Parents' Guide to PKU
An Informational Guide to Dietary Management of Individuals with PKU

This booklet is intended by Mead Johnson to be provided to you by your healthcare professional as part of his or her care plan in the dietary management of PKU. Your healthcare professional is your primary source of information and advice. Always discuss with your healthcare professional any information you receive and any changes you wish to make.
PKU is an abbreviation for phenylketonuria, a metabolic disorder which affects about 1 in 10,000 children. It is an inherited disorder which can be treated very successfully. We at Mead Johnson hope this booklet will help you learn more about PKU.

We put Parents’ Guide to PKU together because we know how “lost” you may be feeling right now. What is PKU? How did our baby get it? Will our child grow and develop normally? How will we deal with this disorder? You are probably looking for answers to these and other questions. We hope that some of your questions about the recent diagnosis of your child will be answered here.

In Parents’ Guide to PKU, we have drawn on our experience providing metabolic formulas for children with PKU. Your experiences and your child will not be exactly like other families, but we hope that you and your family can benefit from what we have learned. We have also included the words of some parents who are truly living with PKU so that you might hear their experiences for yourself.

For simplicity, we have alternated “he” and “she” when referring to your child since both boys and girls will benefit from this information.

*Preface*

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What is PKU?

PKU is an abbreviation for phenylketonuria, a metabolic disorder in which the affected person lacks an enzyme that breaks down a component of protein in foods.

To understand PKU, we have to start with amino acids—the building blocks of proteins. Some amino acids are essential, which means the body cannot make them and they must be supplied by the diet. Phenylalanine is an essential amino acid which is present in most protein-containing foods; it is also converted by the body into another amino acid called tyrosine. Phenylalanine is often abbreviated to “Phe” (pronounced “Fee”). Both phenylalanine and tyrosine are needed for normal growth and development. The person with PKU doesn’t have the enzyme (phenylalanine hydroxylase) that the body uses to make this conversion from phenylalanine to tyrosine.

Because the body can’t get rid of the excess amounts of phenylalanine (Phe) in normal ways (ie, by converting it to tyrosine), phenylalanine builds up in the blood and other body tissues, and some is converted to substances called phenylketones, which are excreted in the urine. This process of the body trying to get rid of excess phenylalanine is where PKU gets its name: phenylketonuria or phenylketones in the urine. The high level of phenylalanine, phenylketones, and low tyrosine levels result in poor growth and mental development.

Will Our Child Grow and Develop Normally?

With proper dietary management and early identification, your child can grow and develop normally. Your child is likely to develop at the same rate as other children his age, to be equally intelligent, and to do as well in school.

Without dietary treatment, your child could develop more slowly and could become mentally retarded. Many studies have shown a relationship between the degree of dietary control, blood Phe levels, and the child’s IQ, so it is very important to follow the instructions you are given concerning your baby’s diet to maintain “safe” blood Phe levels.

PKU is a chronic disorder, which means your child will need to follow a special diet for all of childhood and into adulthood. The most healthy persons with PKU are those who control their
blood phenylalanine levels for life. It is especially important that girls control their blood phenylalanine levels because high levels of phenylalanine in a pregnant mother’s blood can cause serious problems for a developing fetus. Remaining on the diet for life is strongly recommended for all individuals with PKU.

How Did Our Child Get PKU?

You are probably wondering how your child got PKU. You may have other children who do not have PKU, and it’s possible that no one in your family has ever had PKU as far back as anyone can remember.

PKU is a genetic disorder, which means it is inherited from the parents. To understand how your child “got” PKU, it’s helpful to know something about chromosomes and genes. Every cell in the body contains 23 pairs of chromosomes (a total of 46 individual chromosomes) which have all the genetic information the body needs. One chromosome of every pair is inherited from the father, and one chromosome of every pair is inherited from the mother. These chromosomes are made up of genes which are lined up on the chromosomes like beads on a string. The genes are arranged in pairs just like the chromosomes are. Genes work to determine physical characteristics (such as eye color and hair color) and provide “instructions” for developing and maintaining the body, such as making phenylalanine hydroxylase.

In genetic terms, PKU is an autosomal recessive disorder. **Autosomal** means that it is not located on the “x” or “y” chromosome that determines gender. **Recessive** means that both parents must contribute a non-working gene for PKU in order for the child to have PKU. If only one parent passes on the gene for PKU, the child will not have any symptoms of PKU, but will be a “carrier” and still be able to pass that gene along to his or her children. Since the parents are usually “carriers” of the disorder, they will have no symptoms of PKU and may not even know they are carriers.

In fact, PKU may have been passed down *silently* through both families for many generations—that is, no one in either family may have known they carry the gene. It’s only when two carriers have children together that inherit a non-working copy of the gene from both parents that the disorder is expressed with clinical symptoms. Although untreated PKU can cause mental retardation, it does not indicate a family history of mental retardation. PKU can be very successfully treated, and children with PKU can grow and develop normally, both mentally and physically.

Figure 1 illustrates how PKU is inherited. For a child to have PKU, each parent must be a carrier.
of the disorder, which is shown as “Pp” in Figure 1. A carrier has one working gene (P) for producing the enzyme phenylalanine hydroxylase and one non-working gene (p) which does not produce the enzyme, but no symptoms of PKU.

In every pregnancy, the egg and the sperm combine to make a fetus. One of the mother’s genes for a trait (P or p in this case) from the egg and one of the father’s genes (P or p) from the sperm go to make the fetus. The inheritance of genes is a completely random process, and if the baby inherits two p genes, he will be born with PKU. This child (pp, in genetic terms) requires nutritional management to keep excess phenylalanine from being toxic to his body.

As shown in Figure 1, when two carriers for the non-working gene have children together, there is a one in four chance (25%) that they will have a child with PKU. Likewise, there is a 75% chance they will have a baby who does not have PKU; this child may be a carrier (Pp) like the parents or may have two working (PP) genes. These odds don’t mean that a family of four children will have one child with PKU, because the chances are the same with every pregnancy.

With every child, the odds in this example are one in four of having PKU.

Because PKU is hereditary, everyone in the families of both parents should know about the presence of PKU. This will help make certain that all babies born into the family are screened for PKU at birth. Families should talk with genetic counselors for the most up-to-date information on the genetic aspects of PKU.
How Do We Know Our Child Has PKU?

Children with PKU are first identified from elevated blood phenylalanine levels detected with newborn screening tests. These tests are convenient, inexpensive, and allow early identification of several genetic diseases. PKU screening is mandatory for all newborns in the United States and in many other countries. By screening in the first days of life, infants with PKU can receive treatment immediately. Prompt detection and treatment are extremely important because most problems associated with untreated PKU can be prevented if treatment is started early.

To perform the tests, a few drops of blood are taken from the baby’s heel and absorbed onto a piece of special blotter paper. The blood is usually taken in the newborn nursery.

Several laboratory tests are used to screen for PKU, the most common of which is called the Guthrie test. This test uses a certain strain of bacteria which needs phenylalanine to live. If there is excess phenylalanine in the baby’s blood, as in a baby with PKU, the bacteria will grow. The more phenylalanine there is in the blood, the more the bacteria will grow around the blotter paper. The amount of bacteria can then be compared to established standards to determine the phenylalanine level in the baby’s blood. Tandem mass spectrometry is also used in some states to screen for PKU.

If results of screening tests show elevated phenylalanine levels (positive), another blood sample is collected and a repeat test is run to confirm the results. Positive results on the screening test don’t diagnose the child with PKU, but they do indicate a need for further tests. More complete confirmatory tests are done by your baby’s doctor or a clinic that specializes in treating PKU.

High levels of phenylalanine in the blood can indicate yet another type of genetic disorder which is similar to PKU, but extremely rare. It is important to distinguish this group of patients from children with PKU because they must be treated with certain medications in addition to a special diet. The clinic doctors will perform several tests on your baby to be sure the diagnosis of PKU was correct.

In the 1930s, Fölling and others identified in severely mentally retarded patients a condition of high phenylpyruvic acid in their urines. This was later shown to be due to the patient’s inability to utilize phenylalanine. However, it was not until the 1950s that diets could be developed to treat the high levels of phenylalanine in infants. Before simplified screening for PKU became available early in the 1960s, infants with PKU often were not treated with special diets until their parents noticed the baby was not developing normally. By that time, brain damage had already occurred.

The development of a simple sensitive test to pick up phenylalanine in the blood and an effective diet resulted in the state-mandated screening program that picked up your baby’s elevated phenylalanine levels. Studies were carried out to determine if and when the special diet could be discontinued. The research showed that diet was needed for life to assure the infant reaches his or her maximum mental abilities. Today, babies with PKU can develop as normally as their unaffected siblings and go on to lead normal successful lives, as long as the special diet prescribed is followed. Research continues to be carried out looking for alternatives to diet. Until that goal is reached, special diets will be the only solution to help prevent mental retardation.
The PKU Diet

Your child’s special PKU diet is extremely important. Without dietary treatment, your child could become severely retarded. With careful dietary treatment, your child can be healthy and develop normally.

In the PKU diet your child will be on, foods that contain protein and are, therefore, high in phenylalanine (Phe) are typically excluded from the diet. These include cow’s milk, meats, eggs, cheeses, nuts, peanut butter, soy drinks, and tofu. Instead, your child’s diet will consist of a prescribed special medical food and low-protein foods.

This prescribed special medical food is essential since protein and its component amino acids are needed for normal growth and development. Nearly all proteins are high in phenylalanine. Because a person with PKU can’t break down phenylalanine to tyrosine, regular food protein will cause them to have too much phenylalanine and not enough tyrosine in their blood. Therefore, infants and children need a protein source free of phenylalanine but high in tyrosine. The special medical food has little or no phenylalanine, is high in tyrosine, and provides a good source of vitamins and minerals. Your metabolic clinic team will prescribe the appropriate special medical food for your child.

Because each child has a unique absolute requirement for phenylalanine, the metabolic clinic team will prescribe a diet tailor-made for your child. The prescribed special medical food, such as a dietary powder from the Phenyl-Free® family of special medical foods, is the mainstay of management of PKU. The metabolic clinic team will work with you to make sure your child gets the appropriate amounts of special medical foods, as well as other foods, as your child grows and develops. It is vitally important for your child’s growth and mental development to follow the metabolic clinic’s direction on feeding your child.

Your child’s needs change during growing up. The metabolic clinic team will help you design a diet to provide all the calories, phenylalanine, protein, vitamins, and minerals your child needs to grow. The special medical food will not supply all your child’s needs. As your baby grows and begins to eat solid foods, foods such as fruits, vegetables, and low-protein pastas will be added to the diet. The variety of foods and dishes your child can enjoy is quite large. Recipes have been developed already by other parents of children with PKU. The metabolic clinic team will help you find these recipes and ingredients.

The diet for PKU is quite simple to prepare once you understand what your child can and cannot eat. The metabolic clinic team will teach you how to plan your child’s daily meals. They will ask you to keep careful records of how much your child is eating in order to balance this intake with the amount of phenylalanine and protein needed for growth and mental development. For your child to develop normally, the diet must be maintained consistently and accurately. The metabolic clinic team will work closely with you to help you in this goal.

Regular Clinic Visits

Much of your child’s treatment for PKU may occur at a clinic that specializes in this disorder, and you will be asked to bring your child with PKU to the clinic regularly. Your baby’s pediatrician will get reports of the clinic’s diet instructions and your child’s blood results. In general, PKU clinics don’t handle illnesses unrelated to PKU or give routine baby shots, so it’s important for you to have a primary care provider or
pediatrician for your baby’s other medical care. The PKU clinic team can help you find a local pediatrician if necessary.

The PKU clinic team will regularly need information so changes in the diet can be made to match what your child needs to grow and develop normally. This information includes records of your child's food and special medical food intakes, height, and weight. The clinic will also want to measure blood levels of phenylalanine regularly to make sure that your child has every chance of normal development. These visits will give you an opportunity to discuss any questions you may have with the metabolic clinic’s team, and the team to share with you the latest information on PKU.

Just as in the treatment of any disease, doctors and nutritionists vary in their procedures for managing phenylketonuria. A particular clinic may differ from the following description, but there will probably be many similarities.

First Steps in Treatment

Following the initial positive result for PKU, your baby will be started on a diet very low in phenylalanine to get his blood phenylalanine level down quickly. This may take several days. If you are breastfeeding, the metabolic clinic doctor will ask you not to breastfeed your baby during this time. However, you are encouraged to continue breastfeeding your child after his blood phenylalanine levels are lowered, if you wish. Your baby's metabolic clinic team will help you plan a diet that includes breast milk.

Next, your baby will be put on a prescribed diet, and blood levels of phenylalanine will be checked frequently to be sure they aren’t too high or too low. Growth and development will be carefully followed as well. Frequent visits to the clinic will be required to see how your baby is progressing.

Many PKU clinics have a team approach, and you may have appointments with several members of your baby’s treatment team on the same appointment day. The nutritionist will probably be your primary contact—he or she will monitor the therapy, adjust the diet prescription as necessary, and provide advice when problems arise. A physician will examine your baby and help with problem-solving. A nurse will weigh and measure your child to be sure the diet is meeting growth needs. As your child gets older, a psychologist may evaluate him periodically to make sure his psychological development is normal. A social worker or counselor may meet with your family to help you adjust to living with PKU.

The services the clinic provides are essential to you to maximize your child’s development. Be assured the clinic team members are very interested in the health and development of your child. You will want to work closely with them in order to assure your child’s maximum progress. This will include keeping accurate records of your child’s food and special medical food intake, cooperating with the clinic in getting the required number of blood tests, and keeping clinic appointments. Providing the requested blood test, keeping clinic appointments, and following the clinic’s diet for your child are essential. The healthy development of your child depends on these steps.
How Will This Affect Our Family?

PKU is not without its effects on the family. Your child with PKU will need regular PKU clinic visits, especially as an infant and small child, and you will have to spend time preparing the foods and special medical food for your child. However, most families with children with PKU find that this quickly becomes routine. Your time and care will be rewarded by a child who can grow and develop normally.

**Parents:** Responsibility for the diet for PKU should be shared if at all possible. It may be tempting for one person, usually the mother, to handle the diet completely, but in the long run it will be better if both parents learn about PKU and how to prepare the diet your child requires. That way, when Mom is sick, or when “the boys” go camping, there is less worry about managing the diet properly.

**Brothers and sisters:** The controlled PKU diet should involve the entire family, and PKU should be discussed openly and easily. All children in the family should understand PKU so that they can provide support and encouragement to the individual with PKU.

It is important for the self-esteem of your child with PKU that siblings not be allowed to tease him or imply that something is “wrong” with him. Older children can be helpful in preparing the diet, and involving them in a family “project” can help the older children understand PKU better.

**Grandparents:** Grandparents also need to understand the diet and what the consequences are to the child if the diet is not carefully followed. Sometimes, it may seem especially hard for them to deprive their grandchildren of treats and other foods. A good approach is to share the importance of the diet with them and actively involve them in your child’s treatment. One grandmother was overjoyed when she was given a PKU cookbook for her birthday because she could then fix special treats without worrying about hurting her granddaughter.

**Other relatives:** It is important that everyone in your extended family understands that your child with PKU has a special diet and special medical food. You will probably find that most relatives will accept the situation and be very supportive. However, there may be some relatives who are confused about the seriousness of PKU and don’t understand the purpose of the diet. They may be inclined to let your child eat foods that you don’t allow. Careful and repeated explanations will help them understand the critical importance of your child’s diet to his growth and development. Usually, as relatives know more about PKU, they better understand the need for strict dietary management and find PKU less frightening.

**Neighbors:** Other families in the neighborhood, and parents of your child’s friends, should understand that your child cannot have the snacks they may be inclined to offer. In most cases, these families, as well as friends and babysitters, will support your child’s diet once they are told how important it is to your child.

Remember, the more people know about PKU the more they will understand and help.
What Can We Expect As Our Child Grows?

Infancy: Birth to 12 Months of Age

Babies change very rapidly during the first year of life. As parents, you will want to pay close attention to your baby so you don’t miss any new developments. Watch your baby—not because of the PKU—but because it’s fun to watch her develop.

By 3 months of age, your baby will be more responsive to people and will smile and coo in response to your smiling and talking. Increased muscle strength and coordination allow easy movement of legs and arms. When lying on his stomach, your baby will lift his head and shoulders to watch what is going on in the room. By this time, you will probably feel comfortable mixing the special medical food and keeping track of your baby’s formula or breast milk intake.

At 4 to 6 months, you will be introducing pureed foods. Your clinic’s nutritionist will probably suggest starting with purees of vegetables and fruits. The addition of solid foods at this stage may make the diet more difficult for you until you’re used to determining the amount of phenylalanine in food.

Most 6-month-olds have doubled their birth weight. Better control of back and neck muscles means your baby will be able to sit with only minimal support. Your baby will enjoy playing with his hands and simple toys. Most things that your baby can hold will go straight into his mouth, including foods that are within reach. You will want to have low-protein foods available so your baby can experiment with self-feeding. Low-protein foods with texture will help support chewing and oral motor development and to respond to hunger without affecting blood Phe levels.

By 12 months, your baby will have tripled his birth weight and will be able to get around quite well by crawling or walking while holding onto furniture. He will play more with toys and be able to pick up the tiniest things left within his reach. By this age, many babies will also be saying a few simple words.

As your baby grows, her growth rate will decrease. Because she is growing slower, she will naturally eat less. Work with your metabolic clinic team to balance your child’s diet with her appetite and her social and physical needs. You will want to establish a pattern of meals so she will begin to understand hunger and satiety. Feeding may become more difficult. When this
occurs, feed the special metabolic product first followed by the low-protein foods your baby likes. Your baby is also developing social skills. You will need to balance your approval and disapproval of your baby's behavior. Food and feeding should not become a place where excessive playing, pleading, or anger occurs. You could establish eating as a good area for your baby to assert her emerging sense of will.

By the time your child reaches 12 months of age, you will want to keep forbidden foods well out of reach. He will be much more active and able to explore. You will want to make sure that no dangerous household materials are accessible to be picked up and placed in his mouth.

**Toddlers: Ages 1 to 3 Years**

By 18 months of age, your child will probably be able to walk alone, drink from a cup, and try to feed himself. He will try to "talk" and will understand the word "no." His decreased appetite, desire to feed himself, and high level of activity will require attentive parenting. It is important to avoid these tensions as much as possible by positive developmental approaches.

Establishing regular meals and snack times, allowing extra time for self-feeding, and tolerating the resulting mess can help your toddler's motor and social skills. Feed the special metabolic product first to assure adequate intake of protein. Try new low-protein foods and recipes to make mealtimes more interesting for your baby. Your toddler will enjoy finger foods. By offering finger foods lowest in phenylalanine for feeding herself, it will simplify the task of calculating the amount of phenylalanine consumed and enable the child to have "more."

Two and 3 year olds are quite active. They progress from walking up and down stairs with help to unlocking the front door and going for a "walk" on their own. The 2 year old can put two words together and can make his wants known. The 3 year old uses sentences of several words and can point to his eyes and ears. The older toddler can go from happy to sad and back again at a moment's notice. This doesn't mean your child's phenylalanine levels are high, but that he is acting his age. The desire for independence and the tantrums that go along with this age are also typical and are not the result of PKU. If you do have any concerns, however, please discuss them with your clinic specialists.

By 2 to 3 years, your child may
know there are foods he can and cannot have, and you should start teaching him about his diet. But realize that a 3 year old does not understand why some foods are not good for him (especially when he sees other people eating them), and he cannot be expected on his own to refrain from eating them. Many families establish an “ask before you eat” rule. Develop an approach to understanding “yes” or “no” but not “maybe.”

Preschoolers: Ages 3 to 5 Years

Your 3 or 4 year old is obviously no longer a baby. He may ride a tricycle and wash his hands unassisted. Tantrums and stubborn behavior can continue through 3 and 4 years of age because these children often want to do more than they are capable of doing and become frustrated. This frustration (not their PKU) may lead to whining and crying.

Throughout the preschool years, your child will gradually become better able to understand some things about his diet. Try to answer as many of his questions as you can, even if it seems like you are answering the same questions over and over. It is very important that you are consistent. Generally, your child will be happiest with a schedule. Promote formula first and consistency in foods and portion size. Remember, that as your child grows, he will have more and more opportunities to make inappropriate food choices. Good eating patterns should be firmly established before opportunities to make inappropriate food choices present themselves.

At age 3, your child can begin to learn about PKU as well as his special diet, and he should understand the idea of “yes” foods and “no” foods. Reading books together for children with PKU can help you begin to teach your child about why he must follow a special diet. (See bibliography for resources.)

As your child is able, you may want to allow him to choose some of his foods and help with their preparation. Five and 6 year olds will have a general idea of what is allowed on their diet but probably won’t be able to decide if foods that are new to them are allowed. By 5 or 6 years, your child will be more cooperative about his diet, particularly if he has learned that you won’t bend the rules.

Grade School: Ages 6 to 12 Years

By grade school, your child will be mastering skills that she began developing at earlier ages. She will need less help taking care of herself and gradually will become independent in eating, dressing, and bathing. Your child may like
to ride a bicycle and visit with friends at their homes.

The thought processes of the grade-school child are constantly developing, and she will understand time periods and how to see things from another’s perspective. She will learn to deal with more than one detail at a time and to relate one idea to another. At this age, your child will reason and use simple logic and should be encouraged to apply new math skills to her diet as she learns them. Children in early elementary school should begin to keep track of their phenylalanine intake, as well as help with making their formula. It is reasonable and necessary to involve your child more and more in his diet as he reaches school age. By later elementary school, your child should understand what levels of blood phenylalanine are acceptable and should be tracking his own levels. As your child understands more about PKU and is more involved in his diet, you can provide opportunities for him to make decisions for himself, including calculations of Phe in foods. Parents should work with their children with PKU to choose foods and plan menus, record their diets on the diet records, and do their own finger sticks for blood samples. The more involved your child is in his treatment, the more likely he will have the knowledge and skills to make more appropriate choices independently. Children this age prepare formula and take responsibility for drinking it.

**Adolescents: Ages 12 to 18 Years**

Your adolescent may want to be independent, but he still needs parental guidance and direction. At this stage, your child should be developing responsibility for his own diet and food choices. This is good training, not only for the child with PKU, but also for all children.

Most adolescents are fully capable of understanding both PKU and the diet. Your teenager will be able to read and write with ease and to perform math calculations as required to manage his diet. He should be keeping his own diet records and being responsible for clinic appointments. One aspect of adolescence that will make dietary compliance more difficult, however, is the need for peer approval. Many adolescents don’t want to be seen as any different from their friends, and having PKU can make them feel different. These concerns may make it difficult for them to make appropriate food choices. Continue to actively parent your adolescent. Provide positive support and help him develop strategies for dealing with his peers—your child has no reason to feel different if he has the skill, knowledge, self-esteem, and parental support you have provided.
In the past, children with PKU were usually permitted to discontinue treatment (and have elevated blood phenylalanine levels) before adolescence. However, several studies have shown that children who discontinue treatment or have high Phe levels have more difficulties with thinking processes and math skills. Other studies have found that elevated blood phenylalanine levels in older patients were associated with behavior and learning problems which were reversed when the patients returned to more restricted diets. For these reasons, most clinics are now recommending that persons with PKU remain on the diet for life. This “diet for life,” which means good Phe levels for life, is a strong recommendation—discontinuing treatment is not advised at any age.

As they begin their menstrual cycles, girls with PKU should understand the effect PKU can have on developing babies. High blood levels of phenylalanine are very damaging to a developing fetus. Researchers have found that, as the mother’s blood phenylalanine levels increase, her chance of having a baby with an abnormally small head and mental retardation also increased. Incidence of low birth weight and congenital heart disease appears to increase markedly when serum phenylalanine levels are consistently above the “good level.” In women who have not stayed on the diet, it is necessary to reinstate dietary therapy before conception. The prognosis for having a normal child is better, but it is critical that diet therapy begins before conception. The fetus is very sensitive to high levels of phenylalanine. The early months of pregnancy are believed to be the most crucial for normal development.

For these reasons, it’s very important that women with PKU follow their phenylalanine-restricted diet before conceiving and throughout pregnancy. Because it’s so difficult to go back to the diet after being off it, it is especially important that women control their blood phenylalanine for a lifetime.
Did something happen during my pregnancy that could have caused my baby to be born with PKU?

No. PKU is inherited from both parents. Only when the uniting sperm and egg both contain the gene for PKU will the baby develop this condition.

How can you tell who is a carrier of PKU?

When a child is born with PKU, we know that both parents are carriers for the altered PKU gene. Consult with your genetic counselor about determining who else in your families may be carriers.

Siblings of a child with PKU have a 50% chance of being carriers for PKU. Therefore, they may want to have carrier testing done on themselves before they begin a family. If they know they are carriers of PKU but are unsure of the PKU status of their spouses, they will want to be certain that early screening is done on any children they may have.

What will happen to my child with PKU when she marries and has children?

If the father is a carrier for the altered PKU gene, a child they have would have a 50% chance of having PKU. If the father is not a carrier, none of their children would have PKU, but all children would be carriers.

What is “Maternal PKU?”

This is the term given to the special problems that can occur when a woman with PKU has children. Women with PKU need to understand that if they become pregnant when their blood phenylalanine levels are high, there is a greater chance the baby may be born with birth defects or developmental delays. Being on a very low phenylalanine diet prior to conception and throughout pregnancy reduces the risk of these problems.

If we have a child with PKU in the family, how will this affect the lives of our other children?

Having a child with special health needs causes some changes in attitudes and family routine. For example, eating habits may have to be adjusted, and the child with PKU may seem to get more attention than the children without PKU. Parents have to be
careful not to center their attention on the child with PKU, particularly at mealtimes. Older brothers and sisters may try to “mother” the child with PKU, and siblings may even taunt the child with foods he cannot eat. Parents will have to make it very clear that such behavior is not acceptable.

What is the PKU diet?

It is a diet low in phenylalanine, which is the amino acid that the child’s body cannot use properly. The specific diet for your child will be specially designed by your metabolic physician and nutritionist. The diet is designed to meet your child’s individual needs for growth and development. The diet includes a protein substitute, such as Phenyl-Free® 1 or Phenyl-Free 2 dietary powder, which provides the missing protein but has little or no phenylalanine. In addition to these products, patients are given a prescribed specific amount of phenylalanine from breast milk, regular infant formula, or solid foods to provide enough phenylalanine for growth. Measured amounts of fruits, vegetables, cereals, low-protein breads, pasta, and special desserts are added to the diet as the child gets older.

Is the diet expensive?

The formula costs more than regular infant formula because it is specially designed for medical purposes. However, because of the importance of formula for your child’s mental and physical development, most health insurance plans pay for it. In some states, the special medical formula is provided by the State Department of Health. In other states, these products are provided on a sliding fee scale, based on ability to pay. The social worker, nutritionist, or nurse at the PKU clinic can usually help identify a payment method.

Can I breastfeed my baby with PKU?

Yes. Breast milk is lower in phenylalanine than either infant formulas or cow’s milk, and it can provide the supplemental amounts of phenylalanine required for your baby’s growth when used with a special phenylalanine-free formula such as Phenyl-Free 1. Many newborn babies with PKU can be fed with breast milk and phenylalanine-free formulas and still keep their blood levels in an acceptable range. All babies with PKU must have their diet intake and blood phenylalanine levels monitored closely. You’ll want to discuss your decision to breastfeed your baby with PKU with your baby’s doctor and dietitian.

Why are diet diaries kept?

Diet diaries are records of food intake which serve as guides for the clinic team when adjusting your child’s diet. These diaries are a necessary management tool. It is the only way for the nutritionist to know exactly what your child has been eating or drinking so that appropriate changes can be made to assure the proper levels of blood phenylalanine.

How long will my child with PKU be on the diet?

Medical experts state that maintaining the restricted diet throughout life is preferable. Data from the National Collaborative Study of Children Treated for PKU show that intellectual ability and academic achievement in school are negatively affected when blood levels are persistently high before the age of six years. Other studies have shown that children who maintained acceptable levels of blood phenylalanine through age 10 had higher IQ scores than children who didn’t have low Phe levels. A “diet for life” approach is currently recommended.
What happens if my child with PKU refuses to eat part or all of his prescribed diet?

If this goes on for only a few days, it is not a problem. However, if the child gets into a pattern of eating only one kind of food, the prescribed amounts of food and formula may not be ingested. Don’t worry if this goes on for a day or two. If you develop a pattern of special metabolic products along with regular meals, your child will probably not refuse to eat for a longer period than one or two days. If she does, it’s a good idea to check with the metabolic clinic team to rule out a health problem requiring treatment. Call your PKU clinic for help in adjusting the diet during these times, rather than trying to adjust the diet yourself. Staff at the clinic will suggest other ways to get your child to eat and to assure adequate intake in cases of fever, vomiting, and/or diarrhea, or other childhood diseases which result in loss of appetite.

Will my child get sick if he does eat some high-protein food?

No. There may be an elevation in blood phenylalanine levels that will gradually decrease if further Phe intake is restricted. However, frequently high levels of blood phenylalanine can affect your child’s development, even though he may not show obvious physical signs of toxicity.

What are “good” blood phenylalanine levels?

Most clinics generally regard the range of 2-6 mg/dL as “good” blood phenylalanine levels. Please consult your clinic about what is a good level for your child.

What causes blood phenylalanine levels to go up?

In most cases, elevated phenylalanine levels may indicate that your child is eating more phenylalanine-containing foods than he should eat to meet his growth needs. If your child is not getting enough of his special metabolic product, his body will break down muscle and other protein stores, thereby releasing phenylalanine into the blood. Illness can also cause this breakdown and make blood phenylalanine levels rise. If your child is growing rapidly, he may need extra amounts of food or special metabolic product, which can cause a change in blood phenylalanine.

What causes blood phenylalanine levels to go down?

When a child’s diet is changed from regular food to a phenylalanine-restricted diet, levels of blood phenylalanine will go down.

Generally, when a child’s prescribed diet is appropriate for her age and rate of growth, and the diet is closely adhered to, phenylalanine levels should be within the desired range. A level below the range suggests your child is not eating all her “Phe foods.”

Will my child become retarded?

With early dietary treatment, careful dietary management, and resulting good blood Phe levels, the chances are very good that your child will grow and develop normally.

Will my child with PKU look or act differently than other children?

If he has received early treatment and adhered to the diet prescribed by your metabolic clinic staff, your child with PKU should not look or act differently than other children.

Will my child be sickly?

Having PKU will not cause your child to have poor health.

Will my child be able to go to regular schools?

If your child is treated early and carefully, his PKU will not prevent him from attending regular schools.
Tips For Parents

The following tips are compiled from the experiences of many families living with PKU. As you become more comfortable with the PKU diet, you likely will discover other ways to effectively manage the disorder. You should always discuss ideas with the clinic first before making any changes.

- Remember that you aren’t managing the diet of your child alone. The whole family, as well as neighbors and friends, should be instructed about diet restrictions.

- Estimate how long your supply of formula will last, then order early so you don’t have to worry about running out. Remember that you may not be able to go out to the store and buy an extra can of Phenyl-Free® 1, Phenyl-Free 2, or any other PKU formula if you run out.

- Don’t change too many things at one time during the transition from pureed foods to self-feeding. Work in consultation with your clinic nutritionist so this transition can be completed easily and in a stepwise manner.

- Keep in mind that during illness or fever, blood phenylalanine levels may rise. Consult your clinic for guidance on how to minimize this effect of illness.

- Include your child’s needs in the family meal plan. Try to make meals containing foods that your child can eat.

- Keep reconstituted products in a well-marked container that is different from any other container in the refrigerator so that your child can easily identify “her” milk substitute.

- Involve your child with PKU in the management of his own diet when possible. This could include helping to mix his formula, making his sandwich for lunch, or making treats from a low-protein cookie or cake recipe.

- Be consistent in your approach to the diet. A matter-of-fact but firm attitude will help your child adjust more easily to diet restrictions. Keep in mind that all children test their parents, and your child with PKU probably will be no different. When diet restrictions are followed regularly, forbidden foods may lose some of their appeal.

- Avoid the habit of snacking continuously. If your child needs to eat between meals, make that snack an event of its own which you or a caregiver supervises. You may want to have a “tea party” when your child wakes up from his afternoon nap. He will be less inclined to help himself to snack foods when he knows there will be a defined snack time.

- Don’t let your child sample unallowed foods just to taste them. Your child is better off never having tasted foods that, once tried, may become even more appealing.

- Never let your child know you find the smell or taste of the formula objectionable.

Hints While Traveling

- Always take more Phenyl-Free 1, Phenyl-Free 2, or any other PKU formula than you will need when you leave home, since it may be very difficult for you to get it in another state or country.

- Put premeasured amounts of food into small containers to simplify calculating your child’s intake.
• Measure the Phenyl-Free® 1, Phenyl-Free 2, Phenyl-Free 2HP, or any other PKU formula into individual baggies or other small plastic bags. You may find that a small shaker works well for mixing the products.

• Once your child is eating foods from the table, you can order vegetables and fruit for him at restaurants so he feels like eating out is a treat.

• Always carry low-phenylalanine snacks, such as fruit, low-protein cookies, and candy. If your family has ice cream, get a non-milk, iced popsicle for your child with PKU.

**Helping Your School-Aged Child With The Diet**

• Teach your child how to deal with questions about his diet from classmates. Think up good answers together before he starts school.

• Be sure your child’s teachers and lunchroom supervisor know of the diet for PKU and its importance to his health. Most school personnel will be very happy to help. It may be useful for you to give the teacher a guide to what kinds of snack foods your child can have. Children often bring cupcakes or cookies to school to celebrate birthdays or other holidays, and if your child can also have a special treat then he won’t feel so different from his friends.

• With some practice, you can help your child pack a lunch that looks like other kids’ lunches. If you put cold formula in an insulated lunch bag in the morning, it will usually stay cold until lunch time. Measure out the foods your child should eat and pack these in small containers or baggies. Low-protein bread for sandwiches, raw vegetables, fruits, and low-protein cookies can help his lunch look “normal.” If your child knows to bring home any uneaten foods from his lunch, you can keep better track of what he needs for supper to meet his daily requirements.

• Encourage your child to participate in school activities, even though arrangements may have to be made for meals. The diet for PKU should be treated as a small inconvenience in an otherwise normal life.

• Develop the habit of treating each day as important to the health of your child. In addition to keeping a 3-day diet record before blood tests, it is helpful to keep a daily food diary. This will help you stay in the habit of recording your child’s diet. A spiral-bound notebook kept in the kitchen works well for this purpose, and you can also note if your child is sick. Then, when his blood level is high, you can go back through the food diary and try to determine what caused it.

• Teach your child the need to be aware of what she eats and be consistent in drinking the special metabolic product. Help your child learn to make appropriate food choices.
Glossary Of Terms Associated With PKU

**Amino acids** are building blocks of proteins. Phenylalanine and tyrosine are amino acids. Some amino acids are made by the body, but “essential” amino acids are not and must be supplied in the diet. Phenylalanine is an essential amino acid.

**Autosomal recessive** is a form of inheritance requiring the presence of two genes for a trait in order for the individual to show the trait. Persons with only one of an autosomal recessive gene pair are carriers of the trait but are unaffected by the trait.

**Blood levels** of phenylalanine are determined by tests done on blood after it is drawn from the heel, toe, or finger. Blood levels of phenylalanine should be in the range of 120-360 umol/L (2-6 mg/dL).

**Collaborative study** is a research study on PKU. Because no one clinic in the country has enough patients with PKU to answer all the questions we have about the disorder, several major medical centers collaborated in gathering data to learn more about PKU and improve treatment methods. This long-term project determined much of what we now know about PKU, and research continues at many PKU clinics.

**Enzymes** are proteins responsible for causing or speeding up biochemical reactions without being altered themselves by the reactions. The enzyme phenylalanine hydroxylase causes phenylalanine to be broken down into tyrosine.

**Equivalent or exchange** is a measured amount of food, such as cereal, vegetable, or fruit, that has a certain amount of phenylalanine. Some clinics use an “exchange” system to calculate intake of phenylalanine. An “exchange” contains approximately 15 milligrams of phenylalanine. Patients on diets for treatment of PKU should eat a limited number of exchanges per day, determined in consultation with their nutritionist and based on their size and growth needs.

**Genetics** is the study of how characteristics are inherited. Genes contain information the body uses to produce a specific trait or to perform a particular body function.

**Growth measurements** help you and the PKU clinic team know that your child is growing properly. These measurements are made at various times for all children, not just for children with PKU. They typically include measures of your child’s height, weight, and head circumference.

**Guthrie test** is a blood test used in screening for PKU; newborn screening is mandatory in the United States. It does not actually diagnose your child with PKU, but it indicates to the physician that your child has a high probability of having PKU. Further tests are needed to make a definite diagnosis.

**Hyperphenylalaninemia** is a term meaning high levels of phenylalanine in the blood. It is a group of disorders in which elevated blood phenylalanine levels are a primary characteristic.

**Phenylalanine** is one of several amino acids that make up proteins. A certain amount of phenylalanine is needed for normal growth and development. Because the body cannot make phenylalanine, it must be supplied in the diet. Phenylalanine is an “essential” amino acid.

**Phenylketonuria (PKU)** is an inherited disorder in which the body lacks the enzyme needed to convert phenylalanine to tyrosine. In PKU, excess phenylalanine accumulates in the blood and tissues and can damage the brain and nervous system if left untreated.

**Tyrosine** is an amino acid which is needed for normal growth and development. It is typically made by the body from phenylalanine, but the person with PKU can’t make this amino acid and must get it from his diet. Tyrosine is present in phenylalanine-free formulas such as Phenyl-Free® 1, Phenyl-Free 2, and Phenyl-Free 2HP.
Diet Management

A Babysitter’s Guide to PKU. Trahms CM, Cox C. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.

Finger Foods are Fun. Trahms CM, Cox C, Luce P, Padgett D. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.

Games that Teach: Learning by Doing for Preschoolers with PKU. Trahms CM, Cox C, Luce P. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.

New Parents’ Guide to PKU. Trahms CM, Luce P. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.


Cookbooks & Food Lists

Chef Lophe’s Phe-Nominal Cookbook. Barr LA, Trahms CM. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.


Low Protein Cookery for PKU, 2nd edition. Schuett VE. Over 450 recipes for the PKU diet with phenylalanine, protein, and calorie content; suggestions for managing the diet. See www.pkunews.org for ordering information.

Low Protein Food List for PKU. Schuett VE. Over 3,000 foods (including brand names) with phenylalanine, protein, and calorie content. See www.pkunews.org for ordering information.

Making the Change from High Phe to Low Phe: Changing recipes to fit your low phenylalanine meal pattern. Fink KJ, Trahms CM. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.

Creative Family Cooking. The Mid Atlantic Connection for PKU and Allied Disorders. P.O. Box 6086, Lancaster, PA 17607.

General Information


Growth & Development; Parenting


For Children & Young Adults

*The Essentials of PKU: An Informational Pamphlet for Young Adults with PKU and Their Significant Others*. Brohman S, Trahms CM. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.

*A Journey into the World of PKU*. Wessel KW. For children and teenagers. $12.00, 1991. Send orders to: Kenneth W. Wessel, Johns Hopkins University, Center for Medical Genetics, Blalock 1008, 600 N. Wolfe St., Baltimore, MD 21287-4922.

PKU for Children: Learning to Measure. Feucht S, Trahms C. Available from the PKU Clinic at the University of Washington, Seattle, WA. Telephone: (206) 685-3015.

*You and PKU*. Taylor M, Schuett VE. For children aged 3-6 years. See www.pkunews.org for ordering information.

The Young Woman with Mild Hyperphe. Castiglioni L, Rouse BM. $5.00, 1992. Send orders to: PKU Section, Child Development Division, Department of Pediatrics, UTMB, Galveston, TX 77555-0319. Telephone: (409) 772-2356.

The Young Woman with PKU. Castiglioni L, Rouse BM. $5.00, 1992. Send orders to: PKU Section, Child Development Division, Department of Pediatrics, UTMB, Galveston, TX 77555-0319. Telephone: (409) 772-2356.

Other Resources


Genetic Counseling:
National Society of Genetic Counselors Telephone: (610) 872-7608. Available at: www.nsgc.org.
For ordering information, call 800-BABY123 or order online at www.store.enfamil.com