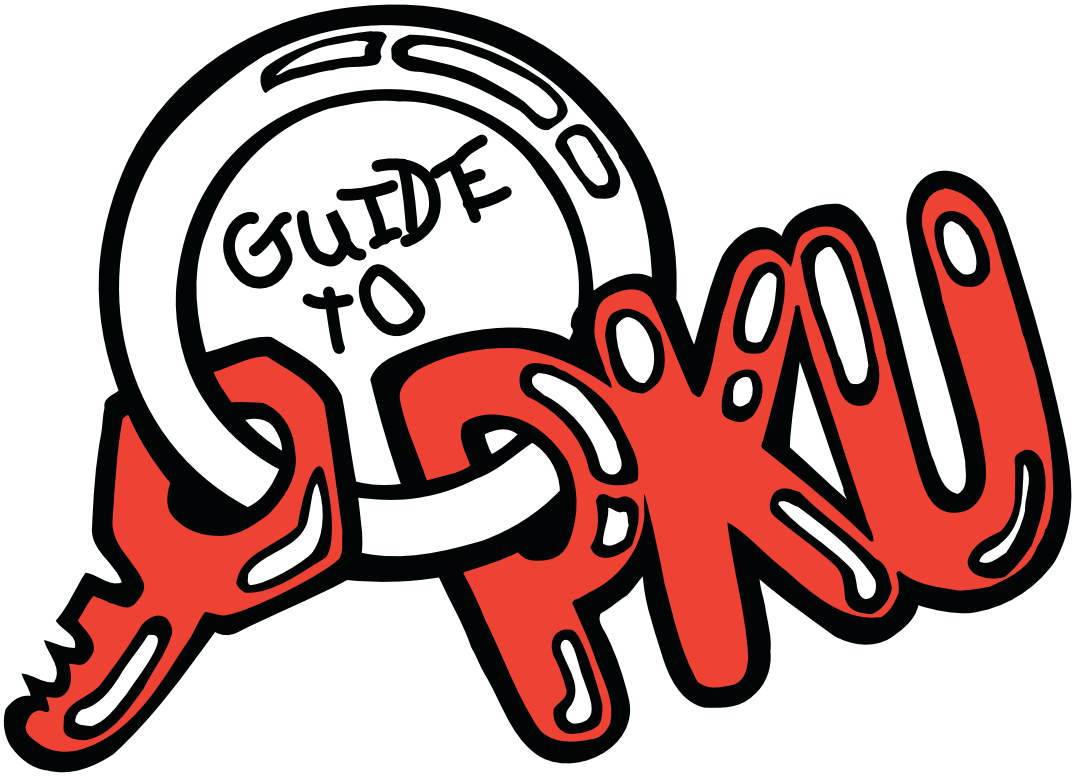


Table of Contents

Preface

The purpose of this booklet is to help to familiarize you, as new parents of a child with phenylketonuria (called PKU), with PKU and its treatment. It will also help to orient you to the PKU clinic and the people there who will help you to manage your child's PKU.

A short glossary at the end of this booklet will help you learn the vocabulary surrounding PKU. The underlined words can be found in the glossary.

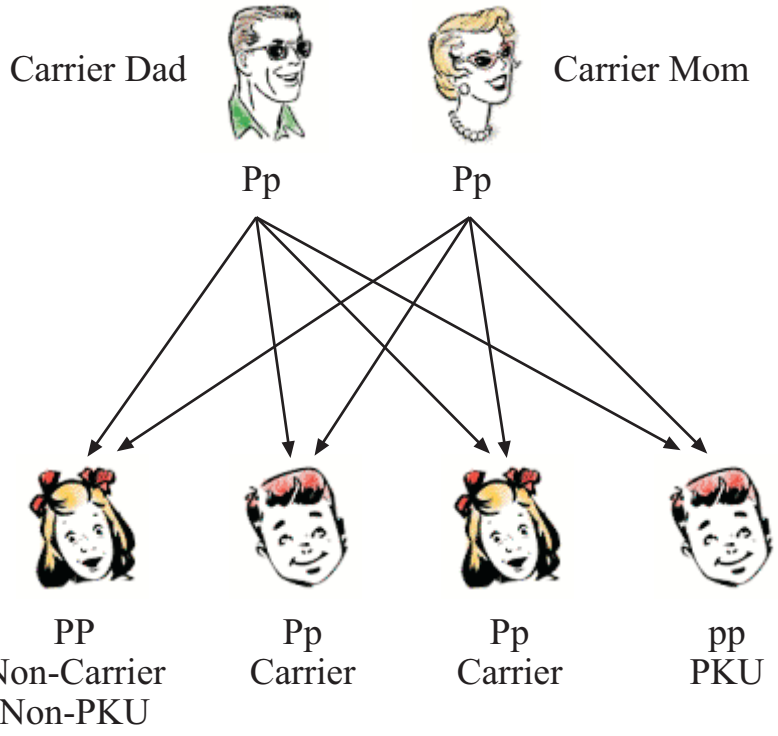
What is PKU?

PKU is short for **phenylketonuria** (pronounced fen'-il-kee'-to-nu'-ria). PKU is a **genetic** disorder in which the child is born without the ability to break down a certain amino acid in the body. Amino acids are the building blocks of protein. This **amino acid** is called **phenylalanine** (pronounced fen-il-al'-ah-nin) or phe (say 'fee'). Since this **amino acid** cannot be completely broken down, it builds up in the blood. A higher than normal level of phe in the blood is called **hyperphenylalaninemia** (hi'-per-fen-il-al'-ah'-nin-e'mia), or hyperphe, (hi'-per'-fee). When phe builds up in the blood it damages the brain and can cause mental retardation in the child with PKU.

Fortunately, if the child is **diagnosed** very early and the **blood phe level** is kept in good **control** throughout the child's life then mental retardation can be prevented. There are many excellent brochures and books which explain PKU in further detail, and you will be able to read through several of these in the future.

Where does PKU come from?

All of our genetic information is stored in the form of genes. Individuals with PKU have received two non-working copies of the gene for PKU. One copy is passed on from each parent. This means that the PKU enzyme (phenylalanine hydroxylase) cannot work properly and the phenylalanine cannot be broken down. Individuals who receive just one non-working copy of the PKU gene are called carriers. The normal copy of the PKU gene makes enough enzyme that they do not have symptoms of PKU and never need a low-phenylalanine diet.



When both parents are carriers, they each have a 50% chance of passing on their non-working gene. In every pregnancy there are four possible outcomes: non-carrier, non-PKU (25% chance), carrier by mom (50% chance), carrier by dad (50% chance) and affected with PKU (25% chance)

How is PKU treated?

PKU is treated by lifelong management of blood phe levels. This probably sounds overwhelming at first, and it does mean that there is a lot to learn, but it will probably not take any longer to learn how to care for your baby with PKU than it does to learn how to take care of any new baby for the first time. It is important to realize that a baby with PKU is a normal infant whose only unique needs are for a special diet. Both you and your child will start learning about the PKU diet as early as possible to insure normal growth and development. Since your child will be learning about phe management from the very beginning and *since this is the only diet your child will know, it will seem normal and natural*.

Treatment of PKU begins with a diagnosis of PKU, which must often be confirmed through a short hospital stay. In order to get to know your baby's individual needs, the doctor does frequent blood tests to measure the amount of phe in the baby's blood. This helps the doctor and the nutritionist determine the prescription for the special formula your baby will need. At first the blood tests may be as often as once a week but once the blood phe levels have stabilized then the blood test is done less frequently, usually once a month during the PKU clinic visit.

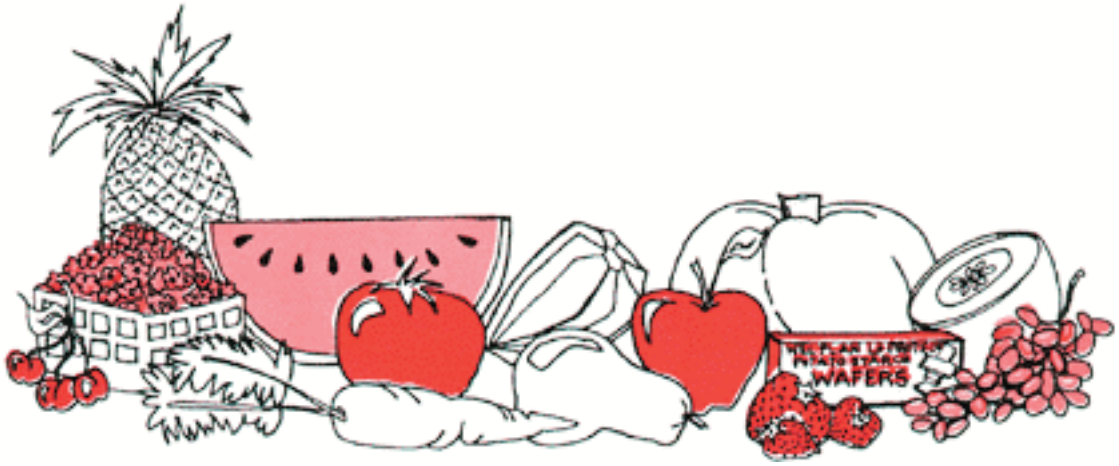
As your child grows older the blood test will help to monitor dietary compliance.

Controlling Blood Phe

At this point, you may be wondering how to keep the blood phe levels down to a safe level. Since phe is part of protein and must be eaten in order to get into the body, simply controlling the amount of phe your child eats is enough to keep the blood phe levels within a healthy range. We know which foods are highest in phe and which are lowest so it is possible to choose the low phe foods once one is familiar with them.

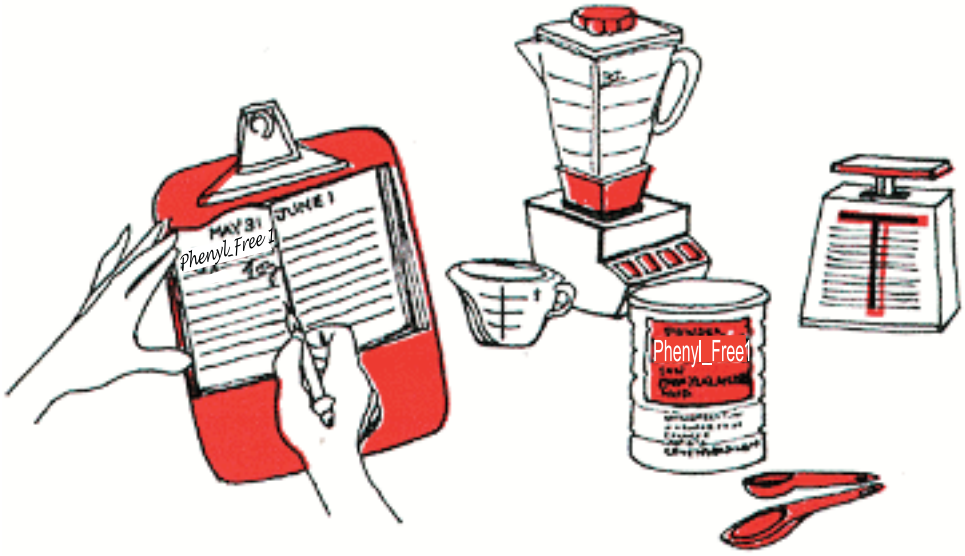
High protein foods are also high in phe, so foods like milk, meat, poultry, fish, eggs, cheese, nuts, beans and grains are omitted from the low phe diet. You will be learning the low phe food pattern along with your child as new foods are gradually added to the diet.

Since children, and especially babies, need protein in order to grow and develop normally, special foods have been developed to replace the high protein foods. The first of these products with which you will become familiar is a milk substitute called Phenyl-Free 1.



Phenyl-Free 1

Phenyl-Free 1 is a special formula designed specifically for babies with PKU. It contains all the needed protein and other nutrients, including vitamins and minerals, that a growing baby needs. It is a "super food" and will be the main source of nutrients for your baby.



If you are new parents, then learning to prepare the Phenyl-Free 1 will be just as easy as learning to prepare other baby formulas. Over the first few weeks the doctor and nutritionist are adjusting the formula in order to stabilize your baby's blood phe levels, so the prescription (recipe) will be changing frequently at first. The prescription and formula preparation will soon become easy to follow. It is very important to measure everything accurately to make sure that your baby is getting exactly the right amount of nutrients and phe.

You will be given a prescription or recipe for enough formula for 24 hours at a time. It will be written in the form of this example:

25 g Phenyl-Free 1 powder;

35 g Enfamil powder;

water to total 15 oz.

The prescription you receive will be specifically for your baby.

How to Prepare Phenyl-Free 1

To make Phenyl-Free 1:

A) Blender method: Using an immersion blender (e.g., Braun hand blender), measure 1 cup of water and place in a large Pyrex measuring bowl. Add the weighed Phenyl-Free 1 powder and blend. Add in the weighed Enfamil powder and add enough water to give the total formula volume required. Quickly blend again. Pour into clean baby bottles and refrigerate.

B) Shaking method: Weigh the Phenyl-Free 1 powder and place in a large pitcher with a tight cover. As about half of the water and shake. Add the weighed Enfamil powder and enough water to give the total volume required. Shake well until mixture is smooth. Pour into clean baby bottles and refrigerate.

To Store Phenyl-Free 1: Phenyl-Free 1 formula should be refrigerated after preparation the same as other baby formulas. The Phenyl-Free 1 formula should only be kept for 24 hours after being mixed. To store the opened can of Phenyl-Free 1 powder simply close it tightly to keep out humidity. Too much humidity can cause the powder to become caked and hard.

The PKU Clinic

The PKU Clinic is made up of a team of trained health care professionals specializing in the management of PKU. You may have already met some of the team members:

The clinic physician performs needed physical and neurological exams. He consults with families on medical matters and monitors current research in the field of PKU. The physician will determine that the diagnosis of PKU is correct.

The clinic nutritionist works closely with the child with PKU and the family to make sure that the child is receiving the necessary nutrients for normal growth and development while complying with the low phe food pattern. At PKU clinic the nutritionist talks with the parents about the child's diet, monthly blood phe level and possible dietary changes. The PKU clinic's educational lessons for specific age groups are supervised by the nutritionist. These lessons are designed to promote self-awareness and compliance with the PKU diet.

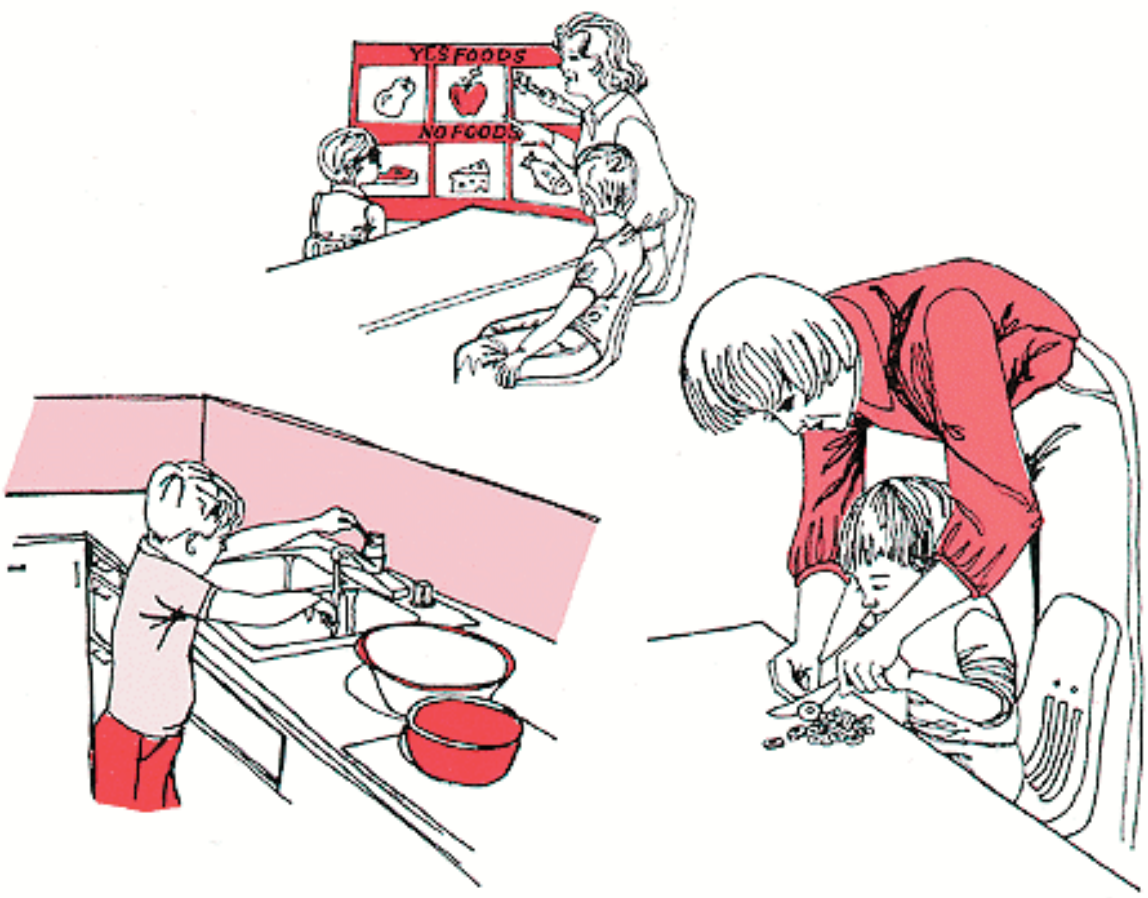
The clinic social worker conducts group discussion sessions for parents that take place on the PKU clinic day. Various issues about PKU, critical stages of diet management and parenting issues common to all families are discussed. Individual sessions with parents and children are arranged to provide extra support as needed.

The clinic nurse screens children with PKU for developmental progress from infancy through pre-school years, monitors routine childhood medical concerns and ensures that the children are medically followed in their community. The clinic nurse organizes parent education groups which focus on parent training for management of usual childhood behaviors and PKU.

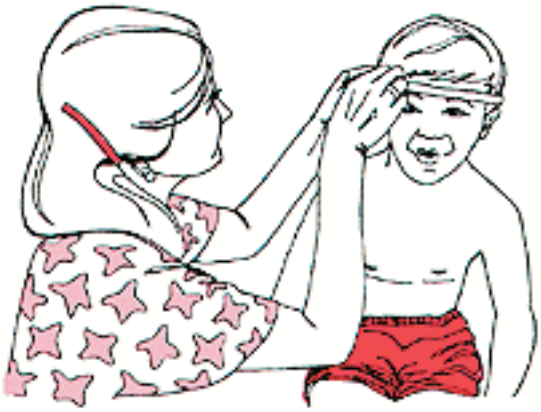
The clinic psychologist does routine psychometric testing on the children with PKU at specific ages to assess whether the children are developmentally "on schedule" and is available to evaluate those children who may have problems in cognitive or social development.

Parental Obligations

It is important that you take the diet for PKU seriously if your infant is to have normal intellectual development. If you are convinced the PKU diet is essential for your baby's good health, your child will learn to feel the same way. During infancy and childhood you are responsible for your child's dietary compliance.



Attending the regular PKU clinic is a very important way for the family of a child with PKU to receive support. Other parents have been through similar situations with their children with PKU and can offer new perspectives that may help you resolve difficult issues.



At PKU clinic the child's blood phe level is measured and height and weight are charted. The child is also able to participate in learning activities and to make new friends. The importance of regular clinic visits cannot be overstressed!

The normal intellectual development of your child is dependent upon dietary control of blood phe levels. You as parents are responsible of your child's diet and growth and intellectual development at this early stage of life.

The PKU clinic team is here to help you each step of the way as you strive to reach the goals of dietary compliance, good control of blood levels and normal intellectual and physical development for your child.

Above all, it is important to remember that you have a healthy child who has the opportunity to develop normally.

Summary

The child with PKU is normal in every respect except that the child's body can only handle a small amount of pine. Babies with PKU drink Phenyl-Free 1, a milk substitute, to keep the blood phe levels in good control and to provide the necessary nutrients for normal growth and development.

When the family and child with PKU attend PKU clinic the child will have a blood test to measure the amount of phe in the blood and will be weighed and measured so the growth pattern can be assessed. Following this, parents and children participate in both group and individual sessions with the nutritionist and other members of the PKU team.

At PKU clinic children are taught self-management skills so that they can make educated food choices for themselves. New foods are introduced to all of the children. The older children are introduced to simple recipes that are used to demonstrate basic food preparation skills.

The ideal of "parents as teachers" is firmly upheld and encouraged at the PKU clinic. You as parents will learn new ways to foster your child's attempts at self-management.

Glossary

Amino acid- a building block of protein. There are many amino acids but in PKU we are concerned mainly with the amino acid called phenylalanine.

Blood phe levels- (also called "serum phe levels" or "blood levels"). This refers to the amount of phenylalanine in the bloodstream. In order for normal intellectual development to occur it is important that the level of phe in the blood be maintained between 1 and 6 mg/dl.

Control- refers to how well the blood phe levels are controlled by the person with PKU. "Good control" indicates that the blood phe level is less than 6 mg/dl. "Poor control" indicates that the blood phe level is greater than 10 mg/dl.

Diagnosis- the identification of a disorder or disease on the basis of its signs and symptoms.

Genetics- the study of heredity.

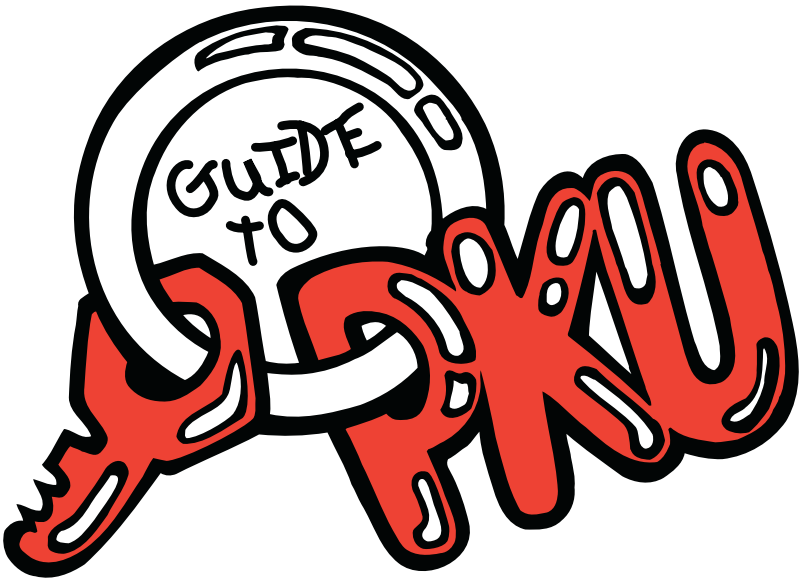
Hyperphenylalaninemia - the physical state of having too much phenylalanine in the blood.

Phenyl-Free 1©- the registered name of a special milk substitute made for children with PKU by Mead Johnson and Company, Evansville, Indiana.

Phenylalanine- is one of 20 amino acids that make up protein.

Phenylketonuria- a genetic disorder in which the body cannot completely break down the amino acid phenylalanine. It is characterized by higher than normal levels of phenylalanine in the blood which can cause damage to the brain and mental retardation.

Protein - long sequences of amino acids. Protein is an important nutrient since it provides the amino acids needed for growth and development.



Developed by Cristine M. Trahms, MS, RD, and Pamela Luce, MS, RD

Illustrations by Teresa Strom

Digital book creation by Greg Owen

Washington State Department of Health

Center on Human Development and Disability,
University of Washington, Seattle, WA 98195-7920

http://depts.washington.edu/pku

Revised, January, 2003