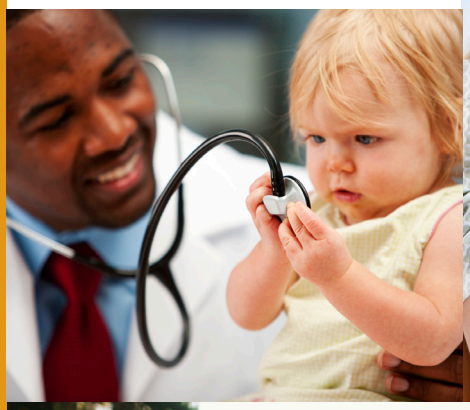
National PKU Alliance



My PKU Binder

P.O. Box 501
Tomahawk, WI 54487
www.npkua.org

PKU Handbook
A Guide for PKU from Diagnosis to Adulthood

Provided by the National PKU Alliance
www.npkua.org

Acknowledgements

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Personal Health Information

HEALTH INFORMATION AND HISTORY			
Name:			
Address:			
Phone:		Cell Phone:	
E-mail:			
Date of Birth :			
Emergency Contact:			
Relationship:			
Phone:		Cell Phone:	
Current Medications:		Daily Prescribed Dose:	
Medical Formula:		Daily Prescribed Amount:	
Clinic Information:			
Address:			
Phone:		Fax:	
Primary Care Physician:			
Address:			
Phone:		Fax:	
PKU Dietitian:			
Address:			

Phone:		Fax:	
PKU Nurse:			
Adress:			
Phone:		Fax:	
PKU Social Worker:			
Adress :			
Phone:			
Genetic Counselor:			
Address:			
Phone:		Fax:	
Other Specialists:			
Address:			
Phone:		Fax:	
Other health issues being treated:			
Major illnesses or hospitalizations:			
Other Information:			

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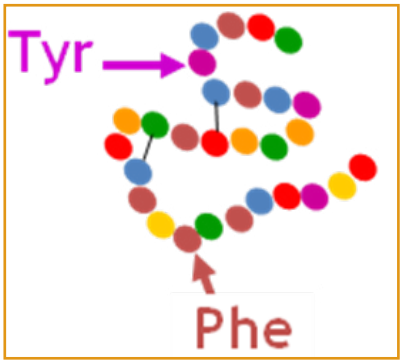
Chapter 1: What is PKU?

A Parent's Perspective

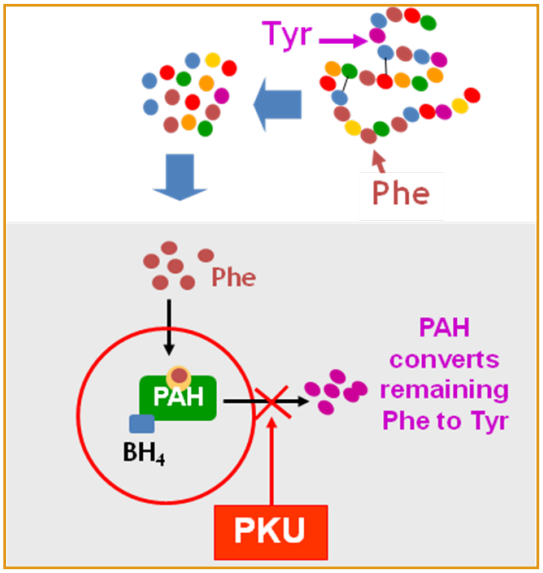
"If our child with PKU had been our first instead of our third, or if we had wanted more children after we had her, we would have done so, even knowing the child could have PKU. Because after three or four years we saw that our daughter's quality of life was no different from that of our other children. She was quite early walking and talking, she is athletic, and lovely. She was just as normal as her siblings."

What is PKU?

Phenylketonuria (PKU) is an inherited genetic disorder that affects approximately one in 15,000 babies born in the United States[1]. A person with PKU does not produce enough of an enzyme in their liver called phenylalanine hydroxylase (PAH). This enzyme is needed to process the amino acid, phenylalanine (Phe), which is found in food items that contain protein[2]. Usually, when people eat protein, it is broken down into different amino acids which are then processed and used for growth and repair of body tissue. Individuals with PKU cannot process Phe in this way, and instead Phe builds up in the blood.



All protein is composed of chemicals called amino acids, joined to each other like beads on a string. When protein is consumed, the string is broken up in the stomach into shorter chains of amino acids and then into individual amino acids in the digestive tract. These individual amino acids are absorbed into the bloodstream and processed for building muscle, making other chemicals in the body, or providing energy.



In people who do **not** have PKU, some of the Phe is converted directly into another amino acid, tyrosine, with the aid of the enzyme PAH. Tyrosine, generally derived from the protein in food, is essential for several functions, including functions in the brain.

[1]National Institutes of Health. Phenylketonuria: Screening and Management National Institutes of Health Consensus Development Statement Available At: http://consensus.nih.gov/2000/2000phenylketonuria113html.htm
Accessed May 17, 2011

[2] National Institutes of Health. Phenylketonuria: Screening and Management National Institutes of Health Consensus Development Statement Available At: http://consensus.nih.gov/2000/2000phenylketonuria113html.htm
Accessed May 17, 2011

What is PKU?

People with PKU cannot convert Phe to tyrosine because their livers do not make an adequate amount of PAH. When a person with PKU consumes protein-rich foods, his or her body cannot break down most of the Phe. As a result, the bloodstream is flooded with excess Phe that is carried to the brain by "transporter cells." The high Phe causes the brain of an individual with PKU who eats protein-rich food to be overloaded with Phe, causing cognitive as well as behavioral problems and potentially insufficient tyrosine levels, an amino acid that is a precursor to neurotransmitters and hormones[3].

There are different categories of PKU that are determined based on diagnostic Phe level, mutation testing (if performed) and the amount of Phe that an individual can tolerate.

- Classical PKU is the most common form of PKU. It results when there are two severe mutations of the PAH gene and as a result there is little or no PAH enzyme activity to convert phenylalanine to tyrosine. These are the most severely affected patients.
  Phe level in blood: Above 20 mg/dl (>1200 µmol/L)

- Moderate/Mild PKU is associated with elevated Phe levels that require treatment but are lower than those observed in Classical PKU.
  Phe level in blood: Above 6 mg/dl but less than 20 mg/dl (360- 1200µmol/L)

- Hyperphenylalanemia (HyperPhe) is used to describe those people whose blood Phe level is above normal, but still low enough that they may not require dietary treatment.
  Phe level in blood: less than 6 mg/dl (<360 µmol/L)[4]

PKU CLASSIFICATION	PHE LEVEL
Unaffected individual	1 mg/dl (60 µmol/L)
HyperPhe	< 6 mg/dl (<360 µmol/L)
Moderate/Mild PKU	6 mg/dl – 20 mg/dl (360- 1200µmol/L)
Classical PKU	>20 mg/dl (>1200 µmol/L)

[3] Acosta B, Yannicelli S, Marriage B, et al. Nutrient intake and growth of infants with phenylketonuria undergoing treatment. J Pediatr Gastroenterol Nutr 1998:27:287-91
[4] Maltzman, S, PKU & Protein : My PKU Toolkit A Transition Guide to Adult PKU Management. New Jersey : Applied Nutrition Corp. 2007:5

What is PKU?

What are the symptoms and treatment for PKU?

With newborn screening being widely used for PKU, infants are diagnosed early and symptoms are prevented with early, life-long treatment. When PKU is untreated, however, blood Phe levels can become elevated. A high level of blood Phe is toxic to the brain, the following symptoms can occur when untreated or treated too late:

- Mental retardation
- Seizures
- Eczema (skin rash)[5]

PKU is treated with a low Phe diet that includes medical formula and foods specially formulated to be low in Phe and provide essential nutrients needed for growth and development[6]. Some patients with PKU are treated with a combination of diet and a medication (Kuvan®, sapropterin, sapropterin dihydrochloride) which lowers the blood Phe level. A small fraction of patients are treated with medication alone.

Regular visits to the PKU clinic are part of the treatment program. Phe levels are checked by performing regular blood tests and treatment is adjusted if necessary to keep Phe levels in the safe range (see chapter 2 for more details on treatment of PKU).

Key Points about PKU

- PKU is a genetic condition that is not contagious.
- Apart from needing a special diet, a person with PKU is healthy.
- A person with PKU can not break down an amino acid called phenylalanine (Phe), found in all foods containing protein.
- Phe can build up in the blood and damage the developing brain.
- Staying on a low protein diet keeps Phe levels in a safe range, allowing for normal development and a healthy life.
- Eating the wrong foods will not make a person with PKU sick right away, but will cause problems over the long-term.
- Having food that is not part of the diet should not be considered a "treat" as it will have implications for an individual with PKU.
- A person with PKU will not outgrow it and must stay on the diet for life.

[5] Mitchell, J.J., Scriver, C.R., Phenylalanine Hydroxylase Deficiency In: GeneReviews [*Internet*]. Seattle (WA): University of Washington, Seattle; 1993-2000 Jan 10 [updated 2010 May 04].
[6] Mitchell, J.J., Scriver, C.R., Phenylalanine Hydroxylase Deficiency In: GeneReviews [*Internet*]. Seattle (WA): University of Washington, Seattle; 1993-2000 Jan 10 [updated 2010 May 04].

What is PKU?

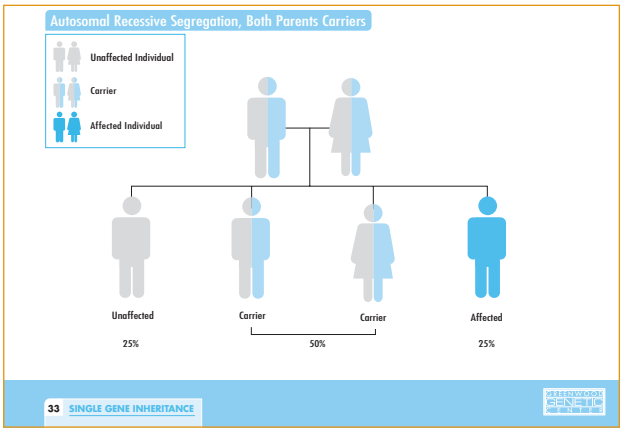
The Genetics of PKU

Genes are present in every cell in our body. They provide the "blueprint" for each characteristic or trait that we possess, such as eye and hair color, height, and gender. Genes also provide the instructions to make enzymes for metabolism. Children inherit two sets of genes, one from each parent. Not every gene always works properly.

What are the chances of inheriting PKU?
Children born with PKU have inherited a defective PKU-related gene from each parent. Around 1 in 50 or 60 of us has a defective PKU gene, yet only 1 in 15,000 Americans inherit two defective genes and has PKU. Boys and girls have an equal chance of inheriting PKU.

The pattern of PKU inheritance is recessive. A recessively inherited condition typically affects males and females equally. For a recessive disorder to happen, a child must inherit one copy of a disease-causing gene from each parent. When this occurs, the child gets a double dose of the disease-causing gene and no normal copy of the gene.

Image of two unaffected carriers and the probability of their child inheriting PKU.



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For a child to inherit PKU, both parents must be a carrier of the PKU gene. Each parent is called a "carrier" because he or she silently carries one copy of the disease-causing gene paired with a normal copy of the same gene. Carriers will not experience any noticeable symptoms. A parent who is a carrier for PKU produces either a normal gene or a PKU gene in each egg or sperm. If both parents are carriers, a child must inherit one copy of the PKU-causing gene from each parent to have PKU.

A baby conceived by two carriers of PKU has a:

- 25% chance of having PKU (inheriting two defective genes)
- 50% chance of being a carrier, who will not have any symptoms of PKU (inheriting one PKU gene and one normal gene)
- 25% chance of inheriting two normal genes

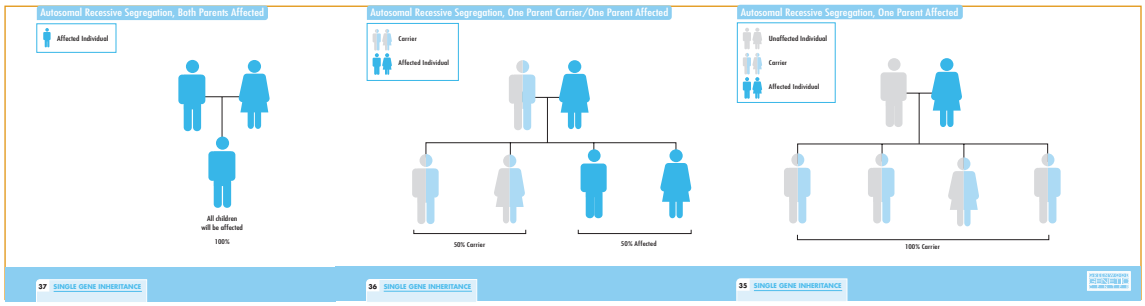
There is a 25 percent chance that another child born to the same parents will also have PKU. Individuals with PKU can have children that do not have PKU if their partner is unaffected. However, their children will be carriers. For more information about PKU patients and pregnancy, see the "Pregnancy" section of this handbook.

What is PKU?

Image of two people with PKU and the probability of their child inheriting PKU.

Image of one person with PKU and one carrier and the probability of their child inheriting PKU.

Image of one person with PKU and one non-PKU/non-carrier and the probability of their child inheriting PKU.



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PARENT 1	PARENT 2	CHILD
PKU	Non-PKU, non-PKU carrier	100% chance of being a carrier of PKU
PKU	Carrier	50% chance of having PKU 50% chance of being a carrier
PKU	PKU	100% chance of being affected with PKU

Parents may be concerned about whether they should have more children. Having more children after having a child with PKU is an individual decision for each family; however, because PKU can be successfully managed, many parents feel there is no reason not to have more children[7].

[7] Mitchell, J.J., Scriver, C.R., Phenylalanine Hydroxylase Deficiency In: *GeneReviews [Internet]*. Seattle (WA): University of Washington, Seattle; 1993-2000 Jan 10 [updated 2010 May 04].

Chapter 2: Treatment and Diet Overview

The PKU Clinic Team

The phenylketonuria (PKU) clinic team consists of a group of healthcare professionals trained to support individuals with PKU. You will see them on your regular clinic visits. These visits are extremely important to monitor your PKU and to adjust your diet if required.

- The physician oversees PKU treatment which includes monitoring Phe levels and other labs, growth and developmental progress, prescribing medication such as Kuvan and monitoring routine childhood medical concerns.
- The registered dietitian works closely with you to plan nutritious dietary choices with formula and low protein foods as well as to monitor blood Phe levels and growth.
- The genetic counselor provides education regarding the genetics of PKU and assists the physician with monitoring PKU treatment. Some clinics also have nurses to assist the physician with monitoring PKU treatment.
- The social worker organizes group and individual discussion sessions with patients and parents to support those affected by PKU.
- Some clinics have a psychologist that performs neurological and psychometric assessments at certain ages to ensure children with PKU are developing at an appropriate rate.

Diet Essentials

Formula: Defining Medical Foods

For people with PKU, medical food is vital to treat PKU and ensure good health. Medical foods include medical formula and foods modified to be low in protein. Because protein intake is restricted, medical formula provides all the essential amino acids found in protein except for Phe. It also provides tyrosine, vitamins, minerals and trace elements that most people who do not have PKU would get from their diet.

Your PKU clinic physician will provide a prescription for medical foods. Taking the prescribed amount of medical formula each day is essential. Low protein medical formulas are made to suit the nutritional needs of people with PKU at different ages, and are available in a variety of forms and flavors to suit different lifestyles and preferences. Your dietitian will assess your nutritional needs and provide a recommended formula to meet these.

Medical formula helps to:

- give energy throughout the day
- build muscle
- increase strength
- maintain brain function and analytical thinking
- keep your Phe levels in control and keep you healthy.

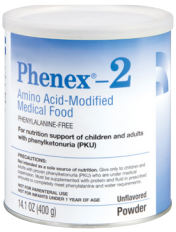
Why is the PKU diet different for different people?
The amount of Phe needed and tolerated by each person with PKU can be different depending on the severity of their PKU. Your PKU dietitian will help you create a specialized diet based on your need. In addition, your PKU team will adjust your diet according to your blood Phe levels, which means that your prescribed diet may vary from time

Treatment and Diet Overview

Foods modified to be low in protein are defined as manufactured products that will deliver no more than one gram of protein per serving. Low protein food products supply needed additional calories without supplying additional phenylalanine containing protein. This helps prevent catabolism (the breakdown of protein in the body ie muscle) which in itself can cause Phe levels to rise. Use of low-protein products, especially when used consistently, greatly improves adherence to the treatment.

Types of medical foods:

- The Drink: Formula in the form of a powdered drink mix. This is the traditional and most common type of PKU formula.
- The Low Fat Formula: A reduced fat version of the traditional formula for adults.
- The Bar: Convenient PKU medical food in the form of a bar that can be eaten on the go.
- The Fortifier: Concentrated amino acid formulas that can be added to any low protein food or beverage you already enjoy. It can also be added to traditional formula to increase the protein content without added volume.
- The Tablet: You can supplement your diet with liquid formula or take multiple tablets daily to meet your formula needs. Just remember to meet your daily fluid requirements to ward off dehydration[8].
- Ready to Drink: These convenient formulas are prepared in advance, ready for you to drink wherever you are. These prepared drinks have the protein you need with the essential amino acids and nutrients you're used to getting from your formulas. You can find a variety of flavors, and some are even freezable for hot days when you need a cool alternative for your formula needs.
- Glycomacropeptide (GMP) foods: GMP is used to make formulas (including some ready-to-drink formulas) as well as some PKU-friendly food. This whey-based protein is produced when making cheese. It is the only known dietary protein that contains a minimal amount of Phe. GMP foods have also been found to significantly lower Phe levels in blood[9]. Foods made with GMP provide an alternative to the amino acid medical foods currently required in the PKU diet.



[8] Maltzman, S, PKU Diet Management: *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey: Applied Nutrition Corp. 2007:9

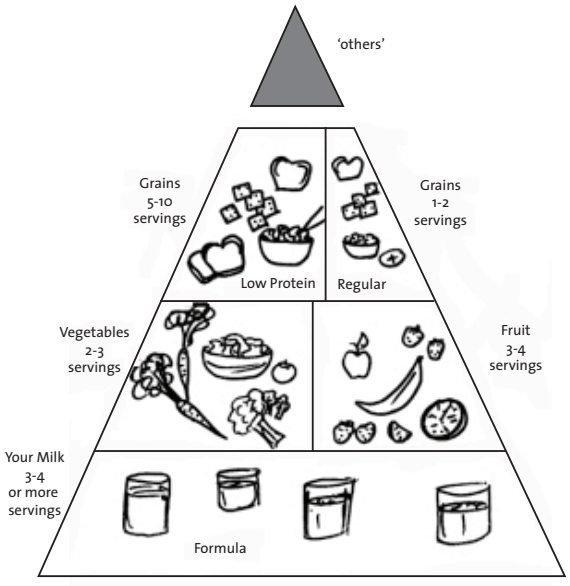
[9] van Calcar, S.C, MacLeod, E.L., Gleason, S.T., Etzel, M.R., Clayton, M.K., Wolff, J.A., and Ney, D.M., Improved nutritional management of phenylketonuria by using a dietcontaining glycomacropeptide compared with amino acids A Journal of Clin Nutrition 2009;89:1068–77

Treatment and Diet Overview

The Low-Protein Diet

People with PKU get most of the protein they need from medical formula, the backbone of the PKU diet. The remaining protein – which includes the essential amount of Phe the body needs for growth and functioning – comes from food. The PKU diet consists of low protein foods such as:

- Fruits
- Vegetables
- Foods modified to be low in protein, including low-protein pasta, rice and breads
- Fats and sugar



Foods such as red meat, chicken, fish, eggs, milk, yogurt, cheese, nuts, soybeans and beans are too high in protein to include in a PKU diet, except when the dietary restriction required is minimal. Foods such as regular pasta, bread, rice and starchy vegetables (potatoes, peas, corn etc) will likely be limited in your diet as well. Your dietitian will instruct you as to which foods and how much you or your child can eat.

Even low protein foods cannot be eaten in unlimited quantities. All foods must be measured to make sure that Phe intake stays within the daily limit.

It is helpful to incorporate the PKU diet into the family meal. This can be done by basing your child's PKU meal on the vegetables or grains the rest of the family will be eating. For example, if your family is having pasta with meat sauce for dinner, you can prepare low protein pasta with tomato sauce for you or your child with PKU.

Counting Phe/Protein

One study shows that the more mothers know about counting Phe, the better their child's dietary compliance is based on the child's blood Phe levels[10]. It is important for all individuals with PKU to count the protein in foods for meal planning and tracking Phe intake. There are many different ways and styles to keep track of Phe in food. Your dietitian will counsel you on the system the clinic uses and which method may work best for you. They will also inform you on how much protein, Phe, or exchanges is needed each day and provide you with a food reference guide.

What is a food reference guide?
A food reference guide shows how much protein and Phe common foods contain, which helps you figure out and plan how much protein and phe you will eat at each meal or snack. It also lists "free foods", i.e. foods that contain little or no protein, which do not need to be counted. The dietitian at your PKU clinic may give you a reference guide and help you use it.

Treatment and Diet Overview

The most accurate way to measure serving sizes is to use a food scale.

How to measure formula or food using a scale

1. Make sure the scale is on a level surface.
2. Turn on the scale. The scale should read "0 g" which means that the scale is "zeroed". If it reads something other than "0 g", push the "On/Off Zero" button once and the scale should read "0 g".
3. When the scale reads "0 g", place the container to measure the powder or food on the scale. Once it has read the weight press the "On/Off Zero" button to zero the scale.
   The screen should read "0 g" again.
4. You can then start adding the food or formula powder to measure the grams needed. Repeat the above instructions each time you need to measure formula or food.

You can also measure formula and foods using measuring cups, although this method is not as accurate as using a scale. Your food reference guide lists both gram weights and cup measurements for many common foods.

This section provides three methods for keeping track of Phe. The common factor between each method is to know your portion size and measure what you eat[11].

Method 1: Counting Milligrams (mg) of Phe

Counting milligrams is the most accurate method of keeping track of how much Phe is consumed. When using this method to keep track of Phe, it is helpful to have a food reference guide that tells you how much Phe is contained in the food and beverages being consumed (see the Chapter 15 Resources for recommended food reference guides). If the food you are preparing is in the reference guide with the amount that you plan on serving, you just need to record the amount listed in the guide. It is also important to pay attention to the brand name of the product. Different brands of a similar item may not have the same amount of Phe due to the ingredients and amounts used.

Measuring using weight

You will need your kitchen scale and a calculator.

1. Weigh the food you are going to serve.
2. Multiple the gram weight of the food by the number in the "Mg Phe/Gm Food" for that food in the reference guide.

For example, if you are serving 9 grams of a particular cereal, find the amount of Phe in 1 gram from the "mg Phe/gm food" column in your reference guide. If the amount is 3.5 mg Phe/gm of food, you would have:

9 grams of this cereal x 3.5 = 31.5 grams of Phe

How do I convert grams of protein into milligrams of Phe?
To convert grams of protein into milligrams of PHE, always multiply the number of grams by 50. So, for example, if a serving size has 1.5 grams of protein, you can do the following calculation:

1.5g protein x 50 = 75mg Phe
Use a calculator, if you need to, to make sure you are getting an accurate count every time.

[10] H. Gokmen Ozel, H., Kucukkasap, T.,Koksal, G., Kalkanoglu Sivri, H.S., Dursun, A., Tokatli, A., Coskun, T., Does maternal knowledge impact blood phenylalanine concentration in Turkish children with phenylketonuria? JIMD Short Report #111 (2008)
[11] Maltzman, S, PKU Diet Management : My PKU Toolkit A Transition Guide to Adult PKU Management. New Jersey: Applied Nutrition Corp. 2007:15

Treatment and Diet Overview

Measuring portions

To measure portions using measuring cups or spoons, you will need the cups or spoons and a calculator. This may be slightly more difficult than using weight.

1. First, you will need to convert any fractions to decimals. To do this, divide the number on the top of the fraction by the number on the bottom. For example, for ¼, 1 ÷ 4 = 0.25. Or ¾ is 3 ÷ 4 = 0.75.
2. Divide the portion size of the serving you are using by the number you will find in the "Measurement" section of your food reference guide.
3. Multiple this number by the number in "Phe mg" column of your food reference guide. This will give you the amount of Phe in the serving you are using.

For example, you want to serve ½ cup of a particular cereal, but the cereal is listed using ¾ cup.

1. 1÷2 = 0.5 is your serving. 3÷4 = 0.75 is the serving size listed.
2. Divide your serving by the serving listed. 0.5 ÷ 0.75 = 0.66
3. Multiply 0.66 by the number in the "Phe mg" from the food reference guide. If it is 45 mg Phe, it would be:

0.66 x 45 = 30 mg Phe

If you are unable to obtain the amount of Phe in a food or beverage, it is possible to estimate how much Phe it contains by the serving size and nutrition information on a food label (see example of a food label below). Commit to memory that 1 gram of protein contains about 50 milligrams of Phe. If you multiply the number of grams of protein by 50 you can get a rough estimate of how much Phe is contained in that food or beverage[12]. Remember, this is only an estimate and is not as exact for calculating Phe content. Sometimes food manufacturing companies may be able to give you additional nutrition information or call your dietitian to see if they are able to find more information on a particular food.

1 gram of protein = 50 mg Phe

For example, if the food has 4 grams of protein, multiply it by 50 to get 200 milligrams Phe.

4 g of protein X 50 (number of mg Phe in each g of protein) = 200 mg Phe

[12] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey: Applied Nutrition Corp. 2007:17

Treatment and Diet Overview

Method 2: Counting Exchanges of Phe

Keeping track of Phe intake by counting exchanges was developed to help make the process of calculating your intake easier. Phe exchanges are values assigned to a food item for easy reference. Exchanges can be found in most food reference guides[13] and make use of decimals rather than percentages to make calculations easier. Counting exchanges is not as detailed and accurate as counting Phe by milligrams, but counting by exchanges is based on:

1 g protein = 50 mg Phe
1 exchange = 15 mg Phe
1 g protein = 3.5 exchanges

If you are using the same measurement as in the book you are using, it is easy to just record the number of exchanges listed. If you are not using the same amount, you will need to measure the milligrams of Phe using method 1. For example, if you measure the food and determine that there is 45 mgs of Phe, you would divide 45 mgs of Phe by 15 (the milligrams of Phe in 1 exchange) to determine that the food you are preparing has 3 exchanges.

If you know that you or your child can have 20 exchanges of Phe per day, that means you or your child can have 300 mgs of Phe per day. Rather than counting to 300 every day, you just have to get to 20!

Method 3: Counting Grams (g) of Protein[14]

Counting grams of protein is generally used for people with a higher Phe and protein tolerance. A "Nutrition Facts" label is required for most packaged foods in the United States. For people with PKU, the most important nutrition facts listed are **serving size** (by grams or pieces) and **protein** (grams) based on the serving size listed. Before eating any packaged food, check the Nutrition Facts label to see how much protein is in each serving, and weigh or count out your serving to make sure that you are only eating the amount of protein you have planned for that snack or meal.

TOMATO SOUP

Nutrition Facts
Serv. Size: 1/2cup (120ml)
Condensed soup
Servings about 2.5

Calories 90	Dietary Fiber 1g
Fat Calories 0	Sugars 12g
Total Fat 0g	Protein 2g
Sat. Fat 0g	Potassium 690mg
Trans Fat 0g	
Polyunsat. Fat 0g	% Daily Values**
Monounsat. Fat 0g	Vitamin A 8%
Cholesterol 0mg	Vitamin C 10%
Sodium 480mg	Calcium 0%
Total Carb. 20g	Iron 4%

While this may not be the most accurate way to keep track of the amount of Phe you consume, counting grams of protein might be the easiest way for you to become accustomed to a low protein diet.

Be aware that Nutrition Facts labels aren't always exact. Due to the food labeling laws, items that say, "Protein 0g" may actually contain up to 0.49 grams of protein per serving. If a label says that one serving of a given food item has 1 gram of protein, it can have anywhere from 0.50 to 1.49 grams of protein. If you do not have a PKU food list or if it is a new product and the label says "Protein 0g", it is best to assume that it has ½ gram of protein[15]. You can also add 0.50g to any rounded number to come up with the maximum grams of protein in each serving.

[13] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:18
[14] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:15-16

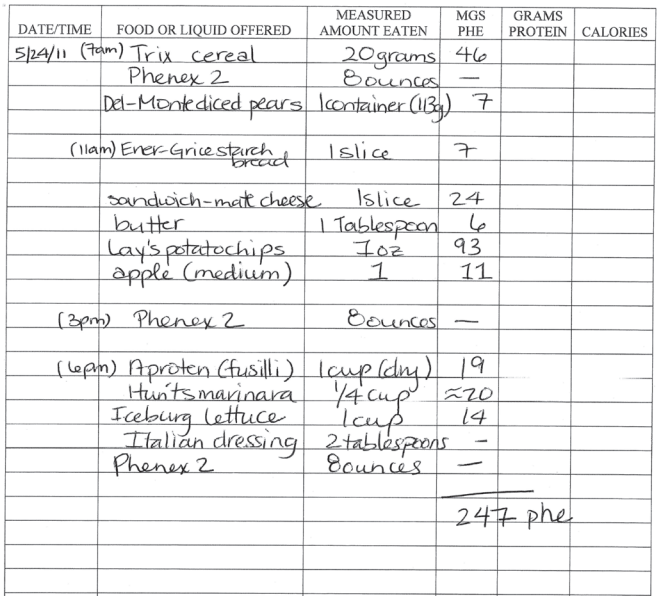
Treatment and Diet Overview

Filling Out a Diet Record

Keeping a diet record, or food log, helps you track the amount of Phe being consumed each day so that Phe intake stays within the daily limit. (A blank diet record can be found in the resources in Chapter 14). Tracking intake of food, drinks and formula will help you maintain the PKU diet and help your dietitian determine how to adjust you or your child's diet when necessary. Your PKU team will typically ask for a diet record when you send in a blood Phe sample.

You will need to keep track of the information below. It may be helpful to add up the Phe as you go so you can see the total that has been consumed so far for that day. This will help you figure out how much more Phe can still be eaten that day to stay within the limit

- **Date/Time:** Record Date and time the food/drink was consumed.
- **Food or Liquid Consumed:** Record the name of the food or liquid (including formula) and be specific as possible (apple vs. fruit, include brand names if possible).
- **Measured Amount Eaten:** Record specific measurements like grams, tablespoons, tea spoons, cups. If a few bites were eaten, record as "3 bites". The amount is just as important as the type of food eaten.
- **Milligrams of Phe:** Look up the amount of Phe in your food reference guide and record the amount of Phe that is accurate to the amount of the food eaten.
- **Grams of Protein and Calories:** This is to record the amount of protein and calories that are in foods. This is to be recorded if this information is available.
- **Daily Totals:** Record the total Phe, Protein and Calories (when available) consumed that day.



DATE/TIME	FOOD OR LIQUID OFFERED	MEASURED AMOUNT EATEN	MGS PHE	GRAMS PROTEIN	CALORIES
5/24/11 (7am)	Trix cereal	20grams	46		
	Phenex 2	8ounces	—		
	Del-Monte diced pears	1container (113g)	7		
(11am)	Ener-G rice starch bread	1slice	7		
	sandwich-melt cheese	1slice	24		
	butter	1 Tablespoon	6		
	Lay's potato chips	1oz	93		
	apple (medium)	1	11		
(3pm)	Phenex 2	8ounces	—		
(6pm)	Aproten (fusilli)	1cup (dry)	19		
	Hunt's marinara	1/4 cup	≈20		
	Iceburg lettuce	1cup	14		
	Italian dressing	2tablespoons	-		
	Phenex 2	8ounces	—		
			247 phe		

[15] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey : Applied Nutrition Corp. 2007:16

Treatment and Diet Overview

Diet and Treatment During Illness

Illness such as high fevers and stomach distress, surgery, and injuries such as bone fractures will affect Phe levels. The most important steps when this happens are:

1. Treat your or your child's illness like you normally would.
2. Formula should be continued as tolerated.
3. Continue the diet if you or your child are able to tolerate food.
4. Stay in contact with your PKU team if you have questions about the diet as it may need to be adjusted during illness.

When a person is sick, energy needs increase and the person may not take in enough calories. When this happens, the body dips into muscles for nutrients, which is called being "catabolic." Your muscles are made up of protein, therefore they contain Phe. When you are catabolic, you break down muscle which releases Phe into your blood, causing your Phe levels to go up. This is why it is important for a person with PKU to continue to consume formula on a daily basis at regular intervals. The nutrition provided by formula will help give you or your child the energy to fight off illness and disease. It is also important to stay hydrated and consume enough calories while sick.

Frequency of Phe monitoring may also need to increase during times of illness to ensure Phe levels are not too high or too low. Call your PKU team with any questions about Phe levels when sick.

<u>Other Treatment Options for PKU</u>

KUVAN®

KUVAN (sapropterin, sapropterin dihydrochloride) is an FDA-approved medication that is used as a treatment method for patients with PKU who respond to sapropterin. KUVAN works by helping the phenylalanine hydroxylase (PAH) enzyme work more effectively to break down Phe in the body.

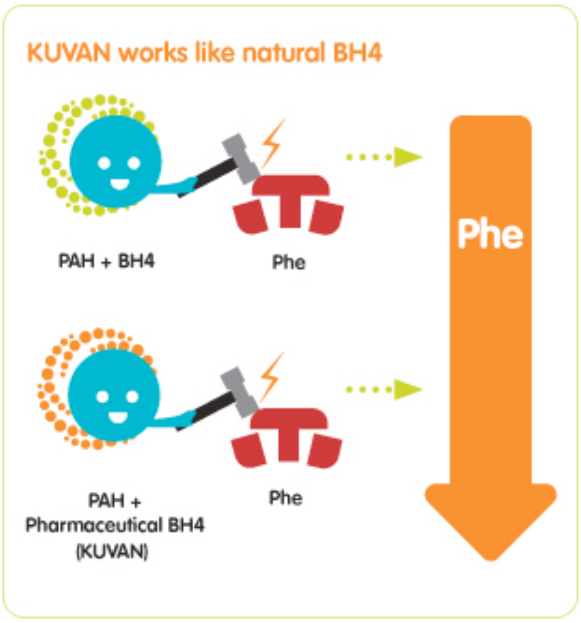
How KUVAN Works

KUVAN is the pharmaceutical form of a naturally occurring substance in the body called BH4. BH4 is a cofactor, or helper, of the PAH enzyme; it aids the PAH enzyme in the breakdown of Phe. As a medication, KUVAN supplies more BH4 to the body, which helps the PAH enzyme break down more Phe and lower Phe levels. KUVAN is not a cure, but a treatment method that, when combined with diet, can help keep Phe levels under better control.

Determining Responsiveness to KUVAN

Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey: Applied Nutrition Corp. 200748-49

Treatment and Diet Overview



While KUVAN works for many, not every person with PKU is responsive to this form of treatment. This means KUVAN may not lower Phe levels for some people with PKU. When first starting KUVAN, there is a trial phase to determine responsiveness; this can last two to four weeks. The trial phase requires more frequent blood sampling, a record of dietary intake, and taking KUVAN consistently every day.

When determining whether an individual responds to KUVAN, there are a few criteria that are used, along with a physician's clinical judgment. These include a significant decrease in Phe levels; an increase in Phe tolerance; and or changes in behavior or mood.

There is no age limit for using KUVAN in the United States. Research has shown that KUVAN reduces Phe levels in KUVAN-responsive patients from infancy through to adulthood (Burton BK, 2011, Mar). The most common side effects of KUVAN are headache, diarrhea, nausea, and abdominal pain. These listed side effects are mild and will usually resolve after a short time.

Starting KUVAN
Ask your PKU team if you or your child would like to start KUVAN. See Chapter 14 Resources for more information about beginning KUVAN responsiveness trials.

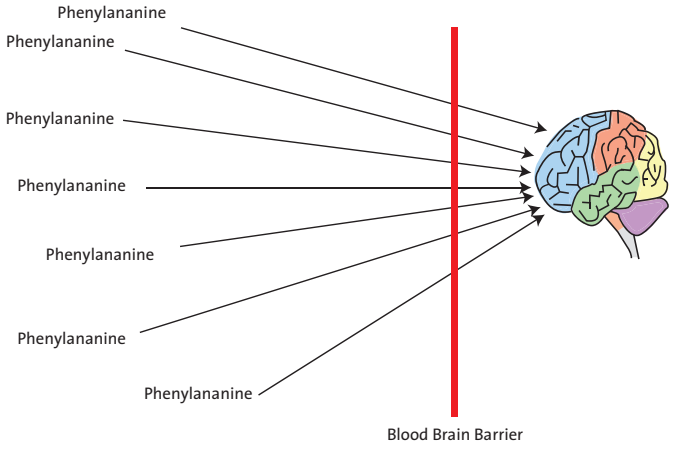
Large Neutral Amino Acids (LNAA)

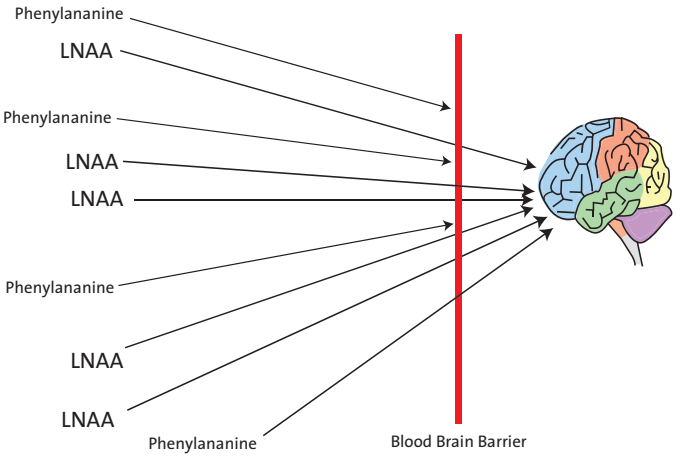
Large Neutral Amino Acids (LNAA) are a treatment option that is mostly used for adults who have difficulty in maintaining the recommended Phe levels. LNAA are considered a medical food product, and come in a powder or pill form containing certain essential amino acids (not including Phe) referred to as large neutral amino acids. LNAA mixtures contain amino acids similar to those that are found in PKU formulas, but in more concentrated amounts.

How LNAA Works
When a person with PKU consumes protein-rich foods, his or her body cannot break down most of the Phe. As a result, the bloodstream is flooded with excess Phe that is carried to the brain by "transporter cells."

LNAA pills or powder have high concentrations of specific amino acids that compete with Phe to latch on to the transporter cells. LNAA are taken with food so that the amino acids are digested along with consumed proteins and enter the bloodstream at the same time as Phe from foods. As LNAA flood the bloodstream with a large number of "safe" amino acids, more of these non-harmful nutrients enter the brain, blocking much of the Phe from being transported into the brain. This process reduces the amount of Phe transported to the brain,

Treatment and Diet Overview





helping reduce the neurocognitive effects of high blood Phe levels.

The purpose of LNAA is to decrease Phe levels in the brain; patients may not necessarily see a decrease in blood Phe levels, although some do. LNAA are used in treating patients that are struggling with the PKU diet or are off diet. LNAA must be taken with each meal and protein-containing snack. Future of PKU Research and Treatment

Future of PKU Research and Treatment

Over the last five years, there have been many great strides to improve treatment for PKU. Below is a brief synopsis of work being done by leading researchers to improve PKU treatment options.

Enzyme Substitution

Researchers discovered an alternate enzyme phenylalanine ammonia lyase or 'PAL' found in plants and bacteria that can break down Phe. On its own, this enzyme is destroyed very quickly in the body. To work, a protector (PEG), or "coater" needs to be placed around PAL to slow down the speed with which it breaks down in the body and to protect it from the body's immune response. The result is PEG-PAL, an injectable medicine that has been shown to lower Phe levels in PKU mice. The first study of PEG-PAL in human patients with PKU began in 2008, and hopes to show a decrease in Phe levels in PKU patients. Successful Phase 1 results were reported in June 2009.

Gene and Cell Therapy

Gene therapy is an ideal treatment for PKU because it would provide functional PAH enzyme in the liver cells which could break down Phe. Unfortunately, the challenge in gene therapy has been the body's immune response, which mounts an attack on the transferred enzymes, like it would attack a virus. While gene therapy has worked in curing PKU in mice, immunosuppressant medications have also been needed to maintain the correction. Some new techniques have shown promise though, to cause less immune response and maintain a longer correction and be safer.

Treatment and Diet Overview

Therapeutic Liver Repopulation

Since liver cells in PKU patients are deficient in phenylalanine hydroxylase (PAH), therapeutic liver repopulation aims to replace the cells that are deficient in PAH (PAH negative cells) with cells that are not PAH-deficient (PAH positive cells). PAH positive cells would then restore function of the liver, curing PKU.

One of the challenges of liver repopulation is that the cells that have PAH do not grow any faster than the cells that do not have PAH. In order for this therapy to work, a method needs to be developed to make the cells with PAH grow faster. If this happens, it may be possible to increase the level of Phe breakdown to normal for individuals with PKU[17].

Special Considerations

Aspartame

"Phenylketonurics: Contains Phenylalanine" means this is likely a product a person with PKU cannot eat or drink.

People with PKU in general should not eat or drink any food or beverages containing aspartame. Aspartame is an artificial sweetener that contains Phe; it is commonly known as NutraSweet™ and is found in diet sodas and some reduced-sugar foods and beverages. A warning for people with PKU is listed on products that contain aspartame, but you must look carefully as the warning is often printed in small type. Some sugarless gums contain aspartame in small amounts. Based on the small amount of Phe in these products some individuals are able to include this in their diet (See chapter 10 for more details on sugarless gum).

Medications

Sweeteners such as aspartame and aspartame-acesulphame are used in some medications, especially for children. Ask your pharmacist to make sure that medications prescribed for your child do not contain these sweeteners. In some situations it is not possible to prescribe an alternative. If you have been prescribed a medication with added aspartame or aspartame-acesulphame, you should call your pharmacist to find out how much Phe is in the medication. The amount of Phe in the medication should be counted in the daily Phe intake. Contact your PKU team if you have questions about blood testing and diet adjustments.

Other medications, including capsules made of gelatin, can be a source of Phe. When buying over-the-counter medications or picking up a prescription, always check labels or ask a pharmacist if the product contains aspartame, aspartame-acesulphame or gelatin.

Exercise

Regular physical activity is an important part of a healthy lifestyle. PKU does not limit the ability to participate in exercise or sports. Regular exercise may even improve the ability

[17] Harding, C.O., Gibson, K.M., Therapeutic liver repopulation for phenylketonuria. J Inherit Metab Dis. 2010 Dec;33(6):681-7. Epub 2010 May 22.

to tolerate Phe. During exercise, you can drink extra formula to keep yourself hydrated and maintain your energy level. After exercise, your body needs fluids, carbohydrate and protein to recover. Drink plenty of fluids, especially water, and eat some foods with carbohydrates, like low protein bread or pasta.

People with PKU should not consume any protein powders or supplements that promise muscle or weight gain. These supplements contain high levels of protein and will increase your blood Phe levels. Your formula acts as a "protein shake". If you feel you may need more protein your diet, ask your dietitian to see if adjustments to your formula need to be made.

Dental Care

People with PKU may be more likely to have some tooth decay and dental erosion (when strong acids in food break down the outer layer of teeth) because their diets are more likely to include sugary foods and acidic drinks. As children with PKU do not get much protein from foods, they eat more carbohydrates and fats to give them energy and may snack more often throughout the day. In addition, medical formulas are acidic and sweetened, which means that sweet and acidic foods are coming in frequent contact with a child's teeth.

To help prevent dental problems, you or your child can:

- Take a sip of water to rinse the mouth after drinking medical formula.
- Offer water instead of juice throughout the day.
- Use a toothpaste containing fluoride.
- Brush teeth twice each day; especially as the last thing before bed at night (avoid snacks or drinks after brushing).
- Schedule regular check-ups with a dentist from an early age, and let the dentist know about you or your child's PKU.

Chapter 3: Monitoring Blood Phenylalanine Levels

A Parent's Perspective

At first it was really difficult and daunting to take my daughter's blood samples. I had to keep reminding myself that parents do what is necessary to protect their child, no matter how difficult. It definitely got easier.

Blood Phenylalanine Levels

Monitoring blood Phe levels is an important part of managing PKU). PKU and the effects of treatment are evaluated by monitoring blood Phe levels. Keeping blood Phe levels under control leads to a lower risk of brain, mood or social problems.

The ideal range for blood Phe levels is around 2-6mg/dl (120-360 µmol/L). For young children, many PKU doctors recommend striving for the lower portion of this range. The target Phe range ensures that the body is provided enough Phe for essential functions. If the level is too high, it may affect the brain, cause mood and behavior disruptions and other issues associated with high Phe levels. If Phe levels are too low for a prolonged period of time, growth may be negatively affected since there is not enough Phe to make new proteins that are part of the structure of the body. A Phe level that is "too low" is one that is below 0.5 mg/dl (30 µmol/L). A number of factors cause blood Phe levels to rise or fall outside the acceptable range, including:

PHE LEVELS MAY INCREASE WHEN...	PHE LEVELS MAY DECREASE WHEN...
Phe intake from food is too high, which causes a release of excess Phe into the bloodstream	Intake of Phe or protein in the diet is lower than recommended
Intake of food or medical formula is too low, which causes the body to break down its own muscle tissue for nutrients, releasing Phe into the bloodstream	Rapid growth in a child causes levels to fall as Phe is rapidly used to build new body tissue
A child's rate of growth has slowed, meaning less phe is being used to build new proteins	Physical activity is increased over long periods of time causing Phe to be used to build muscle
Illness, which may make it difficult to eat or drink medical formula, can cause higher Phe levels from the breakdown of the body's own muscle tissue	

If Phe levels are too high or too low, your clinic team will advise you on ways to get them back to the recommended range.

Monitoring Blood Phenylalanine Levels

Monitoring Phe Levels

Blood Phe levels are measured using a small blood sample taken from the heel or big toe of babies and toddlers, and from the fingertip of children and adults. The procedure is easy to manage once you have had a little practice. Your PKU team will usually supply special filter papers needed for the sample.

A Step-by-Step Guide to Obtaining Blood Samples

You *will need:*

- A lancing device for ample blood supply such as 21 gauge pen. Note that high gauge (ultrafine) lancets do not provide enough blood for testing blood Phe.
- Filter paper
- Rubbing alcohol and a cotton ball, or an alcohol swab
- Bandage

Photos courtesy of BioMarin Pharmaceutical Inc.

Steps to taking blood samples

1. Fill in the required details on the filter paper using a ballpoint pen. This usually includes full name and date of collection.
2. Select (or have your child select, if appropriate for age) the finger to use for the blood sample.
3. Use rubbing alcohol on a cotton ball or an alcohol swab to disinfect the finger or heel. Wipe dry with a dry cotton ball if the skin is obviously wet.
4. Press the lancet or lancet pen firmly against the heel or finger and push down (or push the button) until the lancet is released, piercing the skin.
5. Allow a large drop of blood to form on the heel or finger. If blood is not flowing freely, use pressure on the foot or the finger, pushing toward the site of the needle stick to cause blood to flow.
6. Let the blood drop onto the circle on the card. Continue to fill the circle from the center, making sure that the blood. soaks through to the back of the card. Fill in required circle(s) based on clinic's instructions.
7. Cover the puncture site with a bandage.
8. Lay the card flat so that both sides of the blood spot can air-dry for at least 2 hours.

Monitoring Blood Phenylalanine Levels

9. Place card in an envelope and send to the address provided to you by your PKU team. Don't forget to include a diet record with the sample to help make proper adjustments to the diet, if needed. Results are typically reported back to you with recommendations for any adjustments to your treatment.

While individual preference will vary, you may find you have the most success with a lancet that has some kind of control for how deeply the skin is penetrated. It is usually recommended that you begin with the highest setting and dial down (for a shallower puncture) if you find that the finger or heel bleeds very easily. It is more common for patients and parents to initially have difficulty getting enough blood. This can be because the lancet is too thin, the puncture is not deep enough, or because inadequate pressure is applied to get the blood to flow rapidly before clotting,

Frequency of Blood Sampling

Frequency of blood sampling depends on an individual's age, Phe levels and other physical changes and is determined by the PKU team. Some general guidelines are below.

AGE	RECOMMENDED FREQUENCY OF SAMPLES
First few weeks of life	1-2 times per week
Up to 12 months	1 time per week
1 year and older	2-4 times per month
Pre-pregnancy and during pregnancy	1-2 times per week (see pregnancy section for more information)
Illness	Frequency may be increased during and after illness as directed by your clinic.

Home Blood Phe Monitor

Research is being done to develop a home blood Phe monitor which will allow people to assess their blood Phe levels immediately from home. Similar to a diabetes monitor, individuals with PKU will take their blood sample and a small machine will immediately assess their blood Phe levels. This will allow dietary adjustments and monitoring on a more frequent basis, without the delay that accompanies submission of samples by mail.

Monitoring Blood Phenylalanine Levels

Suggestions for Success and Avoiding Problems

Tips for Successful Blood Sampling

- *Involve children with their blood sampling.* Blood samples are a fact of life for children with PKU. The positive patterns you help establish when your child is young will provide a solid foundation for how he or she manages blood samples throughout life. It may help to allow your child to control the flexible aspects of blood sampling, even at an early age, such as which finger or bandage to use. As your child grows, you can foster acceptance of blood sampling by encouraging him or her to play an active role in collecting blood samples. Find some specific suggestions for age-appropriate ways to involve your child in blood sampling in Chapters 4, 5, 6, and 7.

- *Use your home, office or phone calendar to "schedule" blood samples* to help you remember to take it at the same time every day. This ensures that they are a regular part of your routine.

- *To increase circulation to the hands or feet:*
  - Make sure the hand or foot is warm before taking the sample.
  - Soak in warm water, gently rub the area, or sit in a warm room before taking the blood sample.

- *Use gravity* to help blood flow by letting the hand or foot hang as low as possible.

- *Try different spots on the finger*
  - The side of the finger may be less painful because there are less nerve endings.

- *Use a topical analgesic to reduce pain and anxiety with finger sticks*
  - Pain Ease® or Emla Cream® can be provided with a prescription from your doctor

A Parent's Perspective

The first few months of tests were extremely difficult. We were using the heel and it took over 30 minutes to obtain a sample. All the while our infant was screaming bloody murder. We would get very anxious on the days we had to complete a test. Once we were able to take a blood sample from the big toe our little boy did not even notice. He actually just sits and plays while we fill the two big circles. The other day he actually giggled. So to all you new Moms and Dads it does get better!

Monitoring Blood Phenylalanine Levels

Troubleshooting


Incorrect Sample

Common problems with blood samples that can make measuring Phe levels difficult include:

- The blood spot is too small or circles aren't correctly filled. Do not worry if you "over fill" the spots by going outside the lines as this is not a problem.
- The blood spot has not soaked all the way through and saturated the back of the filter card. This most often occurs when blood flow is very slow and you are just "dabbing" the blood on the top of the card.
- The card has gotten wet.
- The card has not been allowed to dry slowly in open air.


Correct Sample

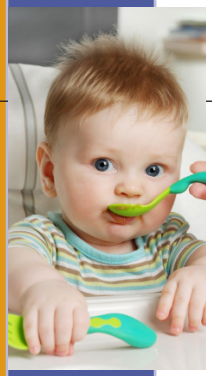
To avoid these issues, follow the step-by-step guide for obtaining blood samples and the tips. If you have any questions about blood samples, talk to your PKU team.

Blood Samples from the Vein

Occasionally a blood sample may need to be drawn from an arm vein at the PKU clinic. These samples will test blood Phe levels, but, depending on the studies ordered by your doctor, may also assess:

- **Bone health:**
  - Calcium and phosphorus levels; other serum chemistries
  - Vitamin D level
- **The adequacy of amino acid and protein intake :**
  - Prealbumin
  - Plasma amino acids
  - Tyrosine
- **General nutritional status**
  - Vitamin B12 level
  - Complete blood count (CBC)
  - Iron
  - Other trace mineral or nutrient levels as determined by your clinic

These labs will help your PKU team evaluate whether the diet needs to be adjusted to ensure you or your child with PKU are getting all the nutrients needed for healthy growth and development. Depending on your clinic, additional tests may be ordered. For example, a bone density scan (type of Xray, also referred to as a DexA scan) is commonly ordered to assess bone density and fracture risk. The frequency of these studies may vary from clinic to clinic.



Chapter 4: Diagnosis to 2 Years

A Parent's Perspective

"I still remember that call from the doctor's office saying our son's newborn screen was positive for PKU. While my husband and I grieved about the diagnosis for several months, we also slowly came to realize that PKU can be managed effectively and is just one part of who our son is."

What to Expect

All babies born in the United States are tested for phenylketonuria (PKU) approximately 24 hours after birth through the state newborn screening test. A blood sample is taken from a needle prick on a baby's heel, and the phenylalanine (Phe) level in the blood is measured in a laboratory. If the Phe level is high, more tests are done to confirm that the baby has PKU. The pediatrician will help link each family to an appropriate metabolic specialist to obtain confirmatory testing regarding the abnormal newborn screening. As soon as the diagnosis is made, treatment will be initiated to lower Phe in the blood to a safe level as high Phe levels for an extended period of time can cause brain damage. With newborn screening, early treatment and regular Phe monitoring, Phe levels can be kept in a safe range.

My baby was diagnosed at 3 days old. Could any damage have occurred?
The period of time between birth and diagnosis of PKU by newborn screening is too short to cause any problems. Children with PKU treated from early infancy are able to live normal, healthy lives.

A new diagnosis of PKU can be a time of uncertainty and even sadness. It is alarming to be told that PKU can cause a problem with your child's brain development and it may not be clear what PKU really means. The first few days or weeks can be stressful after a diagnosis is made. It is natural to have feelings of grief, disappointment, sadness and/or anger about what has happened. Most parents begin to feel more optimistic as soon as they see their child's Phe levels decrease and start to see how PKU is controlled with diet.

The early weeks and months are a time to begin sharing the experience with others and allowing trusted family members and friends to support you when possible. One of the challenges of having a child with PKU is that, because it is rare, few people have heard of it. Finding ways of explaining PKU to your family, your friends, your child, and interested others will become easier over time and as needed.

A Parent's Perspective

"I realized later that calling everyone and telling them about PKU and consoling them on the phone made my own acceptance faster. The more people I told and said, 'Look, it's all right, it's not that bad, the more I was reassuring myself it was OK."

Some people will feel ready to tell family and friends about the diagnosis soon after birth; others may prefer to wait to inform other people about the diagnosis. Waiting to share your child's PKU diagnosis allows you the ability to talk with other individuals about this information in your own time.

Some of the key points that you may want to share about PKU:

Diagnosis to 2 Years

- PKU is a genetic condition that is not contagious.
- Apart from needing a special diet, your child is healthy.
- Your child's body cannot break down an amino acid called phenylalanine (Phe), found in all foods containing protein.
- Phe can build up in the blood and damage the developing brain.
- Staying on a low protein diet keeps Phe levels in a safe range, allowing for normal development and a healthy life.
- Eating the wrong foods will not make your child sick right away, but will cause problems over the long-term. Having food that is not part of the diet should not be considered a "treat" as it will have implications for an individual with PKU.
- Your child will not outgrow PKU and must stay on the diet for life.

A Parent's Perspective

"Our son is a beautiful and extremely bright baby. He is far ahead of his brother at this age, and is growing normally. When I first heard the diagnosis of PKU, I was so worried Connor wouldn't be "okay." I cried for weeks. Now I know that Connor is more than okay; he is thriving."

Many parents find that they need support from other families who have children with PKU to feel reassured that their child will be fine[18]. You may find it useful to talk with parents of older children with PKU about how they prepare the special foods and what it is like living with the diet. Seeing other children with PKU who are growing and developing well is reassuring (see Chapter 15 for more information about PKU support groups). Speak to your PKU team who can assist you in contacting another family who has been in your position and can show you the excellent outcome that is possible for children with PKU.

As you learn to manage your child's PKU, you will gain confidence in your child's future. While managing PKU will be part of your life, it will be just that: a part of your life, and not all of it.

Development

Children with PKU are usually on target for normal developmental milestones during this time; if there are any delays, they are generally not related to PKU. At about 18 months of age, most children experience tremendous growth in cognitive, language and imaginative abilities. They also may begin to experience anxiety around aspects of their PKU diet and treatment as these cognitive shifts occur[19]. It is important to talk to your child about his or her diet and treatment, both to increase awareness of PKU and to reduce anxiety around treatment.

A Parent's Perspective

Don't panic when you read all the frightening information you find on the Internet. I spent my son's first year and a half in tears and terrified that he would be mentally impaired... He's brilliant.

Children under the age of two will usually not understand which foods they can and cannot

[18] Feillet, F., MacDonald, A., Hartung (Perron), D., Burton, B., *Outcomes beyond phenylalanine:* An international perspective Molecular Genetics and Metabolism : 99 (2010) S79-S85
[19] Waisbren, S. THE PSYCHOLOGY OF PKU and ALLIED DISORDERS (AND THE BOSTON MARATHON) National PKU Alliance Annual Parent Meeting, November 2009 Presentation

eat, but they may be able to tell the difference between familiar and unfamiliar foods. From an early age, you can teach your child to ask you before eating unfamiliar foods, or to say no when someone other than a trusted caregiver offers food[20]. You may want to point out that it is important not to accept food from anyone but trusted caregivers for all children, not just those with PKU.

It is also important for older siblings to become familiar with the PKU diet as well. You can involve your older children in meal preparation and encourage them to help feed a younger sibling with PKU so that they learn about foods that are part of the PKU diet[21]. When older siblings understand that a special diet is necessary for their sibling's health, their behavior and positive attitudes can help a child with PKU accept his or her diet and treatment.

Treatment and Diet

A Team Approach

Experience shows that children from families that seek medical help, visit the clinic regularly, and send in blood samples on a regular basis have the best outcomes with PKU. You will gain the tools and knowledge you need to manage your child's PKU through clinic visits and support from your PKU clinic team. Partnering with the PKU team will help you learn to manage your child's PKU and allow for the best possible care for your child. Even though it can be difficult to accept guidance on something that seems basic, such as feeding your baby, feeding a child with PKU is more complicated than feeding a child that does not have PKU. The PKU team can provide you with detailed instructions on how to oversee what can seem to be a complicated diet at first. Following their guidance is crucial and will empower you with the ability to provide an appropriate diet for your child to protect your child's intellectual development and nutritional health. For more information, please see Chapter 2 for the roles of all members of your PKU team.

Breastfeeding/Bottle feeding

Even though your child has PKU, he or she still needs to consume some Phe. The amount of Phe prescribed in the diet is determined by your child's Phe levels. Once your child's Phe levels have come down to a safe level, either breast milk or standard infant formula will be the source of Phe in your child's diet.

A Parent's Perspective

"I was so glad to hear that I could still breastfeed my daughter. We soon got into a regular routine that included breastfeeding and bottle feeding with the medical formula. The bottle feeding times allowed my husband to feel more included in the care of our new baby girl."

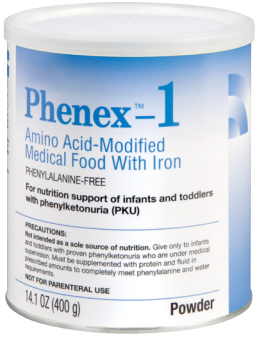
You can choose to use breast milk as the Phe source and still keep your child's Phe at a safe level. Breast milk contains much less Phe than infant formula. However, breast milk alone contains too much Phe to be the exclusive food for babies with

[20] Utah Department of Health. About PKU. Diet Management. Available at: http://health.utah.gov/newbornscreening/Disorders/English/MSMS/AA/PKU/Manual07DietManagement_PKU_En.pdf Accessed May 17, 2011
[21] Utah Department of Health. About PKU. Diet Management. Available at: http://health.utah.gov/newbornscreening/Disorders/English/MSMS/AA/PKU/Manual07DietManagement_PKU_En.pdf Accessed May 17, 2011

Diagnosis to 2 Years

PKU so it must be combined with Phe-free PKU formula. You can work with your dietitian to decide whether you will pump and measure the breast milk or put the baby to the breast. If you choose not to use breast milk, standard infant formula is the appropriate substitute to provide Phe in the baby's diet.

At this age, your child will need you to manage his or her diet, as do all infants.. From infancy to two years of age, parents are generally able to maintain their child's PKU diet without much resistance. Your PKU team will prescribe the right medical formula that, in combination with the breast milk or infant formula, provides the proper nutrition for your child. The amount of medical formula and breast milk or standard i nfant formula will need to be adjusted from time to time to provide the right amount of Phe to meet your child's needs and keep blood levels in the safe range. Keeping a diet record of the amount of formula and breast milk your child consumes will be helpful to accurately monitor his or her Phe intake. Your child's dietitian and PKU team will w ork with you to determine how much breast milk or infant formula and medical formula you should provide.



Mixing Medical Formula

Medical formula, breast milk, or infant formula are the only foods your child needs until around six months of age – any extras, even water, can make your child less interested in the formulas or breast milk that he or she needs. This is especially important for a child with PKU, as it is critical that he or she drinks the prescribed amount of medical formula and standard infant formula or breast milk each day to maintain safe Phe levels.

Will my baby still need medical formula once we start solids?
The medical formula will always be needed. The recipe, amount and type of medical formula will change as your child gets older. Your PKU team will help you figure out when these changes need to be made.

One of the most important ways for you to make sure that your child is getting the nutrition necessary is to measure the medical formula accurately using a gram scale or proper measuring utensils (see Chapter 2 for more information on how to measure medical formula).

Diagnosis to 2 Years

Tips for preparing medical formula:

- Wash and dry your hands thoroughly before handling bottles and nipples and before feeding your child.
- Sterilize bottles and nipples for every use until at least six months of age.
- Measure the medical formula, then add the appropriate amount of water, and mix together, following the instructions you receive from your PKU team.
- Place mixed medical formula that your baby will not be drinking right away in the coldest part of the refrigerator, usually at the back, as soon as it is made.
- When going out, take refrigerated bottles of medical formula in an insulated bag or cooler with an ice pack to keep them cold.
- It is important to let the team know if your baby is sick or not feeding well so his or her formula and diet can be changed if necessary.
- Talk to your PKU team if you have any concerns about feeding your child or feel you need help.

Transitioning to a Sippy Cup

Sippy cups can be introduced at around 6 months of age. To make the transition to a sippy cup easier, start with a small amount of medical formula in a sippy cup and offer lots of praise when it is finished. Gradually increase the amount of medical formula you serve in a sippy cup. You can also let your child pick the cup to drink from.

Transition in Formula

At about one year of age, your child's medical formula will be adjusted so that it continues to meet his or her nutritional needs. The transition will occur in a stepwise fashion to ensure your child's acceptance of the formula. Your dietitian will work closely with you during this time of formula transition.

Starting Solids

A child is ready to start solid foods when he or she is able to sit with support and hold his or her head upright and steady. At this time, a child may start showing signs of interest in what caregivers are eating. This usually happens around six months of age. As your child eats more solid food, it is still important to make sure that your child drinks the prescribed amount of medical formula each day.

So how do you transition to new foods? All parents have to introduce new foods to their child. For children with PKU, there may be a few extra difficulties, but some practical advice all parents can use will help!

- Keep it as simple as possible. If your baby loves pureed carrots, then start there. Boil up some carrots, rough mash them with a fork and add a LITTLE bit of the new food into the smooth puree. This way you are not changing taste, just texture. So if they like mashed potatoes, puree a TINY amount of broccoli and mix it in. Brown rice is also a great way to add some lumps.
- Cook the new food, blend it in a food processor at first and add a LITTLE bit to a food your baby loves. GRADUALLY add more and more. Your baby may gag a little at first (they are born with an oversensitive gag to protect them) and they have amazing straining skills- how they can discern that little lump and separate it from the rest is really quite remarkable when you think about it.
- Almost all kids reject new foods. Keep introducing it and pairing it with their favorites.
- If they are older, encourage them to taste new foods by being a good role model so they can watch you eat it, acting like an idiot (I am a big giant and I am going to eat this little tree, making faces on the plate with the food -- argh please don't eat my nose, etc.), playing into their latest obsession (princesses love nuggets when they are shaped like a heart, pink princess sauce)."

Diagnosis to 2 Years

Once your child starts solid foods, you will need to count the Phe intake from food at each meal. Your dietitian will provide a prescribed amount of Phe to introduce into your child's diet. Using a food reference guide, you can measure foods and the amount of Phe your child eats. See Chapter 2 for more information on counting Phe and the resource section for information on how to get your Food Reference Guide.

Keeping a detailed diet record of the medical formula and solid foods your child consumes will be helpful to accurately monitor their intake. Your dietitian will explain how to fill in a diet record, which you may need to bring to clinic visits or send with blood samples (see "Filling Out a Diet Record" in Chapter 2).

Introducing solid foods to any child generally takes several months as children get used to new tastes and textures. The chart below will give you some ideas of when babies may be ready for new types of foods.

	AROUND 6 MONTHS OR WHEN BABY CAN SIT AND HOLD HEAD UP	8-9 MONTHS	10-12 MONTHS
TYPE OF FOOD	Cooked mashed or strained foods	Soft, diced foods	Self-feeding and finger foods
EXAMPLES OF FOODS FOR THIS SATGE	• Infant rice cereal • Commercially prepared baby food or home-prepared pureed food from fruits and vegetables low in Phe • Pureed vegetables, such as carrots, squash and beets • Pureed fruits such as peaches, pears, apples and apricots	• Mashed, grated, diced, more thickly pureed foods of increased variety • Diced or mashed cooked vegetables • Soft fruit • Vegetable soups • Low protein pasta and rice mixed with pureed	• Toast made from low protein bread • Peeled soft fruits cut in bite-sized chunks • Strips or pieces of cooked vegetables • Low protein pasta spirals • Low protein crackers • Gerber fruit puffs • Low protein dry

To ensure that your child stays within the prescribed Phe limit for the day, you may find it helpful to use the following strategies:

- Decide how much Phe/exchanges your child will eat at each meal or snack.
- Spread the amount of Phe/exchanges your child eats throughout the day, rather than eating most of them at one meal.
- Choose the foods you will offer ahead of time so you can count up each day's planned Phe intake.
- Measure/weigh the amount of food your child may have to provide the amount of Phe/exchanges available for that meal.
- Make note of how much food is left. If your child doesn't finish the meal, subtract the uneaten Phe from the total Phe/exchanges you planned for that meal.
- Offer medical formula throughout the day. If your child finishes the formula and is still thirsty, you can offer water or juice.

Diagnosis to 2 Years

A Parent's Perspective

"We always make sure that everyone in our family has a fruit or vegetable serving that is low in PHE during dinnertime. Then, we just substitute an additional low Phe item for our children with PKU while my wife and I eat something higher in protein. This makes all of our dinner plates have at least something in common."

"Keep trying. I have had numerous cooking disasters where the food looked and tasted terrible. However, I knew that I had to be persistent. I needed to prove to myself that I could take care of my child and give her a wide variety of healthy foods. Eventually I got the hang of it. I still struggle with new foods/recipes because not everything cooks the same way as what I make for the other family members. Being flexible and having a sense of humor made it work, even through the burnt foods!"

Transitioning to Table Food

By one year of age, your child is probably eating fruits and vegetables, and some low protein grains, cereals, bread, pasta and crackers. Your child may show interest in food your family is eating and is typically ready to move away from baby foods. Including your child in family mealtimes from an early age will help with the transition to eating meals with the family.

What if my child is still hungry at the end of the meal?
If your child is still hungry at the end of a meal, offer her something that is very low in Phe or has no Phe. Commonly used low-Phe foods can include: applesauce, low protein rice with butter, Hunt's or Kool Aid jello packs, or low protein bread. Ask your dietitian for more ideas!

To encourage the transition to table food for both children and parents, it may be helpful to begin to use low protein recipes to prepare meals for your child (refer to Chapter 15 for a list of low protein cookbooks). You will need to continue to keep a record of Phe/exchanges – diet records may need to be sent in with blood tests and brought to clinic visits.

How to Make Mealtime Easier

Refusing food is common during the early childhood years, whether or not a child is following a special diet. When parents can understand why children may act as they do, it can help them work with their child to make mealtime easier.

Tips for Making Mealtime Easier

Keep a positive attitude toward your child's diet.
It is important for family and friends to have a positive attitude towards your child's diet. Allow your child to form their own opinions about their diet without negative input from yourself or others.

Diagnosis to 2 Years

Offer choices to satisfy a child's growing independence.
As toddlers start discovering that they are independent people, they may express their likes and dislikes more strongly. It may help to allow your child to choose between two foods, or to be involved in small (and safe) aspects of food preparation. Also, toddlers may want to feed themselves; let your child use a spoon, or offer several finger-food options to encourage self-feeding.

A Parent's Perspective
Be careful about introducing high Phe foods to meet the daily Phe allowance. As your child gets older, he will want larger portions. So it may seem okay to give him a high Phe food when he's 1 and only wants a few bites, but he's going to want a lot more of that same food when he's 3. And you're going to have to tell him he can't have something he's developed a taste for.

Respect that your child may not be hungry.
After the age of 12 months, children don't grow as quickly, which means their appetite may not be as large. Your child will let you know when he or she has eaten enough. This is the time to stop feeding, even if your child hasn't finished the meal. Children's appetites may vary from day to day, and children will eat when they are hungry.

Keep regular routines.
Children respond well to having predictable routines They need to eat regularly to meet the demands of their growing bodies. Seat your child at the table for meals. Children have short attention spans; set aside 20-30 minutes for meals and 10-15 minutes for snacks.

Blood Phe Monitoring
Goal Phe Range: 2-6 mg/dl or 120-360 µmol/L

At this age, Phe levels are usually measured using a small blood sample taken from the heel or big toe of babies and toddlers and from the fingertip as children grow older. Parents are taught how to collect samples from babies and young children at their clinic visits. This usually happens in the first few weeks. Grandparents or other trusted caregivers may also be taught how to take a sample.

The procedure is easy to manage once you have had a little practice. It is important to communicate to your child through your actions and words that blood samples are non-negotiable. Helping your child learn the importance of blood samples will help your child maintain good control of his or her PKU over time.

Your PKU team will usually supply you with special filter papers needed for the test and tell you where to send the cards when you have collected a sample so that they can be tested. Occasionally, a blood sample may need to be drawn from an arm vein.

Tips for Taking a Blood Sample at this Age
- Take every blood sample in the same setting and at the same time of day.
- Help your child know what to expect; providing guidance may reduce fear of the unknown.
- Play in a warm room or bathe your child's heel in warm water to get blood flowing.
- Keep the atmosphere light and matter of fact. Sing a song, or use a pacifier or favorite comfort object to soothe your child.
- Purchase colorful or character bandages, and let your child pick out which one he or she wants.

Diagnosis to 2 Years

Frequency

During the first few weeks of treatment, while your child's diet is getting settled, blood tests may be needed one to two times per week. Once a child has a consistent feeding routine, this may be cut back to once per week, but as the first 12 months are a time of quick growth and change in diet, it may be necessary to test more frequently.

After the first 12 months, blood test frequency may decrease. Typically samples are recommended every two weeks for children over one year of age. Extra blood samples may be needed during or after an illness to figure out whether Phe levels are too high or too low.

Special Considerations for Diagnosis to 2 Years

A Parent's Perspective

"...[Our son's daycare provider] has now created a "yes food" and "no food" book with Kaleb. They cut out pictures of foods from magazines and grocery ads and Kaleb pastes them on either the "yes" or "no" side of the divider. To my husband Paul and me, Connie is a miracle and could never be replaced!" —Mother of PKU Patient

Starting Daycare

Some parents may wonder if their child can still go to the daycare facility they chose prior to their child's PKU diagnosis. Most daycare facilities are willing and capable of managing your child's PKU diet and treatment.

Communication and planning are the most important aspects of ensuring that your daycare provider can manage your child's PKU. To help your daycare provider and ensure your child's safety, the following tips may be helpful:

- *Explain PKU.* It is important to be open with your daycare providers so that they know your child's needs. (See "Explaining PKU" at the beginning of this chapter.)
- *Use lists.* Provide your child's daycare facility with simple and easy to read lists of what your child can and cannot eat or drink.
- *Offer to help communicate to staff.* It may help your daycare provider if you come to the center to talk to staff about PKU. This can help make your child's needs known by all who may come in contact with him or her.
- *Be "on call."* Tell your daycare providers that when in doubt about anything to do with your child's care, they should call you.

"All the children in my care know about PKU. We talk about it openly and honestly in terms they can understand, although we do not dwell on the subject. It is not uncommon for one of the other kids to say they wish they had Kaleb's "special diet." In fact, my two oldest girls (ages 4 and 5) sometimes double check with me to make sure Kaleb has the right cookie or the correct glass of "milk". - *Childcare provider for a PKU patient*

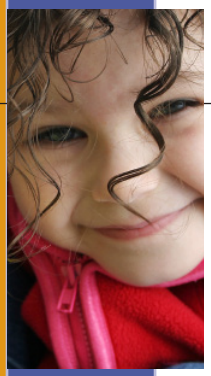
Feeding your child at daycare

Many parents of infants that do not have PKU prepare a one-day supply of bottles with breast milk or infant formula for daycare. Even though your infant is drinking medical formula, you can prepare a one-day supply of formula for your child and bring it to daycare each morning. Recording how much is consumed by an infant in a daycare is not unusual, even when the child does not have PKU. You will need

to make sure that your provider understands the importance of recording how much was consumed at each feeding more precisely – or saves the bottles – so that you can determine if your child will need more medical formula at home. If your child will be eating snacks and meals that you prepare at home, ask your daycare providers to monitor your child's eating and to record the amount of food your child ate that day so that you can determine how much Phe was consumed. You may ask the daycare provider to send home the uneaten food so you can measure it yourself to determine how much Phe is left for the day.

If your daycare facility provides meals or snacks, you can work with your daycare provider to feed your child according to her PKU diet. You may wish to:

- Request that only one provider be responsible for your child's meal preparation. This will help ensure that one person gains necessary experience with the PKU diet. This setup will also allow easier communication about your child's needs as they change.
- Ask to receive the daycare menu each month in advance. Posting the menu in advance is not unusual, and will allow you to choose foods from the menu that your child can eat and decide if you need to supplement the menu with any foods from home.
- Provide a scale to your child's daycare and measuring cups, if necessary, so that food can be weighed and measured.



Chapter 5: Ages 3 to 6 Years

A Parent's Perspective

"...Zachary and Rachel...are fine. You would never know they have PKU unless you saw us weighing the French fries or heard us request a happy meal with salad (no cheese, please) instead of a hamburger." - Parent of two children with PKU

What to Expect

Parenting can be stressful for anyone, but parenting a child with a chronic disorder such as phenylketonuria (PKU) can be especially stressful and may negatively impact the parent's "mental health-related quality of life"[22]. Decreased emotional support – such as what you would get from supportive friends and family – has been reported to decrease a parent's sense of mental health-related quality of life. However, parents also report that as their child with PKU gets older and they feel development has stabilized, there is an increased sense of mental health-related quality of life. Still, there will be times when you will feel stress, such as when you're getting ready for preschool and kindergarten. This is a time of anticipation and, if it is the first time your child will spend long periods of time out of your care, some concern.

A Parent's Quick Tip

Try to network with other PKU families in your region. Even if you have to travel a few hours to get together, it's worth it to socialize with these families. You and your child will both benefit from the interaction.

Your health and sense of well being is important, and may relate to how you parent your child or children. It is important that you keep aware of the emotional support you are receiving, and if you find yourself feeling in need of additional support, vocalize it. Seek support from your friends, family and other parents of children with PKU. Your PKU team can also help with support groups, individual counseling and even match you up with other parents of children with PKU, if you haven't already been introduced[1]. Please see Chapter 15 for website information on accessing state and regional support groups.

Development

While most children begin to develop a strong desire for independence at about three years of age, they still have very little self-control[23]. In order to maintain proper diet and treatment, expectations for a child's ability to manage PKU need to remain age-appropriate. You play a key role in delivering much of the day-to-day care for managing your child's PKU. However, involving your child in management of his or her PKU will help to empower your child with the ability to make decisions about treatment as he or she gets older and can manage treatment independently. This will hopefully improve long-term adherence to the PKU diet[24].

This is a critical age, as children who maintain proper metabolic control at this age have much better health and developmental outcomes than those who do not. From four to six

[22] ten Hoedt AE, Maurice-Stam H, Boelen CAC, Rubio-Gozalbo ME, van Spronsen FJ, Wijburg FA, Bosch AM, Grootenhuis MA. *Parenting a child with phenylketonuria or galactosemia: implications for health-related quality of life* J Inherit Metab Dis 2011:391-8

[23] Waisbren, S. THE PSYCHOLOGY OF PKU and ALLIED DISORDERS (AND THE BOSTON MARATHON) National PKU Alliance Annual Parent Meeting, November 2009 Presentation

[24] van Spronsen, F.J., de Groot, M.J., Hoeksma, M., Reijngoud, D.J., van Rijn, M., J Inherit Metab Dis (2010) 33:671–676

Ages 3 to 6 Years

years, children have little issue following the PKU diet, as rules around foods and medical formula are clear and concrete. Socially, children with PKU develop normally, although some may experience separation anxiety[25]. Developmental progress is typically normal and children with well-controlled PKU can be expected to function normally in a kindergarten setting.

Talk to your PKU team if you have any questions about your child's development.

With your help, a child of this age usually:

- Is aware that he or she has a special diet
- Begins to learn how to deal with other children's curiosity about PKU
- Knows that he or she needs to take medical formula
- Begins to watch you prepare the formula and by 6, can assist in formula preparation
- Knows that he or she needs to have blood tests
- Knows to check new foods with parents
- Begins to learn "yes/no", "red, yellow, green traffic light" or "low PHE/high PHE" foods
- Is aware that the PHE in food is counted

Talking with Your Child About PKU

While your child is not old enough to manage PKU alone, it is valuable for your child to begin to better understand his or her diet and treatment. When speaking with your child about PKU, the following tips may be helpful:

- *Use simple examples to explain ideas.* For example, when explaining to your child why the PKU diet is important, it may be helpful to relate the diet to that of a food allergy. See the resources in Chapter 15 or speak with your PKU team who will also have information about books to read to your child to help him or her understand special diets.

- *Let your child know she can say "no".* If you haven't already, teach your child to ask you before eating unfamiliar foods, and that it is OK to say no to anyone who offers food that is unfamiliar or off limits.

- *Tell your child it isn't his or her fault.* A child may not understand why he or she has PKU when others do not, and may think that he or she did something to 'deserve' it. Explain to your child that everyone is born with different qualities – such as hair color and eye color – and PKU is something that people are born with, not something that anyone causes. The book Everybody Has Something may help show your child that everyone is different with their own unique challenges. It can be ordered at http://www.ucdenver.edu/academics/colleges/medicalschool/departments/pediatrics/subs/genetics/clinical/IMDNutrition/Documents/Everybody_Has_Something_OrderForm.pdf.

How do I talk to my child about medical formula?
Treat the medical formula as special – as an 'energy drink' that will make your child 'tall and strong'. Give the medical formula a name, such as 'special milk'. Give lots of positive prompts and praise – but let your child know that drinking the medical formula is something she or he must do.

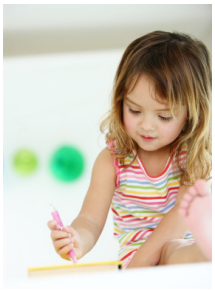
[25] Waisbren, S. THE PSYCHOLOGY OF PKU and ALLIED DISORDERS (AND THE BOSTON MARATHON) National PKU Alliance Annual Parent Meeting, November 2009 Presentation

Ages 3 to 6 Years

> ***A Parent's Quick Tip***
> "Don't approach the diet as a burden (though some days it is, I know) -- think of it as a special part of the day where you get to teach your child about something very important and build trust in you that you are always there to help!"

- *Stay positive*. Sending the right message about foods and treatment is important. It is better to talk about off-limit foods as "high Phe," "no," "red" or "stop" foods (see the next section on the Food Traffic Light), rather than "bad" or "naughty" foods[26]. In the same way, avoid talking about blood sampling in negative ways, such as calling yourself or another parent "mean" for taking a blood sample. Diet and blood sampling are part of life and staying positive will help your child accept and manage PKU as he or she grows. Never say anything negative about the food or formula to your child.

- *Begin to include your child in preparing food.* Empower your child from an early age by including him or her in preparing meals. It may start with something as minor as mixing the formula at 3 and identifying which food should be included in a meal by 6 years.



- *Let your child measure and weigh their food.* Children are naturally curious. Show your child what you're doing when you're weighing and measuring the food you are preparing and help her or him use these tools. Explain how it works and slowly, see if he or she can weigh or measure the food for you, with your assistance.

- *Get your child to help count Phe.* As your child begins to learn to count, he or she can begin to help with recording his or her Phe intake. You can make it into a counting game, and even a memory game about what he or she has eaten. You can use a whiteboard or laminated paper to create a chart to count the Phe eaten at each meal. Write the total amount of milligrams/exchanges your child can have each day at the top of the chart, and list meals under this amount. Next to each meal, write the milligrams/exchanges planned for that meal. As the milligrams/exchanges are eaten during the day, your child can cross them off.

Treatment and Diet

Possible Challenges with the PKU treatment

> **A Parent's Perspective**
> *"PKU has really worked into our life and then it just became a part of it all in a way where I (as a mom and the meal planner/preparer of our house) almost don't even "think" about it anymore! It gets better, it really does!"*

Children at this age are developing a growing sense of independence and sometimes try to gain control over their lives by challenging rules and limits their parents set. For children with PKU, this may include objecting to their diet or treatment.

By allowing your child to have a role in some of the decision making process, he or she will feel like they have more control of their PKU. Above all, it is important to stay positive and matter-of-fact about your child's diet and treatment.

[26] Kids Health Talking to Your Child About Diabetes. Available at: http://kidshealth.org/parent/diabetes_center/living_diabetes/talking_diabetes.html# Accessed May 17, 2011

Ages 3 to 6 Years

KINDS OF CHOICES YOU CAN OFFER
"Do you want to sit on the chair or on my lap to take your sample?" or "Which finger should we use for your sample?"
"Would you like carrots or apple for your snack?"
"Do you want to drink your special milk in the blue cup or red cup?"

The 3 Kinds of Food: Traffic Lights

Many parents use the Traffic Lights example to teach children about the PKU diet. To create a Traffic Light, draw three circles resembling traffic lights on a large poster board or piece of paper and color them green, yellow, and red. Then cut out pictures of many different types of foods from magazines or websites.

To start the game, explain to your child that there are three kinds of foods: "red", "yellow", and "green". These colors are defined as:

- Green = foods that are low in protein/Phe
- Yellow = foods that are only OK in limited quantities
- Red = foods that are high in protein and not on the PKU diet

Work with your child to organize foods according to each color. Once your child understands these ideas, you can create Traffic Light games to improve or test knowledge. New food pictures can create a fun challenge for your child as he or she figures out where each new food belongs on the traffic light, or you can test your child's recognition by placing a "red" food on the green light, or vice versa, and asking which of the foods does not belong.

The Traffic Light may also help you talk about diet choices with your child. Children familiar with the Traffic Light will readily understand what a "green" food or "red" food is, and this offers a way for parents to say no to foods without using the word "no" constantly. Some parents may also choose to refer to foods as "low Phe" and "high Phe" foods for this same reason.

Medical Formula

Medical formula is important for growth and development and keeping blood Phe in the safe range. Your child should drink all the medical formula prescribed each day. Talk to your PKU team if your child does not drink the prescribed amount of medical formula. Some trouble-shooting tips for formula drinking are below.

If…	YOU CAN TRY…
Your child seems overwhelmed by the amount of formula she needs to drink at each siting.	Dividing the amount of medical formula your child needs to drink into three to four smaller servings that will be easier to finish.
Your child does not want to drink the formula during a meal.	Offering medical formula at the beginning of mealtime when your child is hungry.

Ages 3 to 6 Years

Your child does not have an appetite for the medical formula.	Cutting back on other liquids or snacks and offering formula first when your child is hungry or thirsty.
Your child usually likes the medical formula but seems bored with it.	Freezing the formula, then blending it and serving with a spoon for eating. Or serve it in a new cup he or she is excited to use.
Your child doesn't seem to like the taste of the medical formula.	Talking to your PKU team about how to flavor the formula or trying other brands or flavors of formula.

Tips for making mealtime easier

Try to keep family meals and PKU meals similar.
A child with PKU may find it difficult to understand why she can't eat the same foods as others; as a result, she may refuse to eat her food. If possible, make your child's meals similar to the family meal. For example, if the family is having beef stir-fry and rice, serve your child stir-fried vegetables with low protein rice. Your child may want to eat foods that are not appropriate for her PKU diet; educate your child about which foods she can have and support positive food choices.

A Parent's Quick Tip
Order pasta from more than one company and offer your child different brands regularly. Sometimes a certain brand will be on back order when you need pasta, so you want your child to have a taste for more than one brand.

Create a pleasant mealtime environment.
Parents and siblings can be good role models for young children. Eat meals together as often as possible. Try to limit distractions such as TV during mealtime. A relaxed atmosphere during mealtimes can help make this time enjoyable for the family.

Avoid battles over meals.
Parents often become anxious when their children don't eat, and children quickly pick up on this. It can happen especially when you have made a lot of effort to prepare meals for your child with PKU. Some children refuse to eat, knowing it is an effective way to gain attention. It is never a good idea to force-feed a child. This often leads to fear of mealtimes and further refusal of food. Continue to offer new foods multiple times; children may need to see a new food many times before they will try it. Praise your child for trying new foods.

Educate your child about which foods he or she can have.
Your child may want to eat foods that are not appropriate for her PKU diet; educate your child about which foods he or she can have and support positive food choices. Another way to set a good example is to encourage family members to eat plenty of fruits and vegetables.

Ages 3 to 6 Years

Blood Phe Monitoring
Goal Phe Range: 2-6 mg/dl (12-360 umol/L)

Frequency
At this age you should check your child's Phe level every 2 weeks, unless directed otherwise by your clinic.

Tips for Taking a Blood Sample at this Age

Preschoolers and early school-age children are old enough to become more involved in taking blood samples. For example, you can let your child choose where he or she wants to sit when a blood sample is being taken or to choose which finger will be used to take the sample. As with earlier ages, following a routine for blood sample taking can help children know what to expect, which can reduce fear around blood sampling. Other tips for taking blood samples at this age:

A Parent's Perspective
"[My daughter] has made up a little dance she does to shake the blood down and she gets all the supplies ready herself. I wouldn't say this is something she necessarily likes but she accepts it and is a part of it and on we go!"

- Have a calendar with the date marked when blood samples are taken
- Let your child choose which finger to use for the blood sample.
- Encourage your child to help you press the button on the lancet or diabetic pen. Your child can put a finger over your finger and press on the count of three.
- Ask your child to help you count out the blood drops onto the filter paper.
- Ask the child to help "finger paint" the circle(s).
- Explain what a correct sample looks like, and encourage your child to see how well he or she has done in producing a correct sample.
- Give lots of positive feedback on how well your child has done.
- When your child is old enough to write his or her name, have your child help you fill it out on the filter paper.

A Quick Tip
Topical "numbing agents" such as Pain Ease® may help reduce the pain of the finger stick. Ask your PKU team if this is an option to use.

Overall, try to maintain a consistent schedule and matter-of-fact manner about blood sample testing. Blood tests are a way of life for children with PKU, and the patterns you set when your child is young will help create the foundation for how he or she manages PKU throughout life.

Special Considerations

Explaining PKU to Friends

Your child's friends can be a source of support. Although his or her friends cannot tell that your child has PKU at first glance, they may notice his or her special diet and ask questions. Helping your child find ways to explain PKU to friends can help him or her adjust to new situations and help build his or her acceptance for this condition. Your child can share simple information or give a more detailed explanation, but being direct and honest may help other children clearly understand PKU. The book Everybody Has Something may also help

Ages 3 to 6 Years

show your child's friends that everyone is different with their own unique challenges. It can be ordered at http://www.ucdenver.edu/academics/colleges/medicalschool/departments/pediatrics/subs/genetics/clinical/IMDNutrition/Documents/Everybody_Has_Something_Order_Form.pdf

What should I do if my child doesn't want to tell friends?
It is important that friends know about your child's PKU so that they understand or can help offer support in social situations. If your child doesn't want to tell friends herself, you can discuss with her the idea of inviting friends over so that you can help explain PKU. Some parents also provide a presentation to their child's class, offering classmates the chance to ask questions and see the "tools" of PKU treatment, such as formula and blood sampling material.

Listed below are some helpful explanations of PKU:

- No one can "catch" PKU. People are born with PKU, like I was.
- Having PKU means I have to eat foods that keep me healthy.
- I can't eat some kinds of foods, like meat or cheese, or they will make me sick. I won't get sick right away, but if I eat these foods, my body and brain won't grow the way they are supposed to.
- I drink a special milk which is like a vitamin drink. It gives my body good things that come from foods I can't eat.

Offer encouragement to your child by:

- Giving positive messages about the foods he or she can have
- Reinforcing to your child that he or she is special, and that this special way of eating is to keep him or her stay healthy.
- Talking to your child about other people you know who are on a special diet, even if they are adults, so your child knows that he or she is not the only one on a special diet.
- Discouraging negative comments about the taste of the medical formula and the low protein foods from any family members.

See the resources in Chapter 15 for examples of questions your child may be asked and how he or she can respond.

Chapter 6: Ages 7 to 12 Years

A Parent's Perspective

"I have to go to the hospital every 6 months. I get really excited because I get to miss one whole day of school! I go all the way to Toronto to visit my dietitian... She suggests new foods and makes adjustments to my formula. She also tells me how my protein levels have been. After I visit her, I usually go to Doctor Feigenbaum [sic]. She tests my abilities and sees how much [PHE] concentration I have. And then comes the worst part of going to Children's Hospital. I have to get a needle...to see how much protein I've had. I also have a pen at my house that I use to prick my thumb every month and then I send a sample of my blood to the hospital, and they send me my protein levels. I think going to Toronto is one of the best parts of having PKU."

What to Expect

At this age, the focus of a child's life expands beyond family life to include relationships with peers, teachers, coaches and others. This is an exciting time for you and your child as their "world" begins to expand. Having provided them (and those around them) with all the information you have about PKU and their special low protein diet, you are likely feeling quite confident about your child with PKU and his or her ability to go out in the world as independently as they can at this age!

As your child begins to experience his or her expanding world, he or she will encounter new and enticing foods. Some parents may begin to feel a sense of guilt around their child's low Phe diet and not being able to allow your child to have these exciting foods. Like any parent, you want to be able to give your child a small treat. It's important you don't give in to this sense of guilt by giving treats that aren't low in Phe. Don't forget, there are treats that your child can enjoy without compromising their diet. By ensuring your child doesn't over-indulge in these treats, they will remain just that – fun treats you can give your child when he or she needs a little celebration, pick-me-up or change in their regular diet! Enjoy it with him or her and the entire family!

Development

Children of this age spend more time with their friends and often rely on peers and others outside their family for information. During this time, social connections for children with PKU are critical[27] as children with PKU often report feeling socially isolated[28]. Encouraging social connections now will help your child develop a strong social network for the teen years, when social support becomes even more important for their well being and development.

During this time period, the academic demands on children increase. Schoolwork shifts from a focus on memorization to using knowledge for problem solving. Children of this age develop the ability to apply logic to solving concrete problems; parents can help encourage

[27] Advocates for Youth. "Growth and Development, Ages Six to Eight—What Parents Need to Know" Available at: http://www.advocatesforyouth.org/index.php?option=com_content&task=view&id=154&Itemid=206 Accessed May 17, 2011

[28] Jusiene R, Cimbalistiene L, Bieliauskaite R. *Psychological adjustment of children with phenylketonuria Medicina* (Kaunasa) 38 (2002) 424-430

Ages 7 to 12 Years

this skill by asking questions to prompt problem solving, such as[29]:

- "What are you being asked to do?"
- "What do you know?"
- "What information will help you?"
- "Is your approach working? Should you try something else?"

It is not uncommon for children of this age to resist some aspects of their treatment[30]. For this reason, it is especially important for parents to continue encouraging your child's involvement with treatment, such as making formula and preparing their filter cards. Talking to your child about how he or she will handle the PKU diet and treatment at school is also very important at this age. While the school staff will do their best to supervise, you will feel more secure if your child understands and is able to help manage her own diet appropriately.

Things to talk to your child about include:

- Foods that are OK and foods to avoid.
- Bringing home uneaten food in his or her lunch box so that you can calculate milligrams/exchanges for the day.
- Not swapping lunch food or drinks with friends.
- Deciding when to drink medical formula during the school day and how to do it.
- How to explain PKU and their diet and formula to other kids.
- What to do about teasing or embarrassment about the diet.

Children seven to ten years of age should work with parents on:

- Preparing formula with decreasing supervision.
- Choosing an after school snack.
- Learning to pack school lunch.
- Beginning to list foods on a diet record.
- Beginning to weigh and or measure foods regularly.

Children ten to twelve years of age should work with parents on:

- Beginning to prepare and consume formula independently each day (with parental monitoring).
- Preparing simple entrees independently.
- Knowing what blood levels are ideal.

-University of Washington. PKU and Self Management Timeline Available at: http://depts.washington.edu/transmet/process/timeline.pdf Accessed May 18, 2011

In addition, you should continue the process of helping your child learn how to manage PKU. Between seven and twelve years of age, your child will begin to manage – with your help – some aspects of her PKU. Helping your child begin to take responsibility for PKU now will help your child develop the competence and confidence to manage PKU throughout his or her life.

Treatment and Diet

Adherence to Diet

As children grow, they become more aware of those around them and notice people's differences. It is normal for children of this age to place increasing value on peer acceptance and to want to be like their peers. Children with PKU may feel tempted to eat foods that are not on the PKU diet because they want to be like their friends or not call attention to

[29] Waisbren. S. Neurocognitive Functioning in PKU Illinois 2010 Presentation
[30] Waisbren, S. THE PSYCHOLOGY OF PKU and ALLIED DISORDERS (AND THE BOSTON MARATHON) National PKU Alliance Annual Parent Meeting, November 2009 Presentation

Ages 7 to 12 Years

themselves. In addition, children with PKU don't feel different or unhealthy, so they may question the need to stick to their diet or treatment as they don't "see" the effects of their PKU.

A Parent's Perspective
"I used to tell my daughter, some people need glasses, some people are short, others are tall and you have PKU. You have to accept it and make it part of your life but having PKU is not your whole life."

It is important for parents to reinforce to their child that he or she is healthy because of following the treatment plan. By sticking to the diet, formula and treatment, he or she will grow and be able to do the things he or she wants to do. Talking about your child's goals – whether it is playing soccer in the spring or finishing a project for a class – may help your child see how diet and treatment is essential to achieving those goals.

A Parent's Perspective
"I pre-measure cereal, crackers, cookies, grapes, whatever, so that my child can make independent decisions about snacks."

Some suggestions for ways to help your child adhere to diet include:

- Exploring new formula options if formula in take becomes an issue.
- Having them more involved in the process of formula preparation. i.e. becoming more comfortable with using a gram scale and/or measuring utensils with parental supervision.
- Having your child bring formula to school in colored sports bottles or pre-made drink packs if available. This may help minimize interest or questions from other kids.
- Involving your child in meal planning and preparation. If she has some control over difficult aspects of her treatment, it becomes a shared responsibility and she will be more likely to stick to it.
- Letting your child choose her snacks or help you pack her lunch so that she has some control over what she is eating during the day.
- Keeping PKU meals similar to family meals so that your child feels a part of and included in the foods her family eats.
- Keeping low protein treats at school with your child's teachers so that your child has something to eat while others are celebrating a birthday or other event.
- Making sure that the child is comfortable asking for their low protein foods so that they do not get into the habit of sneaking foods.
- Including the child in Phe tracking/counting throughout the day and having them write foods in their diet records.
- Making sure that the child is aware of the importance of keeping their Phe intake within goal.
- Role playing with your child, asking her questions that other students might ask, such as "Why do you drink that milk?" and helping her work out responses she can use in different situations.
- At this age, children tend to be very active especially when involved with after school sports. That is why it is important to have a variety of low protein snacks available.

Ages 7 to 12 Years

A Parent's Perspective

"We started this process very young. My favorite idea was from our 3-year-old preschool teacher. She actually helped us make it. We made a tree out of brown construction paper and mounted it on a poster board. We then laminated the poster board. From there, we cut different colored leaves out of foam sheets (found in the craft store). On the back of each leaf, I put a circle velcro and the matching velcro was attached to the posterboard. Each colored leaf was worth a different number (e.g., 1, 5, or 10). Every night I would put the correct amount of PHE in leaves on the tree. Whenever she would eat something, we would take off the amount of PHE in leaves ("falling leaves"). It didn't take long for her to catch onto the concept."

Learning to Calculate Phe

A great way to teach your child how to calculate Phe is to count the milligrams/exchanges together. You may already be using a whiteboard or laminated paper with a chart to count the Phe eaten at each meal. You may start by writing the total amount of milligrams/exchanges their child can have each day at the top of the chart, and list meals under this amount. To encourage your child's independence, you can begin to get your child to list meals and the amount of Phe he or she can have each day.

Discuss with your child the milligrams/exchanges planned for that meal. Next to each meal, parents or children can write the milligrams/exchanges planned for that meal. As the milligrams/exchanges are eaten during the day, have your child can cross them off. iPhone applications are also available for keeping track of Phe intake, including the DietWell application.

There are many other ways to create games or charts that help children count Phe – you can use your creativity or a solution that works for your family. The most important thing is that you work on counting Phe together so that your child begins to gain the skills he or she will need to manage PKU independently.

A Parent's Perspective

"Teach your child to measure/weigh her own 'milk' and be totally responsible for it by the time she leaves elementary school. Start this around age eight to nine by doing it in stages, maybe having her make her milk one to two times a week with supervision on the weighing. Once you are confident she can tackle the weighing, add quantity, then responsibility. Hopefully, by the time the child is 10 to 11 she will be making her milk on a daily basis with gentle reminders. Remember, they might not do it the way you do. For instance, I would measure out a week's worth in my son's containers, put the lids on and then the night before add the appropriate amount of water and put in fridge for the next morning. I made one mess with the weighing, cans, and scooper and then was basically done for the week. My son does it differently. About half the time he waits until the morning of to make it and the other half the time we get one measured the day before. I am learning very slowly to close my mouth and that other people do it differently."

Ages 7 to 12 Years

Bringing Formula to Class and Preparing Formula
Your child will most likely need to take her medical formula to school in order to spread intake throughout the day. Approximately, 1/3 of the formula for the day is an appropriate goal for formula consumption while at school.

- They can take pre-made formula in a fun sports bottle.
- Ready-made formulas such as Vitaflo Coolers are another option.
- Parents can talk with the teacher as to whether they would be allowed to drink some medical formula in class.
- They can go to the school nurse's office to drink formula.
- Pack the formula with the lunch and they can drink formula with their lunch.
- If they have after school activities, such as sports, they can have some formula then.

Blood Phe Monitoring

Goal Phe Range: 2-6 mg/dL (120-360 µmol/L)

Frequency

At this age you should check your child's Phe level every 2 weeks, unless directed otherwise by your clinic.

Tips for Taking a Blood Sample at this Age

From seven to twelve years, children can begin to take ownership over taking samples. Preparing them for this will help them to feel independent and confident. Tips for taking blood samples at this age:

- Have a calendar with the date marked when blood samples are taken to your child
- Continue to let your child choose which finger to use for the blood sample.
- Have your child fill out their name and date on the filter card.
- Encourage your child to press the button on the lancet pen.
- Ask what a correct sample looks like, and encourage your child to see how well he or she has done in producing a correct sample.
- Give lots of positive feedback on how well your child has done.
- By twelve, some children will be able to do this independently.

Quick Tip
Use topical "numbing agents" such as Pain Ease® that help reduce the pain of the finger stick. Ask your PKU team if this is an option to use.

A Parent's Perspective
"I took my son to CVS and let him pick out a lancet device. Now he loads it and pricks himself. That has helped the process a ton."

Special Considerations

Preparation for Childhood Education

With pre-planning and communication between the family, school and PKU team, children with PKU can transition easily to a school setting. (See Chapter 4 for information on preparing for daycare).

Ages 7 to 12 Years

Each preschool or elementary school is managed differently. The following suggestions are a guide for planning and discussing your child's PKU management with school staff. Most clinics will also have a dietitian or nurse who can talk to the school to help with planning and to provide information on PKU.

Talking with School Staff

Letting your child's school and teacher know about his or her PKU is the first step. It is important for teachers and other staff to understand why your child needs a special diet, and why careful supervision is needed. Anyone who comes in contact with your child will need a basic understanding of your child's dietary restrictions. Some key points that you may want to include:

- PKU is a genetic condition that is not contagious.
- Apart from needing a special diet, a person with PKU is healthy.
- People with PKU cannot break down an amino acid called phenylalanine or Phe, which is found in all foods containing protein.
- Phe can build up in the blood and damage the developing brain.
- Staying on a low protein diet keeps Phe levels in a safe range, allowing for normal development and a healthy life.
- Eating the wrong foods will not make a person with PKU sick right away, but will cause problems over the long-term. Having food that is not part of the diet should not be considered a "treat" as it will have implications for an individual with PKU.
- A person with PKU does not outgrow it and must stay on the diet for life.

A Parent's Perspective

"Since there were many unannounced birthday cupcakes or cookies that we were not able to plan for, I kept a container of food items in my son's classroom that were in separate baggies. He could pick from these if he was unable to have the unannounced food. Each baggy had a label of the food item and then the teacher would send it home so I would know what he had."

You should let the following people know about your child's PKU:

- The preschool director, or the school principal
- The class teacher(s)
- Early drop-off or after-care supervisors and staff
- Any food service staff that might provide meals for your child
- A school nurse, if applicable

Changes in school or classroom practice or policy may also need to be made to help your child adhere to their diet, such as:

- Creating a "no-swapping" rule to prevent children from trading or sharing food.
- Asking teachers to pack any leftover food in your child's lunchbox so you can determine how much was eaten.
- Keeping low protein snacks at school when snacks or treats are necessary.
- Allowing child to drink formula at school or in the classroom.

Ages 7 to 12 Years

School-Provided Lunches

You can pack lunches and snacks from home for your child, but some parents and children prefer or need to use school-provided lunches.

Each preschool or school will vary in what it provides in terms of meals or cafeteria food. Regulations that require schools to provide school lunches for children with special dietary needs differ from state to state. Several federal regulations provide the legal basis for requiring schools to offer nutrition services to children with special needs. These include: The Rehabilitation Act of 1973, The Individuals with Disabilities Education Act (IDEA) of 1990, and the Americans with Disabilities Act of 1990[31].

The Rehabilitation Act of 1973 mandates that students with disabilities not be excluded from any program which receives Federal Financial Assistance (section 504). Further, as part of the USDA's nondiscrimination regulation, federal law requires that schools make substitutions in foods for children considered to have a disability, and whose disability restricts their diet (7 CFR, Part 15b.3). Additionally, USDA regulation explains school requirements and specifies that schools must serve special meals at no extra charge to students whose disability restricts their diet (7 CFR, Part 15b.26(d))[32].

This means that schools need to work with families to provide appropriate meals for children with special dietary needs; however, regulations do vary by state. Some states may only require that schools provide low-protein choices from their regular menus, while others may actually order special low-protein foods for your child's meal. You will need to learn the requirements for your state and work with your child's school to determine the menu for your child's school lunch.

For your child to receive a special school lunch, you may also need medical authorization or need to fill out an Individualized Education Plan (IEP). Talk to your child's school about these requirements.

Regardless of your child's school lunch policies, here are some helpful tips for working with the school to ensure your child receives the diet she needs[33]:

- Get to know the Food Services Director and staff.
- Encourage them to contact you with questions about your child's diet.
- Limit the staff trained to prepare food for your child to one or two people.
- Ensure there is a good understanding of your child's special diet to avoid any mistakes in interpreting diet orders.

[31] Cambrooke Foods. Low Protein School Lunch Program. Parents Guide.
[32] Cambrooke Foods. Low Protein School Lunch Program. Parents Guide.
[33] Cambrooke Foods. Low Protein School Lunch Program. Parents Guide

Ages 7 to 12 Years

Packed Lunches

You may find that it's easier to pack lunches. Allowing your child to choose lunch options will make him or her happier with the choices and preparing a list of options in advance will make it easier for both of you. Some ideas for packed lunches are:

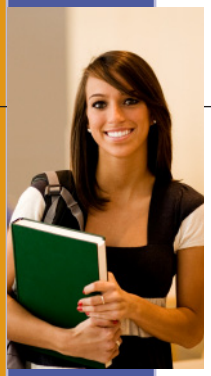
- Sandwiches made with low protein bread
    - o Vegetables sandwiches are delicious – try roasted vegetables for a tasty option
    - o Low protein cheese on its own or paired up with a variety of vegetables to change it up
    - o Low protein peanut butter and jelly sandwiches (remember to ask in advance if your child's school allows peanut butter as some schools are "nut free" due to allergies
- Low protein pasta or rice can be brought in a thermos to keep it warm
- Make a fruit cocktail with a variety of fruits for a refreshing snack
- Cut up lots of vegetables in advance and keep them in the fridge for quick lunch snacks
- Other low protein snacks can be kept in stock for variety such as low protein chips, puddings or fruit cups

After-School Activities

A Parent's Perspective
'I have found that people are so willing to accommodate his needs if I let them know what they are.'

Planning and preparation are needed if your child participates in after school activities. Parents can educate the after-school activity leader or coach about PKU and share that your child may need to take a break to drink formula or have a snack provided from home. Informing your child about the plan and ensuring that your child has extra formula or snacks that are appropriate for the length and physical intensity of the activity can help promote a positive health outcome.

A Parent's Perspective
"We create a menu for our son that is based on the school lunch menu on a monthly basis. Our son's menu has on it everything he can eat that is on the regular menu with the appropriate measurements, as well as what his substitutions should be for the high Phe food."



Chapter 7: Ages 13 to 17 Years

A Parent's Perspective

"I wanted to be sure that by high school my daughter did everything herself so by the time she left for college it was old hat and I wouldn't worry because I already stood by her independence through high school and the trials that come with the freedom there."

What to Expect

During adolescence, all parents have to release some control over their teen's life. This is especially true for the parent of a child with PKU.

Teens with PKU report feeling a lack of independence[34]. Help your teen develop a sense of autonomy by gradually giving up the role of "manager" of your teen's PKU, and move into the role of coach and supervisor. You may find this difficult as you have been continuously told of the important role you have in managing your child's diet, but by helping your child maintain good metabolic control previously, you have already laid the groundwork to ensure that your adolescent will be able to maintain their diet independently[35].

By allowing your teen to take charge of PKU, you will give your teen the opportunity to practice PKU self-management and, with your support, prepare to make the transition to adult care while building confidence in his or her abilities. You can help your teen begin the transition to adult care by[36]:

- Talking to your teen about health care he or she will need in adulthood.
- Trying to find out **how your teen feels** about independently managing the PKU diet and treatment.
- Helping your teen **identify the self-care skills** he or she can do independently
- **Identifying which skills still need to be learned.**
- Talking to your teen about the **warning signs associated with unsafe phenylalanine (Phe)** levels.

Teens, take this quiz to see your level of independence. Strive to achieve checkmarks for all line items – and remember, your parents are always there to help!

- ❒ I speak confidently about food restrictions.
- ❒ I can tell my friends about PKU and the PKU diet.
- ❒ I am comfortable ordering food independently at restaurants and know what kinds of food to look for.
- ❒ I can plan my PKU meals independently.
- ❒ I can pack a bag of PKU foods to take to school or to a friend's house.
- ❒ I know to eat low protein foods and formula before an event to avoid being hungry and overdoing it at a party.
- ❒ I can take my blood samples independently.
- ❒ I keep track of my diet record independently.
- ❒ I speak for myself at my medical appointments.
- ❒ I can discuss my feelings with a trusted person.
- ❒ If I don't feel like I can manage my feelings, I know I can talk to my PKU team, and they will help me or find someone that can.

[34] Jusiene R, Cimbalistiene L, Bieliauskaite R. *Psychological adjustment of children with phenylketonuria Medicina* (Kaunasa) 38 (2002) 424-430

[35] Gleason, L.A., Michals, K., Matalon, R., Langenberg, P., Kamath, S. *A treatment program for adolescents with phenylketonuria* Clin Pediatr (Phila) 1992;31(6):331-5..

[36] University of Washington. Adult Metabolic Transition Project. Available at: http://depts.washington.edu/transmet/whatis.htm

Ages 13 to 17 Years

- Helping your teen **create an action plan** to follow when warning signs appear.
- Asking your teen to **take more control** of their medical appointments. This may include creating a list of "things to do" before appointments so your teen can practice getting ready on his or her own.
- Encouraging your teen to **speak for him- or her-self during medical appointments.**
- Working with your clinic team to **find a primary care physician** for your teen as he or she approaches the age of 18.
- **Meeting with the physician** to ensure his or her familiarity with caring for a patient with PKU.
- Talking to your teen about **education and career goals**, and help him or her work through the steps to achieve these goals (for more information about college, see the college section in Chapter 8: 18-Adulthood)
- **Discussing how sticking to diet and treatment will help your teen reach these goals.**

Even as your teen moves into complete adult care, you will still be an important re source. Embrace your new role, and encourage your teen as he or she assumes full responsibility for his or her PKU.

Warning Signs of High Phe Levels

- Difficulty focusing on tasks (paying attention
- Anxiety
- Depression
- Inappropriate behavior, mood swings, or confusion about reality
- Headaches
- Tremors (the shakes)
- Eczema
- Stiff or weak legs
- Agoraphobia (fear of public places)

A Parent's Perspective

"Allowing her to order in restaurants instead of speaking for her was also tough but once she got the hang of it, we were so proud every time we went out for dinner!"

A Parent's Perspective

"Your PKU child may go from total compliance to throwing caution to the wind at various times in her life. Do not give up on her. Continue to show her that you believe the diet is important by finding ways to demonstrate this. That could mean anything from attending Annual PKU meetings or hosting an event to something as simple as leaving PKU News out on the coffee table. Any little thing you do related to the PKU diet will show her you are continuing to support her even when she seems to be giving up."

"Managing PKU can be difficult but if you are consistent and committed, you can provide your child with the tools he/she needs to manage PKU successfully for a lifetime."

Ages 13 to 17 Years

Development

Developmentally, adolescents continue to gain problem-solving skills and begin to think abstractly. This includes the ability to think through hypothetical situations and imagine outcomes that result from actions, which becomes important as they transition to adult care and long-term planning around PKU management[37].

The primary challenge during adolescence is relaxation of the diet due to the perception that there is no immediate harm[38]. While poor metabolic control is of concern, some research suggests that if PKU has been well-managed earlier in life, the consequences of poorer PKU maintenance at this age are less severe, and deficits can be regained when Phe levels are brought back under control[39]. However, adolescents with higher Phe levels show reduced abilities in problem solving, reasoning, inhibitory control and other cognitive abilities[40]. For many, going back to diet can be more difficult than remaining on it. Encouraging your child's independent management and dedication to his or her health by pointing out the long-term benefits of sticking with the diet and the long-term consequences of relaxing it is important at this time.

Teens 13-17 years of age, you should work with parents on:

- Being able to independently prepare formula.
- Independently managing total Phe intake for the day.
- Learning menu planning.
- Taking responsibility for diet records.
- Being able to take blood samples or schedule blood sample at the PKU clinic independently.
- Explaining the basics of PKU.
- Remembering recent blood Phe levels.

- University of Washington. PKU and Self Management Timeline Available at: http://depts.washington.edu/transmet/process/timeline.pdf Accessed May 18, 2011

As your teen experiences the changes that come with adolescence, there can be challenges in PKU management. As a parent, your help and guidance is essential to ensure that your teen is ready to make the transition into adult life.

Treatment

For active and growing teens, it is important to consume the prescribed amount of medical formula during the day, as well as eat a balanced diet of low protein fruits and vegetables, and low Phe breads, pastas and rice (find tips on drinking more formula throughout the day in Chapter 8).

Teenagers who plan and prepare their own meals may be more likely to take an interest in eating right. This is especially important for teens with PKU. Keeping his or her own diet

[37]Cherry, K. Formal Operation Stage of Cognitive Development Available at: http://psychology.about.com/od/piagetstheory/p/formaloperation.htm Accessed May 18, 2011
[38] Bilginsoy, C., Waitzman, N., Leonard, C.O., Ernst, S.L. Living with phenylketonuria : perspectives of patients and their families. J Inherit Metab Dis. 2005;28(5) :639-649
[39] Waisbren. S. *Neurocognitive Functioning in PKU* Illinois 2010 Presentation
[40] VanZutphen, K.H., Packman W, Sporri, L., Needham, M.C., Morgan, C., Weisiger, K., Packman, S. *Executive functioning in children and adolescents with phenylketonuria.* Clin Genet. 2007; 72(1):13-8.

Ages 13 to 17 Years

record can increase a teen's awareness of eating habits and help him or her take responsibility for diet choices (see chapter 2 for details on how to fill out diet records).

Tips for a Healthy PKU Diet

- **Take the formula** at least three times a day with meals so it's spread throughout the day
- **Eat and count Phe/protein as directed by your PKU team** at meals and snacks
- **Eat plenty of fruit and vegetables** every day, as well as special low protein rice, pasta, bread and crackers based on your tolerance to Phe
- Make sure you're getting **sufficient vitamins and minerals** which comes from your formula. Your team may recommend taking a vitamin or mineral supplement, if needed
- **Drink plenty of water and limit soda, juice and high calorie drinks to 8oz per day**
- Eat the **right sorts of fats such as canola, walnut, olive oil** and limit overall fat intake
- Consume only **moderate amounts of sugar** and of foods containing added sugar

Teens, here are some tips from other PKU patients that may help you manage your diet independently:

- Take it in small steps.
- Set a goal and work on that for a few weeks. Then add in another goal. Before you know it, it will become a lifestyle and habit and not just checking things off the list.
- Use your family and friends to keep you accountable and help you in the process.
- Carry good snacks with you to class and sports.
- Little things can make a difference in what you eat and what routine . you have during the day!
- Stiff or weak legs
- Agoraphobia (fear of public places)

Blood Phe Monitoring

Goal Range: 2-6mg/dL (120-360µmol/L); levels up to 10 mg/dl or 600µmol/L may be permitted in some clinics

Frequency

During the teen years, samples should be taken approximately twice a month, although frequency may vary from time to time. Teens should be responsible for their own blood sampling, although they may still want assistance. This will help develop a sense of self sufficiency and empowerment for a teen with PKU.

Tips for Taking a Blood Sample at this Age

As a parent, all the tips you used for taking samples can now be transitioned to your teenager. Help them to develop good sampling habits with the tips below, and any advice that you may have discovered on your own over the years.

- Have your child set up a reminder on their computer or cell phone with the date marked when blood samples are due
- Encourage your teen to take his or her own sample, but be there to help, especially at first.
- Ask your teen to evaluate if the samples are done properly. If you don't think they are, ask your teen what they think may be wrong with the sample to see if they can identify issues independently.
- Give positive feedback on how well your teen has done.

Ages 13 to 17 Years

- Have your teen record the results of blood tests when they are available; discuss with him or her any changes in management that might be recommended based on the test result.

Special Considerations for 13-17 Year Olds

Peer Pressure

Adolescence can be a challenging time to maintain treatment due to pressures from peers to conform; social support becomes an essential part of managing PKU treatment[41]. Drinking formula and eating a restricted diet may make a teen feel alienated, but it is important for teens not to give in to this pressure to conform. Work with your teen to come up with explanations he or she is comfortable with to explain their diet and formula.

Some teens with PKU start to report psychological disturbances, internalizing disorders (such as depression, anxiety, etc.)[42] and show slower information processing speeds[43]. This means that it can take longer for a teen with PKU to comprehend and respond to new information he or she receives. High school academics require higher executive functioning capabilities – including memory, planning, attention and organization – which can also be impacted by PKU[44]. However, there are many strategies that can be introduced to help teens with PKU manage these issues, and teens who maintain metabolic control can reach their full academic potential. Encourage your teen to speak with other teens with PKU and talk to your PKU team if you and your teen need support in maintaining treatment for life.

A Teen's Patient Perspective

I was always very open with my classmates, friends, and acquaintances about my diet. When people asked what my formula was, I told them it was my "milkshake" that I had to drink because I couldn't eat protein. I then rattled off all the foods I couldn't eat and told them that eating protein would cause brain damage. By explaining everything up front, people realized how serious PKU was and respected any special requests I would make. It made it much easier to suggest PKU-friendly restaurants or tell my friends that I had to stop by at my dorm room to take my formula. Every person you meet provides an opportunity to spread awareness for PKU!

Social Gatherings

Teens with PKU report feeling socially restricted[45] . Encouraging social interactions may help teens feel less isolated and helping your teen feel comfortable in these social situations may facilitate this.

For teenagers, just like adults, many social gatherings involve food. As they gain independence, teens with PKU begin to navigate more of these situations on their own. As a parent, you can help prepare your teen to handle different situations through encouragement and support of their independence.

[41]Waisbren. S. Neurocognitive Functioning in PKU Illinois 2010 Presentation
[42]Sullivan, JE. *Emotional outcomes of adolescents and young adults with early and continuously treated phenlketonuria* J. Pediatr. Psychol. 2001: 477-484
[43]Waisbren. S. *Neurocognitive Functioning in PKU* Illinois 2010 Presentation
[44]Waisbren. S. *Neurocognitive Functioning in PKU Illinois* 2010 Presentation
[45]Jusiene R, Cimbalistiene L, Bieliauskaite R. *Psychological adjustment of children with phenylketonuria* Medicina (Kaunasa) 38 (2002) 424-430

Ages 13 to 17 Years

- Ask your teen if he or she wants to bring a low Phe food to an event to share with everyone. This ensures that there is at least one "safe" food for your teen to eat.
- Prepare a list of foods that are appropriate for snacks with the Phe content. If your teen is invited somewhere unexpectedly, it will be easy to determine what he or she can have and keep track of how much Phe has been eaten.
- Remind your teen that it is always OK to say no to foods that he or she is offered.
- Ask your teen questions that they may get asked in social situations so they can prepare in advance.
- Let your teen know that they can talk to you about their feelings and how they're managing their PKU – even if it's something they think you won't want to hear.
- Remind your teen that you are a resource for them, and that no matter what, you're there to help!
- Encourage your teen to share with a friend and find a network of PKU teens to connect with over the internet or through their clinic (see resource list for websites)

Self-Image

All adolescents may feel judged by their peers on how they look. Many feel pressure to be thin or have a "perfect" body. However, it is especially important for teens with PKU to avoid diets for quick weight loss as restricting nutritional intake can force the body to use its own muscle for energy, resulting in unsafe blood Phe levels.

Having a positive self-image will help your teen focus on what is most important – taking care of his or her body to stay strong and healthy. Teenagers who have a positive self-image are more likely to manage their PKU properly, as well as avoid other risky behaviors. You can help your teen work on a positive self-image by reinforcing the tips for teens. If you are concerned that your teen needs additional support, ask your PKU team for suggestions and support group information.

During your teen years, it's normal to feel uncomfortable in your own skin! Developing self esteem is a never ending process. Here are some tips to help you develop and maintain a positive body image from an expert!

- **Be active**: Regular exercise puts you in a better mood and reduces anxiety. This gives you more confidence in yourself and the way you **feel about** your body.
- **Focus on what you do for your body each day, not on how others respond to it:** You can't control others' responses to you, but you can make healthy choices each day that will leave you feeling good about yourself.
- **Engage in positive activities:** Find time every day for a rewarding activity – do some exercise, go for a swim, play a sport, have a walk with a friend, listen to music. . .
- **Avoid people who give body shape or weight too much importance.**
- **Examine your own self talk and challenge distorted thinking about your body.**
- **Develop many sources of self-esteem:** So much makes you unique and special beyond your appearance. Develop listening skills to be a good friend, practice a skill in sport or the arts or enjoy a good book...
- **Develop perspective:** The older you get the less importance people place on judging people by their appearance. It gets better!
- **Judge yourself as a whole person, not just a body:** Create a list of people you admire who have contributed to your life, school, community, and the world. Was their appearance important to their success and accomplishments? If their appearance was not important what was?

-Kearney-Cooke, A. (2003). 'Helping Adolescents Become Strong Adults'. A Project of the Partnership for Gender-Specific Medicine at Columbia University, U.S.A.

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Risky Behaviors

Alcohol
There are many risky behaviors that a teen will be confronted with. While they are not legally able to drink alcohol yet, teens may encounter situations where their friends are drinking during parties. It is important to advise your teen that alcohol contains Phe, and should be avoided. If he or she intends to have a drink, the amount of Phe consumed from alcohol should be accounted for in the overall Phe intake for the day. See chapter 8 for more information about alcohol consumption.

Becoming Sexually Active

Speaking to your teen about the risk of becoming sexually active is important for all parents. This is especially true for parents of young women with PKU. If women with PKU become pregnant while blood PHE levels are not under strict control, serious damage may occur to the developing fetus, resulting in heart and brain defects associated with Maternal PKU Syndrome (see Chapter 9 for more on pregnancy and Maternal PKU Syndrome).

You may not feel ready for your teen to be sexually active - you may not even feel ready to talk to your teen about being sexually active – but it is essential for any young woman with PKU to use birth control if she becomes sexually active.

Schedule an appointment for your teen with a gynecologist to discuss her PKU condition and birth control options. Let your daughter know that you are always available to talk to her about anything, even sexual questions she may have. Some teens won't be comfortable speaking with their parents about sexual topics. To ensure that this doesn't limit the information they are accessing, assure your daughter that she can go by herself to her gynecologist appointment if that makes her more comfortable. Also, reassure her that you are always there to talk to her without judgment.

Teens, test your knowledge.
Did you know:

- ❒ Girls can get pregnant before their first period.
- ❒ Birth control pills do not protect you from sexually transmitted disease (STDs).
- ❒ Some medications (like antibiotics) can make birth control pills not work effectively.
- ❒ Condoms do not protect against pregnancy and STDs 100% of the time.
- ❒ Abstaining from sex is the most effective method of birth control and STD prevention.
- ❒ If a woman with PKU gets pregnant with high PHE levels, the baby can be born with serious damage, including brain and heart defects (see Chapter 9 for more information about this and how a PKU woman can have a healthy baby).

Chapter 8: 18+ to Adulthood

A Parent's Perspective

"For individuals with PKU, a controlled diet for life is the only answer to unlock the greatest opportunities."

What to Expect

Take this quiz to see your level of independence. Strive to achieve checkmarks for all line items – and remember, your PKU clinic team is always there to help!

- ❒ I can explain PKU.
- ❒ I speak confidently about food restrictions.
- ❒ I am comfortable ordering appropriate foods at restaurants.
- ❒ I can plan my PKU meals independently.
- ❒ I know how to order medical formula and ensure I have enough in advance.
- ❒ I know to eat low protein foods and formula before an event to avoid being hungry and overdoing it at a party.
- ❒ I take my blood samples independently.
- ❒ I keep track of my diet record independently.
- ❒ I attend medical appointments with my PKU team regularly.
- ❒ I have discussed sexual health with my healthcare team and understand how to use birth control effectively.
- ❒ I know the dangers of high Phe levels and know to see my PKU clinic for advice and support.
- ❒ I have information about my insurance and manage it independently.
- ❒ I am aware of the resources available to me.

Adulthood can be a very difficult time for an individual with phenylketonuria (PKU). Some people with PKU have relaxed their strict diet, but begin to realize that their attention, concentration and relationships have been negatively impacted by high phenylalanine (Phe) levels [46].

High Phe levels can impair your executive functioning capabilities, such as memory, planning, attention, and organization. As these are the very skills that are needed for managing PKU, it is important for you to maintain control so that you have the ability to plan your diet, remember Phe intake for your records, remember to drink your formula, maintain your PKU supplies and monitor blood Phe[47]. Following your low Phe diet and treatment is the best way to avoid neurocognitive problems and to maintain your ability to organize your life with PKU. Maintaining your diet and treatment will help you to feel healthy both physically and mentally. Diet for life – maintaining the PKU diet throughout your lifetime – is the best way for you to avoid problems associated with excess Phe.

The good news is that metabolic control for life is possible. The low protein food and formula options available today make the PKU diet more manageable than ever, and research continues to uncover new ways to treat PKU. Most people with PKU find they have more success maintaining treatment for life when they are supported, have a positive attitude, and find a way to make the PKU diet manageable for them. This is known as S.A.M.

46 Waisbren, S. *The Psychology of PKU*. NPKUA annual conference in Dallas, January 2010 Presentation
47 Waisbren, S. *The Psychology of PKU*. NPKUA annual conference in Dallas, January 2010 Presentation

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S.A.M: Support, Attitude, Manageability[48]

S.A.M. is an acronym for the key factors that lead to success in staying on diet or returning to the PKU diet. It is natural to sometimes feel like you can't succeed or the diet is too difficult to follow. However, it is important not to lose all hope and give up. Sometimes you just need a reminder of how to handle a certain situation. Just remember S.A.M. is here to help see you through the hard times and assist you in achieving better health.

S is for Support: This means you have people around you who believe in the treatment for PKU and support you in following the diet.

You are not alone. Building a base of people who support you in maintaining the diet is critical. Whether it be a friend, parent, significant other or a relative, having someone to confide in, talk to, and yes, even complain to is important. Who you choose to talk to is up to you, but you need people who believe in the benefits of "diet for life."

Speak with your local PKU team to find out contact information for other people in your area with PKU. You will not only gain support from someone who understands, but you will also have the chance to make a new friend. There are also online support groups and websites listed at the back of this resource that allow you to voice your thoughts, share ideas, and meet others that have PKU and understand the need for diet.

A is for Attitude: This means that you have a positive attitude about your formula and following a low protein diet.

The attitude you have toward PKU and following the diet is directly related to your success. A positive outlook and attitude is critical toward gaining and maintaining metabolic control. In terms of the PKU diet, a positive attitude means being open to trying new things, exploring new formula and food options, and accepting PKU as a part of you. Being angry and mad about having PKU will not remove the fact that you do need a special diet. Overcoming a negative attitude toward your situation will remove a major barrier to your future success. Viewing the PKU diet as just a different style of eating can boost your overall acceptance of the diet and foster a positive outlook. You may realize that following a strict diet has taught you skills that will help you excel in your daily life, such as increased creativity, self control and healthy meal planning.

M is for Manageability: This means you have found ways to make the diet work for you.

Manageability, as it relates to PKU, is how you are going to implement the diet into your daily life. It can mean anything from obtaining insurance coverage for medical foods to what you do in certain situations, like eating out or social gatherings. Man ageability is your own custom-made plan for following the PKU diet and finding one that works best for you. When you have the right tools, anything is possible. Writing

[48] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey : Applied Nutrition Corp. 2007:24-25.

a plan that includes the time of day you are going to consume your medical food and what you will do if you are away from home, will help you be prepared.

Transitioning to Adult Care

Transitioning

The term transition means change. All people face transitions at various times in their lives. In regards to a person with PKU, transition also means a change in responsibility for your health and medical needs. Now is the time to learn how to rely more on yourself and take charge of your own PKU needs. Transition can also mean[49]:

- Taking on new challenges.
- Gaining a new sense of freedom and independence.
- Having more choices.
- Doing more things on your own.
- More responsibility[50].

Your Transition Checklist

Get in the practice of doing the following things. They will help make your move to adult health care management easier.

- Know how to explain PKU and communicate your health care needs.
- Keep a record of appointments, medical history and medications prescribed.
- Write down your doctors' names, phone numbers and addresses.
- Begin to make your own medical appointments.
- Write down questions for your doctor before your visit.
- Have parents, friends, or your significant other remain in the waiting room while you spend time with your healthcare provider on your own.
- Learn about your health insurance and type of formula coverage your policy offers.
- Learn new information about the health care needs of an adult with PKU.
- Prepare and educate yourself on personal PKU management. For example: How to obtain formula, monitor your blood PHE levels, count PHE in your diet, etc[51].
- If you're more comfortable, you can consider having someone with you for support, like a friend, parent or someone else you trust.

Relationships

Your Family [52]

Up until now, your family or someone in your family has taken care of you and your needs, PKU and otherwise. They have brought you to your healthcare provider and clinic visits, and have taken care of you when you were sick. Now that you are the leader of your own health care and PKU management, your family will have a transition also.

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[49] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:21

[50]Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:22.

[51]Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:22.

[52]Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:29.

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The maturity associated with adulthood will overflow into your daily life. Your family will have to understand that now you make the important decisions regarding your PKU needs. In order to not be viewed as a child, you must act like an adult and accept responsibility for your actions.

Your family will always be there to support you with the endeavors and choices you make throughout your life. Your transition to managing PKU on your own should be no different. Remember, your family is still a tremendous source of knowledge regarding your dietary needs and you can always go to them for support and advice when needed.

Your Friends [53]
The relationship you have with your friends will also experience a transition as you grow and age. As you get older, the friends you already have and the new friends you make may want to know more about PKU and your diet. Embrace this. Talk openly and share with them what life is like with PKU. Having the support from your peers will only make following the diet easier.

Treatment and Diet

Returning to Treatment
As an adult, you may have strayed from the strict, low protein diet and allowed a degree of relaxation to your diet. You may have many questions about how to transition back to the PKU diet and maintain your treatment.If you are an adult who, for whatever reason, has modified or abandoned PKU treatment for a period of time but would now like to return to metabolic control to protect your health and mental functioning, this next section will help you.

Medical Formulas

You may be familiar with traditional powder formulas, which are commonly used as the main source of protein for people with PKU. More recently developed medical formulas come in many new flavors that some may find more palatable than previous medical food, convenient single serving sizes (such as formula packaged in juice box sized containers that are ready to drink) and in powders that can be added to foods and beverages. Some of these formulas are lower in volume than traditional formulas as they are lower in fats and carbohydrates, but most of them have the required vitamins and minerals. You can try out different styles and flavors to find the right formula for you[54].

[53] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:29
[54]Naziri, M. Meal *Planning on a Budget* PKU Bootcamp Children's Memorial Hospital. April, 2011 Presentation

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Low Protein Foods
Offerings of foods modified to be low in protein have also expanded significantly. Specially formulated breads, pastas, non-meat burgers, cheeses, muffins and cookies are now available, among others, and low protein food companies continue to offer new options for people with PKU.

There are also less expensive foods naturally low in protein that available off the shelves such as a few listed below.

PRODUCT	PRICE	PHE
Sunbelt fruit & grain cereal bars (blueberry/strawberry)	$2.00 per box	45-55 mg per bar
Sensible Portions Veggie Straws	$6.00 per 7oz bag	32 mg per serving (38 straws)
Sandwich Mate cheese slices	$1.50 per pack (16 slices)	24 mg per slice
Sun Luck Rice Sticks	~$2.00 per bag	35 mg per serving (1/4 of bag)
KAME bean threads	~$1 per bag	3 mg per 56 gram serving (uncooked)
Pepperidge Farm Very-thin sliced white bread	~$4.20 per loaf	67 mg per slice
Turtle Mountain So Delicious Coconut Milk Yogurt	~1.80 per 6oz	28 mg per 6oz

Ask your PKU team for more examples of low protein foods you can get off the shelf. Also, the resource section of this resource has information on companies who offer medical foods. Some food companies offer free shipping with a certain amount of food ordered.

A Parent's Perspective

"I met David (who is now my husband) and realized that there really could be a future for us. Simply thinking about the future- marriage, children, etc. made me realize that I had to get this under control! This was the first spark in my adjusting back to the regular PKU life and habits."

Transitioning to the PKU Diet[55]
Your PKU team is a good resource as you return to diet, and will help you work out a treatment plan. It is important to have good support from family and friends as you make this transition.

When an individual is off their low Phe diet and not drinking any formula, 100% of their protein comes from dietary intake of natural protein. This will lead to high Phe levels, and

[55] Naziri, M. *Adult Outreach*. December, 2009 Presentation

can lead to health issues previously discussed. When you decide to return to diet, your PKU team will help develop a step-by-step process to slowly reduce the amount of protein coming from natural food items, and increase the protein intake from formula to balance your diet and reduce your Phe levels. This will involve substituting medical formula and low protein foods for high protein foods in your diet over time. An example of how this transition might work is below. As this example shows, you will transition gradually to your diet for life, and your PKU team will work closely with you to ensure that you know how to create a diet that will work for you and your lifestyle. Some people will have been eating a modified low Phe diet, so you may find your diet is more similar to one of the later steps in the diet. You can find where you currently fit, and start from there.

Some people who have been off diet say that when they return to the PKU diet they:

- Feel better
- Look better
- Are less moody
- Find it easier to get along with others
- Have more energy
- Can concentrate
- Can think clearly to study or work
- Can complete assignments or work projects more easily
- Can think more strategically, such as in team sports or at work

5 Steps to the PKU Diet[56]

Example of The "Off Diet":
100% of protein comes from dietary intake of natural protein

BREAKFAST	SNACK	LUNCH	SNACK	DINNER
Coffee 1 egg 2 slices toast	Donut	2 pizza slices 1 breadstick	1 oz. bag of Lay's chips	cheeseburger medium fries soda
TOTAL PROTEIN: 54 GRAMS				

STEP 1:
75 % of protein comes from dietary intake of natural protein;
25% comes from formula protein

BREAKFAST	SNACK	LUNCH	SNACK	DINNER
Coffee ~~**1 egg**~~ **+ 1 apple** 2 slices toast	Donut **+ medical formula**	2 pizza slices 1 breadstick	1 oz. bag of Lay's chips	cheeseburger medium fries soda
TOTAL NATURAL PROTEIN: 49 GRAMS **TOTAL FORMULA PROTEIN: 15 GRAMS**				

[56] Naziri, M. *Adult Outreach*. December, 2009 Presentation

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STEP 2:
50% of protein comes from dietary intake of natural protein;
50% comes from formula protein

BREAKFAST	SNACK	LUNCH	SNACK	DINNER
coffee 1 apple 2 slices toast	~~**donut**~~ medical formula	2 pizza slices 1 breadstick	1 oz. bag of Lay's chips **+ medical formula**	~~**cheeseburger**~~ **+ bun only** **+ 1 leaf lettuce** **+ 2 slices tomato** ~~**medium fries**~~ **+ small fries** soda
TOTAL NATURAL PROTEIN: 36 GRAMS **TOTAL FORMULA PROTEIN: 30-35 GRAMS**				

STEP 3:
20% of protein comes from dietary intake of natural protein;
80% comes from formula protein
+ medical formula
Total natural protein: 17 grams
Total formula protein: 60-70 grams

BREAKFAST	SNACK	LUNCH	SNACK	DINNER
Coffee 2 slices toast 1 apple + medical formula	medical formula	~~**2 pizza slices**~~ **+ low protein pizza crust** **+ 2 tbsp sauce** **+ ¼ cup low protein cheese** ~~**1 breadstick**~~ **+ 1 cup lettuce** **+ 2 tbsp Italian dressing**	1 oz. bag of Lay's chips medical formula	bun only **+ mayo or mustard** 1 leaf lettuce 2 slices tomato small fries ~~**Soda**~~ **+ medical formula**
TOTAL NATURAL PROTEIN: 17 GRAMS **TOTAL FORMULA PROTEIN: 60-70 GRAMS**				

18+ to Adulthood

STEP 4:
15% of protein comes from dietary intake of natural protein;
85% comes from formula protein

BREAKFAST	SNACK	LUNCH	SNACK	DINNER
~~**coffee**~~ ~~**2 slices toast**~~ **2 slices low protein banana** bread 1 apple medical formula	medical formula	low protein pizza crust 2 tbsp sauce ¼ cup low protein cheese 1 cup lettuce 2 tbsp Italian dressing	1 oz. bag of Lay's chips medical formula	~~**bun only**~~ **+ low protein Camburger** **+ low protein bun** **+ mayo or mustard** 1 leaf lettuce 2 slices tomato small fries medical formula
TOTAL NATURAL PROTEIN: 11 GRAMS **TOTAL FORMULA PROTEIN: 60-70 GRAMS**				

STEP 5:
10% of protein comes from dietary intake of natural protein;
90% comes from formula protein

BREAKFAST	SNACK	LUNCH	SNACK	DINNER
medical formula 2 slices low protein banana bread 1 apple	medical formula	low protein pizza crust 2 tbsp sauce ¼ cup low protein cheese 1 cup lettuce 2 tbsp Italian dressing	medical formula ~~**1 oz. bag of Lay's chips**~~ **1 bag of Wise onion rings or ½ cup carrot sticks**	medical formula low protein Camburger low protein bun **+ mayo or mustard** small fries 1 leaf lettuce 2 slices tomato
TOTAL NATURAL PROTEIN: 9 GRAMS OR APPROXIMATELY 450 MG/PHE PER DAY **TOTAL FORMULA PROTEIN: 60-70 GRAMS**				

Tips for Increasing Medical Formula Consumption

As you transition to a low Phe diet, you will be replacing natural protein with protein from medical formula. It is important that you consume the amount of medical formula your PKU team advises to ensure that you are getting enough protein and other nutrients each day. Many adults try to restrict the protein in their diet without drinking the formula. This may result in health issues such as having low vitamin levels, poor bone health and difficulty maintaining a healthy weight.

The following tips can help you get enough medical formula:[57]

1. Drink a glass of formula with/or before every meal.
2. While grocery shopping or walking around a store, aim to finish a serving of formula before you check out or leave.
3. Drink a glass of formula on the way home from work, school or picking up your children.
4. Use a sports bottle to drink formula on the go or at the gym.
5. Substitute formula for other beverages you may have during the day.
6. Add crushed ice to make formula colder.
7. Spread your formula intake throughout the day. This allows your body to absorb all the nutrients consumed.
8. Change the taste of your formula with flavored extracts.
9. Be creative by adding formula to already made low protein food or flavored drinks.
10. Carry a small bottle of mouthwash or toothpaste to freshen your mouth after you finish your formula.

Getting Satisfaction from Food

Experiencing satisfaction from food is part of being human. Although many foods may be restricted or limited due to PKU, you do not have to compromise on taste.

Identifying the items and tastes that you crave is important. Knowing what you like and choosing foods that make you feel satisfied is part of enjoyable dining. Eating foods that don't satisfy you may leave you wanting more.

The first step is to identify foods that make you feel satisfied. Is it salty, sweet, creamy, spicy, fatty, juicy, crunchy or mushy that you look for in a meal?

Got a Craving?

- Add spicy seasoning, cracked pepper, crushed red pepper, or tabasco sauce to steamed vegetables to give them the fiery kick you crave.
- End your meal with some pineapple or other fruit to get the sweet and tangy taste that signals your body that the meal is done.
- Satisfy your sweet and creamy taste buds with some low protein chocolates or marshmallow treats.
- Change the texture of a low protein baked item by using applesauce instead of oil to make products lower in fat, soft, and fluffy.
- If crunchy is what you want, make your salads snap. Toast low protein bread to make your own croutons. Season them to meet your savory taste needs and toss over greens. 1

[57] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey : Applied Nutrition Corp. 2007:47.

18+ to Adulthood

Food and eating includes a variety of factors. Not only are taste and smell large factors in the foods we choose, but texture, sight, and our culture play a large role. So, make the most out of your dining experience and keep the zesty seasonings handy. Be creative!

If you are still hungry at the end of your meal, there are many low Phe or Phe free foods that you can eat to satisfy yourself while not going over your Phe limit for the day. Commonly used low Phe foods include: fruits and vegetables, low protein rice or pasta with a little butter or canola oil and seasonings, low protein egg mix or low protein bread. Ask your dietitian for more ideas![58]

Tips for successful goal setting:

- **Aim for a realistic goal that is right for you.**
    - *For example,* if you are not currently drinking any medical formula it might be best to start with 1/3 or 1/4 of the goal recipe and work your way towards a 100%. Starting small will make sure that you get there!
- **Put in place a goal that is specific.**
    - *For example*, drink 2 cups of formula with breakfast and dinner each day of the week. That is a more specific goal than saying "I will start drinking my formula".
- **Plan for potential setbacks & reassess/adjust your goals as needed.**
    - *For example*, if you set a goal to completely cut out all high protein foods from your diet and find that the sudden restriction is too drastic it might be more realistic to first start by cutting back on the number servings of high protein food per day.
- **Think short term and long term.**
    - *For example*, a short term goal would be to decrease the amount of Phe you are consuming in your diet. This will get you closer to the ideal Phe intake and may take only a few weeks to reach. A long term goal would be to keep your Phe levels within treatment range. This long term goal may take several months or longer to achieve and maintain.
- **Focus on the process.**
    - *For example*, the long-term outcomes of the goals you'll be setting are to ultimately help you decrease your Phe levels. During this process you will set short-term goals like drinking the prescribed amount of formula every day. This process will help create behaviors and habits that will help you reach the long-term goal.

Kuvan® Treatment

If you are an individual that has been on diet since childhood, you may have already been tested for responsiveness to Kuvan® (sapropterin dihydrochloride). If you are responsive, you may have incorporated this into the management of your PKU. If Kuvan is part of your treatment, don't forget to take it every day. You can use your phone or computer to set up reminders.

[58]Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey : Applied Nutrition Corp. 2007:38.

18+ to Adulthood

For individuals who are returning to diet after an extended time, you may or may not have been tested for responsiveness to Kuvan. See Chapter 2 for information about treatment with Kuvan to support managing your PKU.

Blood Phe Monitoring
Goal Phe range: Although a Phe level under 6 mg/dl (360 µmol/L is ideal, levels up to 10 mg/dl or 600 µmol/L are often viewed as acceptable for adults

Once an individual with PKU is an adult, blood Phe is usually be monitored once or twice per month. Described below are some factors that may alter how frequently you should monitor your blood Phe levels.

AGE AND OTHER FACTORS	RECOMMENDED FREQUENCY OF SAMPLES
Pre-pregnancy and during pregnancy	Levels should be monitored 1-2 times per week. (see pregnancy section for more information)
Illness	Recommendations vary; frequency may be increased during and after illness to ensure consistent Phe levels
Returning to diet	Levels should be obtained after each adjustment in treatment
Physical training	Training may affect Phe levels. If you are concerned, discuss Phe level monitoring with your PKU team.
Dieting for weight control	Dieting may affect Phe levels. If you are concerned, discuss Phe level monitoring with your PKU team.

Special Considerations in Adulthood

College[59]

The first few weeks of college will be exciting and full of change. You will meet a variety of new people and have many new experiences. The one thing that remains constant is managing your PKU. Keeping levels in control when you don't have family watching over you may be more difficult than anticipated. Focus! Committing yourself to your PKU treatment and keeping your PHE levels in control is more important than ever.

Long lectures, late nights studying, and cramming for exams all require you to be at your best. Adhering to your treatment regimen, continuing your low protein diet and consuming your formula daily should be a top priority. If you are on Kuvan, you will need to develop a routine that helps you remember to take it daily, Elevated blood Phe levels result in

[59] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey : Applied Nutrition Corp. 2007:47.

compromised executive functioning skills such as memory, recall and concentration. Keeping your levels in control, especially before an exam, will help ensure your success in school.

Dining on Campus

Some schools require all first year students to be on a meal plan. Although you may not consume as much as your fellow students, meal time is a great social experience and can lead to many new friendships. If the meal plan is required, speak with the school nutritionist and general manager in the dining department. They may be able to stock and prepare some low protein foods for you.

A Patient's Perspective

"My freshman year of college, I think I missed out on a lot of things because I was stressed all the time and so overly emotional [from being off diet]."

How much Phe is in beer?

One 12 fl oz can of beer = about 40mg of Phe!
(The darker the beer the higher the Phe.)

For Phe content in other alcoholic beverages, look in the low protein food list for PKU.

Group dining usually offers a wide array of options, including a salad bar stocked with fresh fruits and vegetables. Be sure the people who prepare your food understand the true nature of your PKU and what accommodations you will need if you are not exempt from the meal plan. Maybe a discounted rate is possible so you can still enjoy the social aspect of group dining your first year.

If you are granted an exemption or live off campus, planning your meals is up to you. Choose a day to make a few different items so you spend less time cooking and more time learning. Once again, the key is to be creative and plan ahead.

Tips for Drinking Formula on Campus

- Add powder to an empty water bottle and fill with water later so you can drink your formula when you need it.
- Mix formula in advance and freeze it. It will defrost while you are on campus and you can drink it when you need a cold boost of energy.
- Is your dorm room far from campus? Use a locker at the school gym or student union to keep formula closer to your classes!
- Add formula straight to the bottle of popular store-bought beverages and drink while you are on campus.
- Use the ready to drink formula pouches

Alcohol[60]

There are many decisions you will make as you figure out how to make diet for life manageable for you. One of these choices includes whether or not to consume alcohol, once you are of legal age. Like all other choices involved with becoming an adult, consuming alcohol brings additional responsibility. You should already be aware of the dangers of drinking too much. If you have PKU and you choose to drink alcohol you need to be aware of additional issues involved with drinking.

[59] Maltzman, S, PKU Diet Management : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey : Applied Nutrition Corp. 2007:37.

Some beer and mixers contain significant amounts of phenylalanine. If you choose to consume these types of beverages you need to account for this in your total allotment of phenylalanine for the day. Both alcohol consumption and high phenylalanine levels impair your judgment. Even if you choose alcoholic beverages that are low in phenylalanine, impaired judgment can lead to poor food choices and overeating resulting in elevated blood phenylalanine levels. Moderation is the key.

Family Planning and Maternal PKU

If you are female and sexually active, you need to use birth control to prevent an unplanned pregnancy. Becoming pregnant while your blood Phe levels are not under strict control could cause serious damage to a developing fetus, causing a variety of heart and brain defects known as Maternal PKU Syndrome. Schedule an appointment with a gynecologist (a doctor who specializes in women's reproductive health) to discuss your PKU condition and birth control options. It is essential to use your birth control exactly as prescribed to ensure that you do not become pregnant until you are ready and able to plan for a safe pregnancy. See Chapter 9 for more on pregnancy and Maternal PKU Syndrome.

Weight Management

Adjusting eating habits for weight management is not unusual for adults. For an individual with PKU, changing your diet may affect your blood Phe levels. When an individual with PKU goes without food or medical formula, the body starts to break down some of its own stores of protein, resulting in an increase in the individual's blood Phe level.

Before altering your diet, it's very important that you contact your PKU team for information about how you can manage your weight without losing control of your Phe levels. Advice may include:

1. Reduce calorie containing beverages (soda, juice, sports drinks, energy drinks)
2. Increase fruit and vegetable consumption (limit starchy vegetables such as corn, potatoes, peas)
3. Make sure you are getting plenty of formula - protein from formula helps make you feel full. Talk to your dietitian about finding the right formula to limit calories.
4. Eat small, frequent meals
5. Drink plenty of water
6. Limit added sugar (candy, cookies) and fats

Increasing physical activity is a great way to help maintain a healthy weight. Physical activity will help you feel better, give you energy and may help reduce Phe levels.
Here are a few tips to increase physical activity:

1. Start with realistic goals
2. Increase walking by taking the stairs or walking places, get a pedometer
3. Join a gym
4. Have an exercise partner to help keep you on track
5. Join a class like dancing, pilates, yoga, local sports league

Chapter 9: PKU and Pregnancy

A Parent's Perspective

"Having PKU during pregnancy seemed like the biggest issue I would deal with; I was so wrong. There are worse things to deal with. PKU is manageable and if you follow your diet and stay in control, what seems scary becomes second nature." -Nicole, NJ

What to Expect

Women may experience a variety of emotions about having a child, both before and during pregnancy. Most parents feel concerns at one time or another about their ability to conceive, the health of a developing fetus, and how they will care for an infant. These feelings are all normal. For women with PKU, emotions around pregnancy may be heightened by worries about how their PKU disorder impacts the health of a developing fetus. With proper precautions and adherence to diet and treatment, women with PKU can have a normal pregnancy and a healthy baby

It is unlikely that an individual with PKU will have a child that also has PKU, although the child will always at least be a carrier. Testing to see if a partner carries the PKU gene is available. The partner must be a carrier in order for an individual with PKU to have a child with PKU. In a family where one child has PKU, prenatal testing may be available. However, because the treatment of PKU is so successful, it is rarely requested.

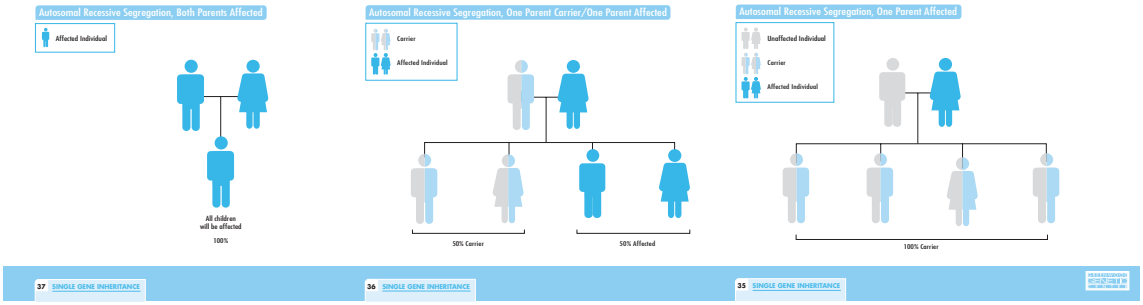

Illustration from Genetic Counseling Aids 5th Edition, Copyright 2007, permission for use granted by Greenwood Genetic Center

PARENT 1	PARENT 2	CHILD
PKU	Non-PKU Non-PKU carrier	100% chance of being a carrier of PKU
	Carrier	50% chance of having PKU 50% chance of being a carrier
	PKU	100% chance of being affected with PKU

A Parent's Perspective

"At times the PKU diet may seem hard or even impossible to follow, but once you hold that perfect baby in your arms, you realize it wasn't hard at all!" - Laryssa, NY

PKU and Pregnancy

Maternal PKU Syndrome
While having a child with PKU may be a concern for individuals with PKU, an even more serious concern may be the effect a woman's PKU may have on her fetus. High Phe levels during pregnancy can result in miscarriage or cause Maternal PKU Syndrome (MPKU).

MPKU affects the fetus and is believed to be caused by high, uncontrolled Phe levels, which may include:

- Heart problems
- Small head size
- Physical deformities
- Slow development
- Brain damage[61]

A Parent's Perspective

"It's important to remind yourself on a regular basis that you CAN do this, that you ARE doing it, and that, at the end, you'll have a beautiful, perfect, healthy baby in your arms! That outcome makes anything you face totally |worth it!" -Kerry, NY

Studies show that the later a woman with PKU gains control of her Phe levels during pregnancy, the more likely it is that the child will have:

- behavioral problems such as aggressive "acting out" behaviors[62]
- attention issues
- difficulty developing friendships
- lower intelligence

It has also been found that timing of when the mother gains control of her Phe levels is connected to the child's intelligence quotient (IQ), with earlier control being related to higher IQs[63].

The potential damage high Phe levels during pregnancy can cause to your child can be very frightening. To prevent damage from occurring to the fetus, it is extremely important that you plan your pregnancies by:

- Meeting with your PKU team 3-6 months before pregnancy
- Fine tune your diet with formula and Phe intake
- Increase Phe level monitoring to 2-4 times per month
- Keep Phe levels in the range of 2-6 mg/dL for 2-3 months prior to conception

It's important to use birth control until you are able to lower your Phe to the recommended level for pregnancy. Following the recommended diet to ensure that Phe levels are low when you become pregnant – and stay low throughout your pregnancy – is the only way to ensure your fetus develops safely.

Contact your PKU team if you are thinking about planning a pregnancy. They can provide guidance on achieving and maintaining safe Phe levels before and during pregnancy. They can monitor your Phe levels closely to ensure that your levels are safe before conception. Any questions you have about your treatment during your pregnancy can be addressed by your PKU team.

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[61]Acosta PA, Yannicelli S. PROTOCOL 2 – Maternal Phenylketonuria (MPKU) Nutrition Support of Pregnant Women With Phenylketonuria (PKU) With PHENEX™-2 Amino Acid-Modified Medical Food *The Ross Metabolic Formula System Nutrition Support Protocols 4th Ed.* Colombus Ohio, 2001:12-27
[62]Ng TW, Rae A, Wright H, Gurry D, Wray J. *Maternal phenylketonuria in Western Australia: pregnancy outcome and developmental outcomes in offspring.* J Paediatr Child Health 2003;39 358-63
[63]Maillot F, Lilburn M, Baudin J, Morley DW, Lee PJ. *Factors influencing outcomes in the offspring of mothers with phenylketonuria during pregnancy: the importance of variation in maternal blood phenylalanine.* Am J Clin Nutr 2008:700-5

PKU and Pregnancy

If you become pregnant unexpectedly, contact your clinic team immediately. The PKU team can help you reduce your Phe levels which will improve the outcomes for your child. Also, if you are not already on diet, immediately go back on diet and take the medical formula as prescribed.

Treatment

The amount of natural protein and phe that each woman with PKU can have varies widely depending on the severity of her PKU and how rapidly the fetus is growing. During periods of rapid fetal growth, phenylalanine tolerance increases because the fetus has a higher demand for phenylalanine in order to build its body tissues. This will allow you to eat more natural protein. Frequent monitoring of blood phenylalanine levels show if a woman is getting the right amount of phenylalanine. If your level is less than 2mg/dL you may be able to tolerate more phenylalanine from natural foods. If your level is greater than 6 mg/dL you are most likely getting too much phenylalanine and/or not enough formula. Your dietitian will advise you about adjusting your diet[64].

Formula will remain the most important part of your diet throughout pregnancy, since it is your main source of protein. It provides all the amino acids needed for a woman and her developing baby, minus phenylalanine which cannot be properly metabolized in PKU. Formula also provides calories, vitamins and minerals, and increases your feeling of fullness. During pregnancy with PKU there is no way other than formula to provide sufficient protein for proper fetal growth and development.

For many women, especially those who are returning to diet for pregnancy, drinking the formula may be a major challenge. Taking the time prior to pregnancy to find a formula you enjoy drinking is important.

Formula is best used by the body and baby when you drink it throughout the day; usually three or four servings are recommended. Your formula prescription will be designed specifically for you by your metabolic doctor and dietitian to meet your changing needs.

Sometimes formula may be difficult to tolerate, especially during the early weeks of pregnancy when nausea and vomiting are common. Since formula is critical to your success, contact your metabolic clinic and Ob/Gyn if nausea and vomiting are prolonged[65].

Tips for Staying on Diet and Drinking Formula During Pregnancy

- Mix formula with lemon or straw berry sorbet for a treat.
- Add a splash of seltzer for carbonation. Bubbles may help the formula go down easier.
- Increase the amount of water in your formula to meet additional fluid needs and prevent dehydration.
- Food cravings are sometimes common to pregnancy. Make a list of foods you desire and match with foods you can have. For example; low protein cakes, muffins, and chips.
- Keep a journal of your food and formula intake. It is the best way for you & your metabolic team to examine and control your diet.

[64] Maltzman, S, Maternal PKU : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey : Applied Nutrition Corp. 2007:56-57
[65]Maltzman, S, Maternal PKU : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey : Applied Nutrition Corp. 2007:56

PKU and Pregnancy

Controlling Phe

As your needs and the needs of your developing baby change each trimester, your PKU team may adjust your diet or medical formula as necessary, keeping a close watch on Phe levels to ensure they stay in the safe range. Both Phe and tyrosine blood levels are monitored closely throughout pregnancy. Tyrosine supplements may be prescribed if your level is low. It is important during pregnancy that you:

- *Take your medical formula consistently.* Drinking your formula as it is prescribed will help you get the caloric intake your body requires each day. Eating a small amount of low protein bread, pasta or crackers 15 minutes before you take your medical formula may help settle your stomach so you can drink it more easily.
- *Drink formula in small, frequent doses to help with formula tolerance.*
- *Eat regular small frequent meals.* Gaining or maintaining weight will help keep Phe levels in the safe range. Losing even a little weight at this time can make your blood Phe levels higher. Eating small frequent meals will also help prevent heartburn.
- *Maintain frequent blood tests.* Your clinic team will advise you if Phe levels need to be monitored more closely so that your diet and formula can be adjusted as needed.
- *Keep track of your Phe intake.*

Phe levels may start to drop in the 2nd and 3rd trimester due to increased growth of the fetus. You may be able to start eating more Phe in your diet. Your dietitian will monitor your Phe levels and make recommendations about the best way to increase your Phe intake.

Common Difficulties during Pregnancy

Many women feel sick to their stomach during pregnancy, especially in the first trimester, which may make it difficult to hold down your medical formula and food. Heartburn and constipation may also be a problem. These tips may help:

- Eat small amounts of prescribed food hourly while awake.
- When you wake up, eat some prescribed low-protein crackers.
- Drink fluids, especially water, between meals.
- Avoid fried or spicy foods and unpleasant smells.
- Eat only lightly seasoned foods
- Try plain fruits with skins and vegetables (raw vegetables may help with constipation).
- Eat some prescribed food before preparing meal.
- Sit up straight while you're eating, chew carefully and eat slowly.
- Go for a walk after eating.
- Avoid stooping or lying down after eating to reduce heartburn.
- For constipation, try having high fiber cereal in the morning and drinking prune juice.
- Daily exercise is always healthy, and continues to be during pregnancy. It can help with constipation and make you feel better overall[66].

[66] Acosta PA, Yannicelli S. PROTOCOL 2 – Maternal Phenylketonuria (MPKU) Nutrition Support of Pregnant Women With Phenylketonuria (PKU) With PHENEX™-2 Amino Acid-Modified Medical Food *The Ross Metabolic Formula System Nutrition Support Protocols 4th Ed.* Colombus Ohio, 2001:41

PKU and Pregnancy

Kuvan® during Pregnancy

In a small number of women with PKU studied, Kuvan® (sapropterin dihydrochloride) has been found to help control Phe levels during pregnancy[67]. There have been no harmful effects of the fetus reported with Kuvan use. However, the number of pregnancies exposed is too small to be certain that there are no adverse effects. Additional studies are needed on the safety and effectiveness of using Kuvan during pregnancy[68].

Blood Phe Monitoring and Health Assessments

High levels of Phe at any time during your pregnancy may harm your baby. Your fetus will be monitored with ultrasound examinations throughout your pregnancy to ensure that development is on track. It is essential that you follow your diet and take the medical formula as prescribed before and throughout your pregnancy to control your Phe levels and to ensure that you have enough tyrosine in your blood[69].

Blood Phe monitoring is also especially important while planning and throughout your pregnancy. In addition to monitoring your Phe levels, your clinic will also monitor your nutritional status to ensure you have the nutrients required to support you and your developing fetus throughout your pregnancy. Other assessments may include

- Complete blood count (test for anemia)
- Metabolic profile (chemistry panel)
- Prealbumin
- B12
- Selenium
- Total cholesterol
- Ferritin
- Folate

If your PKU team finds that your diet isn't providing enough of certain nutrients, supplements will be recommended.

Special Considerations

Parenting with PKU

Congratulations! You're the parent of a beautiful newborn baby! Like all parents, this is an exciting and stressful time. It may be even more stressful for a mother with PKU. Many women with PKU think that once the baby is delivered, relaxing their diet is acceptable. Many mothers report that they no longer use their medical food once their child is born[70].

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[67]Koch R. Maternal phenylketonuria and tetrahydrobiopterin Pediatrics 2008:1367-8
[68]Trefz FK, Blau N. *Potential role of tetrahydrobiopterin in the treatment of maternal phenylketonuria.* Pediatrics 2003:1566-9
[69]Acosta PA, Yannicelli S. PROTOCOL 2 – Maternal Phenylketonuria (MPKU) Nutrition Support of Pregnant Women With Phenylketonuria (PKU) With PHENEX™-2 Amino Acid-Modified Medical Food *The Ross Metabolic Formula System Nutrition Support Protocols 4th Ed.* Colombus Ohio, 2001:35
Rohr F, Munier A, Sullivan D, Bailey I, Gennaccaro M, Levy H, Brereton H, Gleason S, Goss B, Lesperance E, Moseley K, Singh R, Tonyes L, Vespa H, Waisbren S. *The Resource Mothers Study of Maternal Phenylketonuria: preliminary findings* J Inherit Metab Dis 2004:145-55

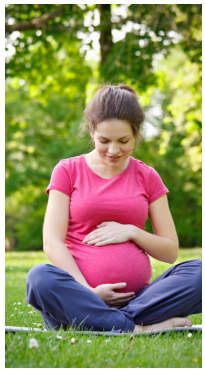
PKU and Pregnancy

However, high Phe levels result in a variety of issues that may impair your ability to properly parent your child. It has been shown that home environment has a direct impact on a child's development. Controlling your Phe levels may help you provide your child with the stimulating environment that helps a child thrive[71,72]. Diet for life is recommended and important for your health and may help with your ability to parent.

Increased Phe levels are associated with:

- Lower IQ
- Thought disorders
- Mood disorders, like depression
- Learning difficulties
- Mental processing issues
- Personality disorders
- Anxiety
- Behavioral problems[14]

You may also be concerned about the potential for Maternal PKU Syndrome, and whether your baby has PKU. Newborn screening will be done to determine whether your child has PKU. Even if your baby does have PKU, he or she can still be breastfed (see Chapter 4 for more information on breastfeeding and managing PKU for an infant).



Mothers with PKU can breastfeed their child. If you maintain your diet, breastfeeding will not result in your baby being exposed to high Phe levels. In fact, breastfeeding may even keep your Phe levels a little lower.

Working closely with your PKU team to ensure that your Phe levels remain controlled is important, and if your baby has PKU, that his or her Phe levels are also controlled. Your PKU team is there to support and guide you, especially at this important time in your life.

[71]Waisbren SE, Hanley W, Levy HL, Shifrin H, Allred E, Azen C, Chang PN, Cipcic-Schmidt S, de la Cruz F, Hall R, Matalon R, Nanson J, Rouse B, Trefz F, Koch R. *Outcome at age 4 years in offspring of women with maternal phenylketonuria: the Maternal PKU Collaborative Study.* JAMA 2000:756-62

[72]Waisbren SE, Chan P, Levy HL, Shifrin H, Allred E, Azen C, de la Cruz F, Hanley W, Koch R, Matalon R, Rouse B. *Neonatal neurological assessment of offspring in maternal phenylketonuria.* J Inherit Metab Dis. 1998:39-48

[14] American Academy of Pediatrics Committee on Genetics Policy Statement: maternal phenylketonuria. Pediatrics 122 (2008) 445-449

Chapter 10: Dining Out and Other Tips

Eating out and following a restricted diet may seem like two things that do not go well together. But dining out with PKU can be enjoyable and easy if you take the time to prepare in advance.

Working with Restaurants

Form a Relationship

If you have a local restaurant you go to often and really enjoy, get to know the management and staff. Many times, people in the food business also want to get to know their clientele and understand their special diets. If you speak openly about having PKU, you may be surprised to learn that the owner has a cousin or a family member with similar dietary restrictions. A restaurant may have no problem with changing or modifying a menu item to meet your needs. They may even let you bring your own low protein bread or pasta for them to prepare to enhance your dining experience[73].

To create a positive dining out experience:

- Research the type of restaurant where you will be dining.
- Gain knowledge of menu options prior to your arrival. Many restaurants have menus online.
- Feel free to call ahead with a question about what is available. Be clear about your dietary needs.
- Some places may allow you to bring in your own low protein pasta or bread.

Ask Questions

Don't be shy! Ask your waiter or waitress questions about ingredients used. Tell them that you need to limit protein. Remember that it is their job to make your dining experience as pleasurable as possible. They should be able to inform you of all ingredients (and especially hidden ingredients) in your desired dish. If they do not know the answer or seem unsure, politely ask them to consult the chef since accurate information is important to your special diet[74].

Watch Out for Hidden Ingredients

In the chart on the right are some items that contain protein due to a hidden ingredient that makes up the final product. If you are not 100% sure, you may want to ask the server if these hidden ingredients are present. Stating that you have a 'food allergy', which is a common occurrence, will assure an accurate answer[75].

Menu Item	Hidden Ingredient
Worcestershire Sauce →	Anchovies
Miso Paste →	Soybeans
Tahini Sauce →	Sesame Seeds
Caesar Salad Dressing →	Anchovies
Soup Broth & Gravies →	Chicken & Meat
Soy Sauce →	Soybeans

[73]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:35
[74]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:32
[75]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:32

Dining Out and Other Tips

Create Your Own Dish

A good restaurant is a customer-friendly restaurant. Since you are free to ask questions, request a custom entrée that is suitable for your diet. If they have the ingredients in house, they may be able to make you something not on the menu. Chefs are food professionals and may enjoy the chance to be a bit more creative and make a special low protein entrée[76].

Request Sauces and/or Gravies on the Side

If you are not sure about a sauce or topping, request that item on the side. Sometimes a little bit of sauce, gravy, and/or dressing can go a long way. Since you need some phenylalanine in your diet, a small amount of a "High-Phe" item can allow you to enjoy the flavor or theme of a dish. Remember you still must keep track and control the amount of Phe you take in[4].

Suggestions When Dining Out

Dining out is a wonderful social experience. Following a restricted diet does not mean you must avoid the experience of dining out. Choosing a place to dine, with PKU, just requires a little advanced planning[77]. Many restaurants provide their customers with special dietary needs with resources. Check the internet before going to see if the restaurant you're visiting has information for PKU customers.

Types of Restaurants to Try
Note that the Phe content listed below may change. Please use this only as a general guide and check with the restaurant at the time of your visit.

American
Visiting your local diner or other establishments offering typical American fare may offer the most menu variety and ease of ordering.

Try: baked potato, coleslaw, mixed vegetables

Boston Market

1 serving green beans	1.6 g protein / 46 Phe
1 serving coleslaw	1.8 g protein / 39 Phe
1 serving cinnamon apples	0.4 g protein / 15 Phe

Vegetarian
Adhering to the PKU diet is similar to being a vegetarian, but there are key differences. Many traditional vegetarian items are rich in cheese and/or soy products. These items are high in phenylalanine and natural protein, therefore, not a part of the PKU diet plan. Typical vegetarian items to watch out for and avoid include cheese, tofu, and other soy products[78].

[76]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:32
[77]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:36
[78]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:35

Dining Out and Other Tips

A vegetarian restaurant can be a new and exciting place for you to dine. Be sure to ask your server about what dishes would be most suitable for the PKU diet. Vegetarian protein is soy based and very high in Phe. Be sure to ask questions. Here you may find your server to be very knowledgeable about the food and ingredients and your dietary needs[79].

Try: Stir fried green beans or egg-plant, mixed steamed vegetables

Au Bon Pain

12 oz carrot ginger soup ..... 1 g protein / 56 Phe
12 oz garden vegetable soup..... 3 g protein / 89 Phe

Buffets

Buffets can be an ideal dining situation for anyone with PKU. You can select from a variety of healthy choices and customize them to fit your needs in controllable portions. Even the most diligent person following a PKU diet can overindulge at a buffet, so be careful. Buffets are an all-you-care to -eat, not an all-you-can-eat environment[80].

Try: theme salad, fruit , mashed potatoes

Old Country Buffet

1 serving spoon sautéed zucchini .....1 g protein
1 serving spoon candied yams .....1 g protein
1 serving spoon grilled vegetables .....1 g protein

Mexican

Bring your own low protein tortilla, or order a corn tortilla to enjoy at a Mexican restaurant to enjoy a variety of tasty food! The different toppings like lettuce, tomato, salsa, sautéed greens, guacamole and even a little bit of sour cream can be a nice change.

Try: Try: vegetable fajitas with low protein or corn tortilla or Spanish rice

Chipotle

1 crispy taco shell .....1 g protein
1 soft taco shell ..... 1.5 protein
2.5 oz fajita vegetables .....1 g protein
3.5 oz guacamole.....2 g protein
1 serving fresh tomato/green tomatillo salsa .....1 g protein

[78]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:35
[79]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:36
[80] Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:36

Dining Out and Other Tips

Japanese
Japanese restaurants tend to prepare fresh entrees to order and have an abundant selection of fresh fruits and vegetables. This can allow for a great dining out adventure. Many menu items are naturally low in phenylalanine, and they may provide a a good chance to try something new. Be creative and talk to your server[81].

Italian
Italian restaurants are abundant. Optimize your dining experience by asking questions and making special requests. Italian dishes often have a lot of sauce and cheese so ask for them on the side. Bringing you own low protein pasta and/or bread may increase you menu choices[82].

Try: vegetable based sushi rolls: cucumber, avocado, mushroom, squash

Souplantation & Sweet Tomatoes

½ cup carrot/raisin salad.....1 g protein
½ cup pineapple/coconut salad.....1 g protein
½ cup steamed veggies with lemon butter.....1 g protein

Chinese
Take out or eat in, Chinese restaurants are known for using a variety of fresh vegetables and cooking food to order. There are many options that will fit into the PKU diet. Be sure to ask about the ingredients in the sauce to rule out hidden Phe in a soy or meat-based sauce. Requesting a light sauce or sauce on the side can increase options and the size of your portion[83].

Try: low protein pasta with broccoli/ garlic oil, cold grilled vegetable antipasto, vegetable du jour vegetables

Panda Express

Side of mixed veggies.....2 g protein / 80 Phe
1 serving mandarin/sweet & sour sauce.....0.1 g protein / 3 Phe
1 fortune cookie.....1 g protein / 25 Phe

To feel more satisfied with your dining out experience:

- Drink a serving of your formula before going out – the Phe free protein in your formula helps with satiety or making you "feel full".
- Eat a low protein snack such as fruit, vegetables, low protein bread or low protein crackers.
- Drink plenty of fluids before going out and while you are at the restaurant.

[81]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey: Applied Nutrition Corp. 2007:37.
[82]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey: Applied Nutrition Corp. 2007:38.
[83]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management*. New Jersey: Applied Nutrition Corp. 2007:38.

Dining Out and Other Tips

Beverages

Whenever you go to a restaurant, you will have something to drink. Many may choose only to have water with their meal, but many people choose something else. By now you probably know that regular soft drinks are Phe free – but don't forget they are not calorie free. Make sure you drink these in moderation.

Also look for the soft drinks that are sweetened with Splenda (sucralose) which is a phe free sweetener. Coke sweetened with Splenda, Pepsi One and Diet Rite in all flavors are diet soft drinks that are sweetened with Splenda.

Coffee/Tea
Many people enjoy drinking coffee at home and at restaurants and coffee shops. Most places have low Phe options such as regular coffee, tea, apple cider and fruit juices. Be aware of what is added to drinks such as milk, soy milk, half and half, cream. These will add Phe to the beverage.

While many of the flavor syrups are Phe free, be aware that some of the sugar free syrups are sweetened with aspartame, but some are sweetened with Splenda (sucralose) which is Phe free. Always ask before adding anything to your drink.

Alcohol[84]

Some beer and mixers contain significant amounts of phenylalanine. If you choose to consume these types of beverages you need to account for this in your total allotment of phenylalanine for the day. Both alcohol consumption and high phenylalanine levels impair your judgment. Even if you choose alcoholic beverages that are low in phenylalanine, impaired judgment can lead to poor food choices and overeating resulting in elevated blood phenylalanine levels. Moderation is the key.

How much Phe is in beer?

One 12 fl oz can of beer = about 40mg of Phe! (The darker the beer the higher the Phe.)

For Phe content in other alcoholic beverages, look in the low protein food list for PKU.

[84]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey : Applied Nutrition Corp. 2007:37.

Dining Out and Other Tips

Special Considerations
Sweeteners and chewing gum

Sweeteners are used as an alternative to sugar, but while some are PKU-friendly, aspartame is not. *"Phenylketonurics: Contains Phenylalanine"* means this is likely a product you cannot eat or drink. Aspartame is an artificial sweetener that contains Phe; it is commonly known as NutraSweet™ or the brand name Equal® and is found in diet sodas and some reduced-sugar foods and beverages. A warning for people with PKU is listed on products that contain aspartame, but you must look carefully as the warning is often printed in small type.

Phe free Sweeteners
- Stevia
- Splenda (sucralose)
- Honey
- Corn Syrup
- Sugar
- Saccharin
- Sugar alcohols

Chewing Gum
Below is a list of chewing gum from the Wrigley company. Each stick of gum contains 2-3mg of Phe.
- Extra Winter Fresh
- Orbit
- Eclipse
- Big Red
- Double Mint
- Hubba Bubba
- Spearmint
- Juicy Fruit

Some gums contain aspartame in small amounts. Based on the small amount of Phe in these products some individuals are able to include these in their diet. Check labels carefully to be sure the product you choose is PKU-friendly. Below is a list of Phe free sweeteners.

Chapter 11: Traveling

A PKU Perspective

"There is no reason to let having PKU ever hold you back from going where you want to go and doing what you want to do. Traveling, be it by air or any other mode of transportation, opens new horizons and literally lets you look at things from a different perspective that should not be missed simply because you have PKU."

Traveling

Whether or not you have PKU, preparation and planning are important when you travel away from home. No matter if you are traveling near or far, you will find it easier to manage your diet and treatment if you do some research on the kinds of foods and resources you will have access to while traveling and once you reach your destination.

Air Travel

Going on a plane for a trip can make some people excited and others a little nervous or both. Having PKU should not stop you from traveling by plane. But it does mean you need to plan ahead and keep a few extra things in mind.

Flight delays are beyond your control. For this reason it is important to always pack your formula in your carry-on bag. Do not mix with liquid until past the security screening checkpoint. If you are traveling for an extended period of time, be sure to bring at least a week's worth of formula in your carry-on, just in case your luggage gets lost or stolen[85].

Have you packed these items?

- Medical formula
- A mixing container
- Scoops and measuring cups, if used
- Low protein foods
- A scale, if needed
- Food for your trip
- Your prescription
- Insurance card
- A medical foods necessity letter from your doctor

Always bring a travel letter from your clinic explaining medical necessity when you travel, especially when you are traveling internationally. This letter should list the name of the PKU-related products you are carrying with you in your luggage, state why these products are needed, and who will be using them. If you have to carry liquid formula, such as for an infant, this letter should specifically mention this. It will help you handle any questions that may arise at security. A sample letter of medical necessity can be found in the resource chapter.

For ease of getting through security, it is also helpful to keep your medical formula in its original sealed containers (packages, cans or sachets) and take a copy of your prescription with you. If you are traveling domestically, you can also ship your medical foods to your destination in advance to reduce the weight of your luggage. Be sure to allow enough time for transit should there be any delays in shipping.

[85]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey : Applied Nutrition Corp. 2007:44.

Traveling

Most domestic flights charge a fee for in-flight meals, and the foods are generally not PKU friendly. Be sure to pack (or purchase prior to boarding) any food or snacks you think you might need during the flight. Most international flights still offer meal service, so be sure to specify your need for a special meal when booking your international ticket. You may find that the only PKU-friendly meal option is a vegetable plate, salad or fruit plate (as even meals listed as low protein or vegetarian/vegan may not be low enough in protein for the PKU diet). Take plenty of snacks for the flight to cover the possibility of travel delays.

Call Hotels in Advance

To make your stay easier, call your hotel in advance to speak with management about your needs. Many hotels have rooms with a kitchenette where you can keep and prepare foods. Some hotels offer small refrigerators in each room where food or medical formula can be stored, but it is best to check prior to travel. If the hotel where you will be staying does not have kitchen facilities or a refrigerator, ask about having a mini-refrigerator brought to your room for use during your stay if you need one.

PKU Traveler's Tips

When traveling, you may also find these tips to be helpful.

- If you are traveling overseas, ask your PKU team for information on where low protein supplies can be obtained in your destination country.
- If you are traveling on an organized tour overseas, ask your dietitian for a letter explaining your diet.
- Many hotel chains offer a free continental breakfast. In addition to coffee, fruit and cereal, many have a 'make your own' Belgian waffle option. If you bring low protein powdered pancake mix with you, you can easily make your own low protein waffle at the hotel. Just call ahead and ask.
- Disney World is also PKU-friendly as long as you call ahead to let them know you're coming and what your dietary needs are.
- If you are shipping your formula or food to a hotel to arrive prior to your arrival, be sure to label the box clearly with your name and arrival date on the package. Call and alert the hotel that a shipment will be arriving for you.

Chapter 12 - Emergency Preparedness

How to Prepare Yourself for an Emergency

When you have PKU or any other special dietary needs, being ready to handle difficult situations requires special planning, in addition to the typical things everyone needs to consider in the event of an emergency or natural disaster. Here are some key things to think about and discuss with your family today[86].

Hospitalization[87]

If you need to be admitted to a hospital, you will need to bring your formula (and anything you mix into your formula) with you. If you are admitted for an extended period of time, you will also need to bring some low protein foods or have someone bring them for you.

Medical professionals you meet in a hospital or acute care setting who do not regularly work with PKU may not know how to manage your diet. They will need to be educated on your diet and the items that are appropriate for you.

Be sure to notify your metabolic team if you are in the hospital so they can aid in obtaining the correct care you need. They will be able to alter your dietary prescription, if needed, in response to your condition and direct the hospital staff as necessary.

And remember, some over the counter medicines may contain aspartame or gelatins, which contain PHE. Be sure to check the labeling on all over-the-counter and non-prescription medications such as vitamins, aspirin and cough syrups.

Emergencies

People with PKU require specialized care and medical foods, both of which may be unavailable or in limited supply in the event of an emergency.

You may not be able to follow your PKU diet perfectly during an emergency but it is important to adhere to it as closely as possible and continue to take your formula! Almost by definition, an emergency is something that brings added confusion and stress. Not following your diet can affect your ability to deal with and manage the situation at hand effectively[88].

Some emergencies may mean you can't obtain what you need for your PKU diet in the normal way. You may be able to stay in your home during the emergency, but you may not have electrical power. Other emergencies require evacuation from your home to a location that may be far from your PKU team or pharmacy.

It is important to prepare for both of these possibilities BEFORE THEY HAPPEN.

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[86]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:48.
[87]Maltzman, S, PKU & Your Life : *My PKU Toolkit A Transition Guide to Adult PKU Management.* New Jersey: Applied Nutrition Corp. 2007:49.
[88]Southeast NBS & Genetics Collaborative Region 3 Genetics Collaborative Emergency Management *Strategic Plan: An Approach to Emergency Preparedness for the Medical Genetics Community.* May 2008:3.

Emergency Preparedness

In the Event of Evacuation[89]

It is important to create an evacuation plan to use in case of an emergency evacuation. This plan should include details on when and how to evacuate, where to evacuate, and the preparation of an evacuation kit. If you have to evacuate, you may have to be away from your home longer than a few days. Even if you are able to stay in your home during an emergency situation, you may not be able to obtain what you need for your PKU diet in the normal way.

It is safer to plan for at least a 2 week emergency or evacuation period.

When evacuating, it is also helpful to have access to your medical records so that you can establish PKU care in a new location. Ask your PKU team how to access your medical records in the event of an emergency and keep a copy of your formula prescription, most recent lab and test results in your wallet or evacuation kit[90].

Your PKU team may also have to evacuate during an emergency. Be sure to ask someone on the team how you can reach team members in the event of an emergency that requires them to be away from their offices.

Creating an Emergency Plan Ahead of Time

In preparing for an emergency you should create an emergency response plan for you and your family[91]. It is important to do the following things before an emergency situation occurs.

- Decide where you will go if you need to evacuate during an emergency.
- Remember that friends or relatives who live in the same neighborhood will also be evacuating and choose a place a safe distance from your town.
- Remember you may need to be gone for more than a few days. Either choose a place where you know you and your family can stay for an extended time, or have a second place in mind if the evacuation time is extended.
- If you have pets you will be taking with you, choose a place where they will be accepted.
- Tell your PKU team the place where you think you will be evacuating to and a phone number (cell phone if possible) where you can be reached.
- Keep a copy of your formula prescription, recent lab and test results.
- Ask your PKU team where there are genetics programs or other health care providers close to the location where you plan to evacuate.
- Create an extra supply of medicine, low protein food, and medical formula to last at least two weeks. Your PKU team may need to help you with prescriptions for an emergency supply.
- Put together an Emergency Kit. You may want to include additional things, but this kit should include:
    - Medication and medical formula (at least a two week supply)
    - Low protein foods (at least a two week supply)

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[89]Southeast NBS & Genetics Collaborative Region 3 Genetics Collaborative Emergency Management Strategic Plan: *An Approach to Emergency Preparedness for the Medical Genetics Community*. May 2008:14-15.
[90]Southeast NBS & Genetics Collaborative Region 3 Genetics Collaborative Emergency Management Strategic Plan: *An Approach to Emergency Preparedness for the Medical Genetics Community*. May 2008:18.
[91]Southeast NBS & Genetics Collaborative Region 3 Genetics Collaborative Emergency Management Strategic Plan: *An Approach to Emergency Preparedness for the Medical Genetics Community*. May 2008:31.

  - Contact information for the companies that supply your medication, medical formula, and low protein foods
  - A list of PKU resources in the community you would evacuate to
  - Contact list for family members
  - Emergency contact numbers for your PKU team
  - Water and snacks appropriate for car travel
  - Manual can opener
  - Blankets and pillows
  - Maps and flashlight
  - Cell phone
  - Extra batteries
  - Chargers for all electronic devices
- You may want to consider buying a small hot plate or similar appliance that can be used to prepare food in a hotel room.

During some emergencies you may not have to leave your home – but you may lose electricity or even water supply. It is important to plan how you will follow your PKU diet for this kind of emergency as well. In addition to the things listed above, it is helpful to have:

- Bottled water – enough for several days (estimate one gallon a day for each family member).
- Some low protein foods that don't require cooking or refrigeration.
- A camp stove (that uses propane gas) that can be used to prepare food without electricity, and a manual can opener.

Following Your Emergency Plan

If an emergency occurs you should be ready to immediately follow the emergency or evacuation plan you have put together.

- If you have warning that there will be an emergency (for example, a hurricane is predicted for your area), begin preparing to follow your emergency plan as soon as possible.
- After an emergency situation occurs, make contact with your PKU team as soon as possible.
- Your PKU team may not have access to your diet information, so be prepared to tell them:
  - Your daily Phe restriction
  - What medical formula you use and how much each day
  - If you take PKU medication – what you use and how much each day
  - How long your supply for each of these things will last
- You may need to contact the companies who supply your medication, medical formula, or low protein foods and tell them your new contact information if you do not have enough supply to last until you can return home or to your usual way of obtaining these things.
- You may need to contact the PKU team and/or other health care providers in your new location to continue your PKU care.

Chapter 13: Tracking Your Progress

Sample Diet Record

Instruction for filling out a diet record:

- **Date/Time**: Record Date and time the food/drink was consumed.
- **Food or Liquid Consumed:** Record the name of the food or liquid (including formula) and be specific as possible (apple vs. fruit, include brand names if possible).
- **Measured Amount Eaten:** Record specific measurements like grams, tablespoons, tea spoons, cups. If a few bites were eaten, record as "3 bites". The amount is just as important as the type of food eaten.
- **Milligrams of Phe:** Look up the amount of Phe in your food reference guide and record the amount of Phe that is accurate to the amount of the food eaten.
- **Grams of Protein and Calories:** This is to record the amount of protein and calories that are in foods. This is to be recorded if this information is available.
- **Daily Totals:** Record the total Phe, Protein and Calories (when available) consumed that day.

It may be helpful to add up the Phe as you go so that you can see the total that has been consumed so far for that day. This will help you figure out how much more Phe can be eaten that day to stay within the limit. Remember to weigh or measure every food and drink item! Below is an example of a diet record already filled out with actual information followed by a blank record for you to make copies of this blank record for your use.

DATE/TIME	FOOD OR LIQUID OFFERED	MEASURED AMOUNT EATEN	MGS PHE	GRAMS PROTEIN	CALORIES
5/24/11 (7am)	Trix cereal	20grams	46		
	Phenex 2	8ounces	—		
	Del-Monte diced pears	1container (113g)	7		
(11am)	Ener-G rice starch bread	1slice	7		
	sandwich-melt cheese	1slice	24		
	butter	1 Tablespoon	6		
	Lay's potato chips	1oz	93		
	apple (medium)	1	11		
(3pm)	Phenex 2	8ounces	—		
(6pm)	Aproten (fusilli)	1cup (dry)	19		
	Hunts marinara	1/4 cup	≈70		
	Iceburg lettuce	1cup	14		
	Italian dressing	2tablespoons	-		
	Phenex 2	8ounces	—		
			247 phe		

Chapter 13: Tracking Your Progress

PKU DIET DIARY

Name: _____ **Birth date:** _____

Date/Time of Specimen:_____/_____am___pm___ **Time of Last Meal:** _____am___pm___

Recorded by: _____ Weight_____Height_____

Phe/Protein Prescription: _____ **mgs Phe/grams of protein per day**

Other Diet Modifications (if any): _____

Vitamins or minerals taken (if any): Kind _____Amount _____

Kind _____Amount _____

How is Formula Mixed? (Please specify what product is used.)

Amount: _____ (# grams/packets/scoops/tbsp/cups): **Formula Name:**_____

Amount: _____ (# grams/packets/scoops/tbsp/cups): **Formula Name:**_____

Amount: _____ (# grams/packets/scoops/tbsp/cups): **Formula Name:**_____

Add water/ juice (kind)_____**to make a total volume of**_____**ounces.**

Kuvan: Current dose_____tabs per day N/A

Any missed doses? No Yes, _____

Appetite: Poor_____Usual_____Better than usual_____

Any illness?: Yes_____ No_____ Date/s:_____

Was medication required? Yes_____No_____What was thermometer reading?_____

Vomited food or formula? Yes_____No_____Diarrhea? Yes_____ No_____

Describe Illness/Other Comments: _____

_____

Please record food/formula consumed for 3 consecutive days prior to obtaining a blood sample or prior to a clinic visit.

Tracking Your Progress

	PHE/PROTEIN INTAKE	FORMULA INTAKE
Day 1		
Day 2		
Day 3		
Average		

DATE/TIME	FOOD OR LIQUID OFFERED	MEASURED AMOUNT EATEN	MSG PHE	GRAMS PROTEIN	CALORIES

Tracking Your Progress

Sample Phe Log

When you take a blood sample and send it to a laboratory for testing, your PKU team will receive the result. When you get the results from your clinic, you should record them in a Phe Log, like the one below. Record your Phe level in mg/dL or µmol/L. Record the date, Phe/protein intake, medical formula intake and Kuvan intake from the date the sample was taken (not the date you receive the results).

DATE	BLOOD PHE MG/DL OR UMOL/L	PHE/PROTEIN INTAKE	MEDICAL FORMULA INTAKE	KUVAN INTAKE	RECOMMENDED DIET CHANGES

Chapter 14: Insurance Coverage for PKU Treatment

Insurance Overview

Medical foods (formula and foods modified to be low in protein) are a medical necessity for people with PKU. However, many adults and families face challenges in obtaining coverage from their health insurance.

Coverage for PKU-related medical foods varies from state to state, although 38 states have passed legislation that requires at least some coverage. Some states have passed legislation to mandate insurance coverage for medical foods, while others provide medical formula and some low protein foods directly to PKU patients through newborn screening or state health department programs.

To find information about legislation around insurance coverage of medical foods in your state, visit our state coverage resource tool at: http://npkua.org/index.php/state-coverage-for-pku[92].

If you are facing insurance denials, the National PKU Alliance may be able to help with its newly launched insurance ombudsman project. To learn more, visit http://npkua.org. Here you find additional information on the insurance appeals rights, your rights under the recently passed Affordable Care Act, as well as how to request a volunteer advocate to assist you with your cover issues. The first step to understanding your coverage for medical foods is to have a basic understanding of your health insurance plan.

Questions to Consider When Choosing a Health Insurance Plan

Many employers change health insurance companies on a frequent basis. This typically happens with the start of the benefits year. If you are facing this situation, it is critical that you be pro-active and work with your employer to try to ensure coverage of medical foods for PKU.

Questions to ask when your company informs you that they will switch health insurance companies:

- Who is the first point of contact for questions?
  - Your employer's HR Manager
  - A Benefits Consultant/Advisor hired by your employer, if offered
  - Toll free phone number for the insurance company
- Is the plan insured or self funded?
  - Under an insured plan, the employer purchases commercial health care coverage from an insurance company, and the insurance company assumes the risk for payment of claims. Insured plans are regulated by the individual states. If the plan is self funded, (often times called a "self-insured" plan), the employer keeps the risk to pay the bills and usually hires a plan administrator to process the claims. This differentiation is important because self funded plans are not subject to state insurance laws or regulations – a state cannot require a self-funded employer to cover PKU medical foods (formula and foods modified to be low in protein).
  - The best way to determine if the plan is self-funded is to ask the employer or call the plan. Generally, most very large employers and union plans self-funded.

[92] National PKU Alliance. "State Coverage for PKU." http://npkua.org/index.php/state-coverage-for-pku

Insurance Coverage for PKU Treatment

- Another way to determine if the plan is insured or self funded is to be aware of the documentation you receive. If your employer has an insurance policy, the plan is insured. If they have a plan document, the plan is self funded.
- Be aware that many self-funded plans use insurance companies to process their claims and perform other administrative duties. To further the confusion, the insurance company may also "rent" their provider network to the plan, so a self-funded plan may look very much like an insured plan.
- Sometimes self-funded plans are called "ERISA plans." ERISA is a very broad federal law that also regulates employer health plans. Don't be misled by this term - virtually ALL employer plans, whether they are insured or self-funded, are regulated by ERISA and technically are "ERISA plans." But generally, if a plan is called an "ERISA plan" it is probably self-funded.
- Another good way to determine if the plan is insured or self-funded is to read the documentation you receive. It may tell you directly if the plan is insured or self-funded.
- In addition, all ERISA-regulated plans are required to provide certain information to plan participants, but many employers with insured plans do not fully comply with this rule. The plan sponsor (usually the employer) is responsible for this, not the insurance company. Thus, if the employer has an insured plan, you may not see federally required information in the plan documents – typically missing would be the name of the "plan administrator", the designation of the plan's fiduciary, the plan year and plan number (used for federal reporting), and the plan name. If this information is missing, it's probably an insured plan.
    - Often your first point of contact at an insurance company will not know if there are state mandates. For example, when they first run a query, they will often get a message back stating that the formula is not covered because it is available over the counter. This can be disproven with just a few documents.
    - It may be necessary to submit your state's insurance coverage laws to the insurance company's appeals department in writing after your coverage begins. Current laws can be found on the NPKUA website at www.npkua.org under the legislation tab.
    - Be aware that private insurance companies may have different rules.
        - If your company opts for a self-insured plan, ask your company to work with you to ensure that formula is covered. Self- insured plans can design their own coverage spectrum and are not obligated to follow state mandates for coverage.
- Understand what benefit category medical foods fall under. Typically medical foods are covered either as a pharmacy product or a medical product. Some insurers may provide coverage for the formula, but not the foods modified to be low in protein.
    - Pharmacy benefit – your medical foods will be covered like a prescription drug and you will need to pay a monthly co-pay.
    - Medical benefit – your medical foods will be covered like Durable Medical Equipment (DME). The cost may be $0 per month or a percentage to be determined, after the deductible is met.
- In some instances the insured will pay out a percentage of the cost and be reimbursed.
- Some plans also establish annual benefit maximums.

Insurance Coverage for PKU Treatment

- What choices are available for plans? Does one meet your needs better than the others?

o Three plans are often offered:
  - HMO – (Health Maintenance Organization) plans are often the most restricted and some may not cover medical foods.
  - POS - (Point of Service) plans usually provide good coverage but with limited choice of providers.
  - PPO - (Preferred Provider Organization) plans are sometimes the least restrictive in terms of both coverage and choices.

It's important that you do the math. Find out the contribution to the premium for each plan. This contribution is the amount of money you pay to an insurance company for insurance coverage. It is important to note that even if a certain plan has a lower contribution (meaning you will be paying less each month for your insurance coverage), it may mean you have to pay more out of pocket for medical services. So if you are going to be accessing healthcare often, it may be worthwhile to pay a higher premium up front to get better coverage later when you need it most. Consider the following scenario:

- Standard Plan- Biweekly premium $100 or $200/mo. Benefits pay 50% of medical food costs. Medical formula is $1200/mo. You would pay a total of $800/mo ($200 for insurance and $600 for formula) under this plan.

- Premium Plan- Biweekly premium $200 or $400/mo. Benefits pay 80% of medical food costs. Medical formula is $1200/mo. You would pay $640/mo.($400 for insurance and $240 for formula) under this plan.

  - o If an itemized benefits package is not handed out, request one and read it over. Pay close attention to sections relating to the needs of PKU: pharmacy copays, Durable Medical Equipment coverage, and specialist doctors. If it is unclear you can ask your first point of contact for clarification or more details.
  - o If none of the plans meet your needs, it doesn't hurt to ask for more options. It is not unheard of for an HR Manager in a small company to make changes to better meet the needs of the employees.
  - o Find out if there is a Flexible Spending Account available. Your deductible and copays can be tax-free if planned out in advance. Keep in mind that these accounts do not roll over into the next year.
  - o If your needs allow, you may find it beneficial to opt in to a Health Savings Account. These are similar to a Flexible spending Account in that they are not taxed, but may require you to choose a very high deductible plan. A benefit is that they do roll over at the end of the year. A Health Reimbursement Plan is similarly not taxed, but may not roll over year to year.
  - o Know your expenses. Keep a record of how much you spend on PKU products per month and per year to help you determine which plan fits your needs best.

- Is your preferred PKU clinic in-network?
  - o If not, contact your clinic and ask if they can try to join the network, or if they already have a plan for such situations. You can also ask the plan for an "out of network" exception.

Insurance Coverage for PKU Treatment

Documentation to have available:

- Soft and hard copy of your state's legislation regarding coverage for medical foods.
- Proof of prior coverage (will be sent by previous insurance company within 30 days of cancelation of coverage).
- Contact information for your previous insurance company, as well as your old group number, individual ID number, and toll free phone number (ID card or copy of both sides).
- Copy of your prescription for medical foods.
- Full name of your formula and manufacturer contact information.
- PKU quick facts sheet: description, diagnosis, symptoms, treatment, etc.
- Send your clinic your new health insurance information as soon as possible. Your new insurance company may require correspondence with your clinic to approve your formula coverage. It is much easier to get assistance from your clinic if they have had time to prepare for the near-inevitable request for Letters of Medical Necessity (see Appendix A).

Other Tips

- Your HR Manager or benefits consultant may request a list of your diagnoses and treatments, including all prescription products and procedures required. This helps them determine how to help you get the best coverage option. If they don't ask for it you may offer it.
- The internet and search engines can be a valuable research tool to find information about health insurance companies, look up terminology, and reach out to your PKU communities for support.

Understanding Your Current Health Insurance Plan

In order to learn about your current coverage, or if you receive a denial for medical foods coverage, you need to get a copy of your master insurance policy (for insured plans) or the plan document (for self funded plans) and read it. If you have a self funded plan, ERISA requires that the plan administrator (generally the employer) provide this documentation within 30 days of a request. Many employers don't know they are required to do this and will often tell employees that they don't have anything. Sometimes people think they have 100% coverage for all prescriptions, services and medical foods, just because they have insurance. This is not true[93]. It's important to read your coverage document carefully.

- Find out if medical foods for PKU are a covered benefit and if you will be responsible for any co-payments, co-insurance and/or other deductibles[94].
- If medical foods are covered, you need to learn whether they are a medical benefit or a prescription benefit. This determines who will supply the formula. If your formula is covered under the prescription part of your benefits, then you can have your prescription filled at an approved pharmacy. If your medical food is covered under the medical part of your benefits then you will need to have a medical supply company provide the formula. To find an "In-Network" pharmacy or medical supply company, ask the insurance representative for a provider list or contact your metabolic team and/or the formula manufacturer[95].
- If you are a federal employee and have insurance through the Federal Health

[93]From My PKU Toolkit, Applied Nutrition Corporation, 2007, p. 50.
[94]From My PKU Toolkit, Applied Nutrition Corporation, 2007, p. 50.

Employee Benefits (FEHB), medical formula is normally covered for children up to the age of 26. Foods modified to be low in protein are normally not covered. In addition, it's important to know that federal plans do not have to follow any state or most federal mandates for coverage.

- If you are on Medicaid, your formula should be covered and foods modified to be low in protein should be covered as part of the Early and Periodic Screening, Diagnostic and Treatment program as a health benefit under medical supplies.
- Contact your HR manager at work to learn if your health insurance plan is a self-insured plan. This will help you determine whether or not your plan has to follow any mandates that exist in your state regarding coverage for medical foods. ERISA prevents many adults and families from getting coverage for medical foods despite these state laws. Self funded plans can design their own coverage spectrum. In many cases, these plans choose to exclude medical foods for the treatment of PKU from their coverage. Many companies provide a self funded plan to their employees because it costs less to do so. If your employer moves from an insurance plan to a self-funded plan, they will often maintain the same benefits. They will often not remove the coverage until they find out they have large claims, but it is a violation of The Health Insurance Portability and Accountability Act (HIPAA) to remove benefits for a particular condition that a participant is being treated for.
- You still have a right to appeal a denial for medical foods coverage if you have a self funded plan. See the section on Appeals Process for more information.

Letter of Medical Necessity

In order to obtain insurance coverage or appeal a denial, you will need a letter of medical necessity from your doctor. The letter should detail your specific needs for medical foods and laboratory coverage as treatment for your PKU condition. It should also clearly state why these treatments are necessary for managing your PKU. A sample letter of medical necessity can be found in the resources section.

Dealing with Your Insurance Company [96]

To get the maximum coverage for medical foods to which you are entitled, you need to contact your insurance company. Call the member/customer service number on your insurance card and ask for the benefits department to find out if a prior approval is required. Always write down the name of the person with whom you spoke, the date and any information discussed. If the representative won't give out personal information, get his/her first name, ID number or direct phone extension, at the very least.

Inform the representative of your needs. Be sure to provide him/her with the product name, description, national drug code (NDC), HCPCS code, and manufacturer name and phone number. These codes can be located by calling the manufactuer of your medical food. Explain that you need this product for the dietary management of Phenylketonuria (ICD-9/ diagnosis code: 270.1), which is an inherited disorder of metabolism and that this product is medically necessary for your treatment. Specific information in commonly used codes can be found in Appendix B.

[96]From My PKU Toolkit, Applied Nutrition Corporation, 2007, p. 51.

Insurance Coverage for PKU Treatment

Your insurer should be able to tell you if medical foods are a covered benefit and whether or not you will need prior approval for coverage. If you do need prior approval, find out where to fax or mail the copy of your state law, prescription and letter of medical necessity to obtain a prior approval number. Keep records of any fax transmittals and return receipt via mail.

Sometimes the insurance representative may not be familiar with the policies regarding medical foods. If this is the case, ask to speak with a supervisor or case manager and begin again. At this point it may be helpful to mention that there is a state law that mandates coverage (if one exists in your state).

Keep an insurance file where all of the paper work, documentation and receipts can be kept for future reference. You can also request a case manager from the insurance company. Most insurance companies provide case management services for people with chronic health issues. A case manager can prove to be very helpful in navigating the insurance benefits.

Flexible Spending Accounts

A Flexible Spending Account (FSA) is an employer-provided benefit that allows employees to set aside income from their paychecks to pay for medical expenses. The major benefit to FSA account holders is that this income is not taxed, saving both money on income taxes as well as increasing the amount of money that can be spent on medical expenses (as it is actual income, rather than taxed income).[97]

If you have medical food expenses that are not covered or partially-covered by your insurance company, an FSA can be a convenient way to save money over the course of the year from your paycheck. Usually you will determine how much money should be allocated from your paycheck into your FSA at the beginning of each year. Generally, only the cost of foods modified to be low in protein that exceed the cost of normal foods can be reimbursed.

Not every employer has an FSA benefit. Talk to your employer or benefits administrator about your employer's FSA plan and how it could work for you.

What is ERISA?

The Employee Retirement Income Security Act of 1974 (ERISA) is a federal law which is primarily concerned with pension plans. However, it also sets minimum standards for many employee benefits, including employer provided health coverage.

ERISA governs approximately 2.5 million health benefit plans sponsored by private employers nationwide. It does not apply to government and church employee plans.

ERISA plans are not subject to state insurance laws or jurisdiction. Thus, even in your state has a law requiring some sort of coverage for medical foods, if your plan is self-funded, is does not have to follow the state law. This called an ERISA exemption.

Insurance Resources

National PKU Alliance: The NPKUA works to improve the lives of individuals with PKU and pursue a cure. This tool-kit is part of our Insurance Ombudsman Initiative to

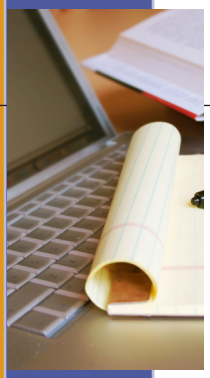
[97]FSAFeds. "What is a Flexible Spending Account?" https://www.fsafeds.com/fsafeds/summaryofbenefits.asp#WhatIsFSA

Insurance Coverage for PKU Treatment

provide information and support to adults and families struggling with insurance coverage for medical foods. A central part of this new program will be to offer support and guidance through a network of lay advocates. www.npkua.org

The Patient Advocacy Foundation: This foundation provide pro bono case management and insurance mediation assistance for those with chronic, debilitating, or life threatening diseases. They may be able
to provide assistance to PKU adult and families.
www.patientadvocate.org. 1-800-532-5274.

Nutricia Product Coverage Navigator: Nutricia's program in Massachusetts, New York and Texas provides assistance to families using their metabolic products. They can provide assistance with prior authorizations, claims submissions, medical necessity letters, billing errors and the appeal process. 1-800-356-7354, ext. 1200.



Chapter 15: Resources

Resources are available to support you in managing phenylketonuria (PKU) and its treatment. First, and most importantly, your PKU team is always available to support you as your primary support and source of information. Resources have also been developed and refined over the years to help you. Here we provide you with samples and direction to other resources that will continue to support you with managing PKU from infancy through to adulthood.

Sample Letters for School and Other Activities

The following sample letters of medical necessity should be provided by a PKU team member. He or she can prepare a customized letter for you. Keep a letter of medical necessity with you when you are travelling to ensure you can easily respond to any questions asked at airport security or customs.

SAMPLE LETTER FOR INSURANCE COVERAGE OF FOODS MODIFIED TO BE LOW IN PROTEIN

(Date)

RE: (patient name)
D.O.B: (patient date of birth)

To Whom It May Concern:

We are writing a letter of medical necessity regarding the treatment of (patient first name & last name). (patient name) has been under the consultative care of the (clinic name). He/She has an inborn error of metabolism, a genetic disorder, known as phenylketonuria (PKU, ICD 9 270.1). We are writing to request that low protein modified food products be covered by his/her current medical insurance.

PKU is a lifelong problem that requires a phenylalanine-restricted diet including low protein modified food products and the prescription of medical foods/formulas by a licensed physician with the support of a registered dietitian in order to control the blood phenylalanine level. Low protein modified food products are defined as manufactured products that will deliver no more than one gram of protein per serving. Low protein modified food products supply needed additional calories (to help prevent catabolism, which in itself can cause phenylalanine levels to rise), without supplying additional phenylalanine containing protein. Use of low protein modified food products, especially when used consistently, greatly improves adherence to the treatment program.

PKU results from a deficiency of the enzyme responsible for metabolizing the amino acid phenylalanine. This results in the build-up of phenylalanine to toxic levels. An untreated child with PKU will suffer irreversible brain damage as well as severe and progressive neurological disorders. Normal growth and development are possible if an infant with PKU is treated appropriately. In adolescents and adults, neurological deterioration, phobias, difficulty in concentration and impulse control, and loss of IQ points can occur if treatment is not sustained.

Patients are treated with prescribed medical foods/formulas, as well as a phenylalanine-restricted diet which includes low protein modified food products. This diet excludes all foods high in protein (i.e. meat, poultry, fish, dairy, nuts and legumes) and markedly restricts

all grains, including rice, breads, and pastas. Medical foods/formulas provide the primary protein constituent (80-85% of RDA protein) for the PKU dietary treatment regimen. Low protein modified food products other nutrients which includes additional calories to prevent catabolism which can cause a rise in phenylalanine levels. Use of these products is medically supervised by a physician and implemented by a registered dietitian specially trained in the nutrition management of inborn errors of metabolism. Nutrition therapy must also provide a sufficient and balanced intake of other nutrients to avoid nutritional deficiencies. Nutrition therapy of PKU solely via protein restriction is not possible, because it will result in protein malnutrition, calorie deprivation, vitamin and mineral deficiency, failure-to-thrive, and potentially death.

The standard of care for PKU requires the use of the medical food/formulas and a phenylalanine-restricted diet which includes the use of low-protein modified food products, as well as routine nutrition follow-up with a specially trained registered dietitian. The two primary goals of treatment are:

1. To maintain the blood phenylalanine at a level that is not toxic, but still allows for normal growth and development.
2. To ensure that the individual's overall nutritional requirements are met, allowing for normal growth and development, and the avoidance of nutritional deficiencies.

The recommended treatment range of blood phenylalanine levels for individuals with PKU is between 2 and 6mg/dL (120 and 360µmol/L). There is good correlation of cognitive function and maintenance of blood phenylalanine levels in this treatment range. Elevated blood phenylalanine in patients has been associated with behavior and learning problems which can reverse when the blood levels return to the treatment range. Currently, indefinite continuation of dietary management is recommended to all patients with PKU. These recommendations are based on a growing body of evidence indicating there is a decline in average IQ and development of difficulties in school performance after diet discontinuation.

We appreciate your attention to this request for (patient's name)'s low protein modified food products to be covered by his/her current medical insurance. Please do not hesitate to contact us if you have any questions at (clinic contact info).

Sincerely,

(dietitian name), RD, LDN

(Physician name), M.D.
(physician credentials, clinic name)

cc: (parents name)

Resources

SAMPLE LETTER FOR INSURANCE COVERAGE OF MEDICAL FOODS

(Date)

RE: (patient name)
D.O.B: (patient date of birth)

To Whom It May Concern:

We are writing a letter of medical necessity regarding the treatment of (patient first name & last name). (patient name) has been under the consultative care of the (clinic name). He/She has an inborn error of metabolism, a genetic disorder, known as phenylketonuria (PKU, ICD 9 270.1). We are writing to request that medical food/formula be covered by his/her current medical insurance.

PKU is a lifelong problem that requires a phenylalanine-restricted diet and the prescription of special medical foods/formulas by a licensed physician with the support of a registered dietitian in order to control the blood phenylalanine level. The term medical food/formula as defined in section 5(b) of the Orphan Drug Act {21 U.S.C. 360ee (b) (3)} is a "food which is formulated to be consumed or administered internally under the supervision of a physician and which is intended for the specific dietary management of a disease or condition for which distinctive nutritional requirements, based on recognized scientific principles are established by medical evaluation."

PKU results from a deficiency of the enzyme responsible for metabolizing the amino acid phenylalanine. This results in the build-up of phenylalanine to toxic levels. An untreated child with PKU will suffer irreversible brain damage as well as severe and progressive neurological disorders. Normal growth and development are possible if an infant with PKU is treated appropriately. In adolescents and adults, neurological deterioration, phobias, difficulty in concentration and impulse control, and loss of IQ points can occur if treatment is not sustained.

Patients are treated with prescribed medical foods/formulas (in a variety of forms powder, capsule, liquid, bar etc.), special low-protein modified food products as well as a phenylalanine-restricted diet. This diet excludes all foods high in protein (i.e. meat, poultry, fish, dairy, nuts and legumes) and markedly restricts all grains, including rice, breads, and pastas. Currently, (patient name) is prescribed (name of medical formula) which is a medical formula used to manage PKU. Medical foods/formulas provide the primary protein constituent (80-85% of RDA protein) for the PKU dietary treatment regimen. Use of these products is medically supervised by a physician and implemented by a registered dietitian specially trained in the nutrition management of inborn errors of metabolism. Nutrition therapy must also provide a sufficient and balanced intake of other nutrients to avoid nutritional deficiencies. Nutrition therapy of PKU solely via protein restriction is not possible, because it will result in protein malnutrition, calorie deprivation, vitamin and mineral deficiency, failure-to-thrive, and potentially death.

The standard of care for PKU requires the use of the medical food/formulas and a phenylalanine-restricted diet, as well as routine nutrition follow-up with a specially trained registered dietitian. The two primary goals of treatment are:

1. To maintain the blood phenylalanine at a level that is not toxic, but still allows for normal growth and development.
2. To ensure that the individual's overall nutritional requirements are met, allowing for normal growth and development, and the avoidance of nutritional deficiencies.

The recommended treatment range of blood phenylalanine levels for individuals with PKU is between 2 and 6mg/dL (120 and 360µmol/L). There is good correlation of cognitive function and maintenance of blood phenylalanine levels in this treatment range. Elevated blood phenylalanine in patients has been associated with behavior and learning problems which can reverse when the blood levels return to the treatment range. Currently, indefinite continuation of dietary management is recommended to all patients with PKU. These recommendations are based on a growing body of evidence indicating there is a decline in average IQ and development of difficulties in school performance after diet discontinuation.

We appreciate your attention to this request for (patient's name)'s medical formula, (name of medical formula) to be covered by his/her current medical insurance. Please do not hesitate to contact us if you have any questions at (clinic contact info).

Sincerely,

(dietitian name), RD, LDN

(Physician name), M.D.
(physician credentials, clinic name)

cc: (parents name)

Resources

SAMPLE LETTER FOR TRAVEL

Date: _____

RE: _____ traveling with medical formula and low protein foods

To Whom It May Concern:

My patient,_____, is a _____year old boy / girl with an inborn error of metabolism called phenylketonuria (PKU). The treatment for PKU is dietary management and restriction of dietary
phenylalanine (an amino acid) and daily consumption of a medical nutritional formula.

_____'s medical formula is a mixture of a formula called_____
For a PKU
patient, it is crucial for all this formula to be consumed daily with special low protein foods which are part of treatment for PKU.

During _____'s travels – including the flight - it is essential that he / she have access to the formula (in powder and liquid form) and specialty low protein foods. They will bring what they need with them on the plane, including the formula, and the rest will be in their checked bags. The following non-perishable formula and perishable/non-perishable food items will be brought with them during their travels:

- Name of medical formula: _____ (in powder and or liquid form)
- Low protein foods _____
- Gram scale to measure foods and formula

Please allow the family of _____to bring formula and low protein foods on the plane during their travels so that they may properly care for their child while on vacation.
_____'s medical care is coordinated by Dr._____
and the clinic staff at_____. If you have any questions

Sincerely,

_____

Dr.

Resources

Handouts

For any parent, their child's first time experiences like having dinner with friends or sleepovers can be stressful. For the parent of a child with PKU, it can be especially stressful. You may worry about whether your child will be able to eat anything, or if he or she will end up eating something that is inappropriate for a low Phe diet.

You may not be the only one. It can be stressful for a new daycare provider, teacher, parent of a friend or babysitter to know that a child in their care has a special diet.

An easy way to help put your mind at ease – and theirs – is to provide them with a handout of information. Consider giving them a copy of the "FAQs" as well, if that level of information is required, or, if your child will be with them for a meal, fill out the blank menu form that will help them prepare a simple meal that is nutritious and appropriate for your child with PKU.

Consider meeting with people personally; reassure them that you are there to provide support, especially for new daycares and schools.

Resources

PKU Information for Adult Friends of the Family

You will be in close contact with_____ who is a healthy child, but has a rare genetic disorder called phenylketonuria or PKU. This means that _____has special dietary requirements that are very important, but is otherwise very healthy and can do anything else that other children enjoy. It's important that you feel comfortable and confident that you can handle the diet, too, so here is some information you might find helpful.

Here are some key points that you may want to know:

- PKU is a genetic condition that is not contagious.
- Apart from needing a special diet, a person with PKU is healthy.
- People with PKU cannot break down an amino acid called phenylalanine (Phe), which is found in all foods containing protein.
- Phe can build up in the blood and damage the developing brain.
- Staying on a low protein diet keeps Phe levels in a safe range, allowing for normal development and a healthy life.
- Eating the wrong foods will not make a person with PKU sick right away, but will cause problems over the long-term. Having food that is not part of the diet should not be considered a "treat" as it will have implications for an individual with PKU.
- A person with PKU does not outgrow it and must stay on the diet for life.

Foods that are safe as snacks are_____ _____. It's important that we always know what_____has eaten, so please let us know if any snacks have been served, and how much. We will record this so we can make sure_____has all the nutrients needed every day. If ever _____will be over for dinner, we can send a PKU meal, or an easy low Phe menu for you to prepare. And please don't be afraid to ask any questions you may still have!

Resources

PKU Information for Friends of a Child with PKU

Hi! My name is _____. I have a disorder called phenylketonuria or PKU. This means that there are some things I can't eat – kind of like having allergies. I have a special drink that helps make sure I grow and have big muscles. You will see that I drink it a lot!

Some things you may want to know are:

- You can't catch PKU. People are born with PKU, like I was.
- Having PKU means I have to eat foods that keep me healthy.
- I can't eat some kinds of foods, like meat or cheese, or they will make me sick. I won't get sick right away, but if I eat these foods, my body and brain won't grow the way they are supposed to.
- I drink a special milk which is like a vitamin drink. It gives my body good things that come from foods I can't eat.
- Having food that I'm not allowed isn't a "treat", but there are treats that I can have – like_____!
- I will always have PKU, it won't go away, so I will always be on a special diet.
- I can run, jump, play and do anything any other kid can do!

Just so you know:

- My favorite treat is _____ .
- My favorite activity is_____ .
- My favorite toy is _____ .

In case you have any questions or want any more information, you can call:

_____at_____ .

Resources

PKU Information for Babysitters

As a babysitter for_____, you may wonder what phenylketonuria (PKU) is_____. is a healthy child, but has a rare disorder called phenylketonuria or PKU. This means that_____ is healthy, but has a special, very important diet. It's important that you understand this, so I'd like you to review some information that will help you while babysitting_____.

Here are some key points that you may want to know:

- PKU is not contagious.
- Apart from needing a special diet, a person with PKU is healthy. It's kind of like having allergies, but sometimes more serious.
- People with PKU cannot break down parts of protein – which is something found in meat, eggs, milk and a lot of other foods.
- Phe can build up in the blood and damage a person with PKU.
- Eating the wrong foods will not make a person with PKU sick right away, but will cause problems over the long-term. Having food that is not part of the diet should not be considered a "treat."
- A person with PKU does not outgrow it and must stay on the diet for life.

When we leave you with_____, we will leave out prepared meals and some foods that are safe as snacks such as_____. It's important that you do not give_____ any other food than what has been provided or approved at any time as it can make him or her sick.
It's also important that we always know what_____has eaten, and how much, so please let us know what has been eaten (or if something hasn't been eaten that we left out).

Please don't be afraid to ask any questions you may still have about_____.
It's important to us that you're comfortable and able to help us with_____'s special diet.

Resources

PKU Information for Daycare Providers and Teachers

As daycare provider for_____, you will be responsible for _____independently. This is an important responsibility, and I want to make sure you are comfortable in this role. This is some information to help you understand phenylketonuria (PKU) and how it affects my child.

_____is a healthy child. PKU is a rare genetic disorder. This means _____that has special dietary requirements that are very important, but is otherwise very healthy and can do anything else that other children enjoy.

Here are some key points that you may want to know:

- PKU is a genetic condition that is <u>not</u> contagious.
- Apart from needing a special diet, a person with PKU is healthy.
- People with PKU cannot break down an amino acid called phenylalanine (Phe), which is found in all foods containing protein.
- Phe can build up in the blood and damage the developing brain.
- Staying on a low protein diet keeps Phe levels in a safe range, allowing for normal development and a healthy life.
- Eating the wrong foods will not make a person with PKU sick right away, but will cause problems over the long-term. Having food that is not part of the diet should <u>not</u> be considered a "treat" as it will have implications for an individual with PKU.
- A person with PKU does not outgrow it and must stay on the diet for life.

We will work with you to help you learn the details about planning meals for PKU. We can also review the menus that you prepare, identify what _____can eat, and supplement it with what is required for a nutritional PKU diet.
Foods that are safe include_____. We can provide treats that are appropriate for _____so that when special events like birthdays happen, he or she will have a treat to enjoy as well, that's safe for him or her to consume.

It's important that we always know what_____ has eaten, so please let us know everything what_____has eaten, and how much. This

includes if he or she has not eaten some of the food that has been provided as we have to make up for it throughout the day. If any Phe-free snacks have been served, we need to know this, too. We will record this so we can make sure_____has all the nutrients needed every day.

Attached is a list of Frequently Asked Questions. Please don't hesitate to ask any additional questions you may have. We will work closely with you to assist with this transition in any way possible.

Sincerely,

Contact information: _____

FAQs for Adults[99]

Q. How common is phenylketonuria (PKU)?
A. Approximately one child in every 15,000 births has PKU[100]. All states have effective PKU newborn screening programs, so most children with PKU start a low protein diet within the first month of life. The diet is well-controlled to ensure that the children grow and develop normally and attend regular schools.

Q. Since PKU is inherited, do all the children in that family have PKU?
A. No, there is a 25 percent chance for other children to have PKU. For a child to have PKU, one parent must have PKU or each of the parents must be non-symptomatic carriers of the PKU gene meaning that they have one normal gene and one PKU gene. Two carrier parents have a 25 percent chance of having a baby who is free of the PKU gene, a 50 percent chance that the baby will be a carrier, and a 25 percent chance the baby will have PKU. Families with one child with PKU may have others that do not.

Q. How is the PKU diet planned?
A. The diet depends on how much Phe is allowed in the diet and how much of the medical formula the individual needs. Below is a typical low protein menu.

MEALS	FOOD ITEMS	PHENYLALANINE
Breakfast	3 CBF Mixquick pancakes	2
	2 tbsp Pancake syrup	None
	1 Apple	11
	Medical formula	None
Lunch	1 ½ cups (cooked) Loprofin Spaghetti	7
	½ cup Marinara Sauce	39
	1 cup Iceburg Lettuce	14
	2 tbsp Italian dressing	None
	Medical Formula	None
Dinner	1 PKU Perspectives Chicken-flavored patty	39
	1 CBF bun	6
	½ cup Fried potatoes	76
	2 tbsp Ketchup	18
	10 baby carrots	30
	1 Mandarin Orange	16
	Medical Formula	None
TOTALS		**258 mg(~5.2g protein)**

Q. How long will they be on this diet?
A. A person with PKU is put on a strict low Phe diet as soon as he or she is diagnosed with PKU. It is recommended that anyone with PKU remain on their strict diet for their entire life, as high Phe levels can cause health problems for a person with PKU at any age.

Q. Can a child grow with this strict diet?
A. The formula or drink contains most of the protein, vitamins and minerals needed for growth. The food eaten provides the rest of the nutrients needed, and there are "free foods" that do not contain Phe that a person with PKU can eat when he or she is hungry.

Q. Can a person with PKU have any treats?
A. Some treats a person with PKU can have include lollipops, popsicles (without ice cream) and other sugar candies.

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[99]Kaufman, M, Nardella, M. *A Teacher's Guide to PKU* Texas Department of Health Available At: http://www.ub.edu.ar/centros_de_estudio/ceegmd/documentos/TeachersGuide.pdf Accessed June 15, 2011
[100]National Institutes of Health. *Phenylketonuria: Screening and Management National Institutes of Health Consensus Development Statement* Available At: http://consensus.nih.gov/2000/2000 phenylketonuria113html.htm Accessed May 17, 2011

Q. Does a child with PKU look or act differently from other children?
A. No, a child with PKU is just like other children in your classroom except that he/she has a special diet.

Q. How can I explain the PKU diet to children?
A. Young children can understand that since cars with different engines use different fuel (gas, diesel, etc.), some children have bodies that work in different ways than others and they need different food. Older children can understand the similar concept of a "food allergy." Don't hide the fact that the PKU child's lunch is different if asked, but no long explanation is needed. Ask the child's family for suggestions on how best to answer this question. You may also want to review our list of questions that children may encounter to prepare yourself for young, curious minds.

Q. If a child with PKU eats a high protein food, will he or she feel sick?
A. If a PKU child does eat a high protein food, he/she will probably not feel sick or different in any way. It is the long term, that elevated blood Phe level interferes with mental development. People with PKU may report feeling irritable and may have difficulty paying attention if they are not following the det completely. The changes may not be seen for several months to a year or more.

Q. What is the connection between aspartame and PKU?
A. Aspartame is used as a sweetening sugar substitute. When aspartame is broken down in the body, over half of it is Phe. Since individuals with PKU limit Phe in their diet, products containing aspartame needs to be avoided. A warning is required on all food products sweetened with aspartame, typically found in small print near the ingredient list:

PHENYLKETONURICS: Contains Phenylalanine

FAQs for Young Children

Children are very observant and will likely notice quickly that your child with PKU is eating different food. Also, while other children may share, it's important to teach your child with PKU that he or she cannot share from his or her friends and classmates. This will likely create questions from these curious children. Prepare your child for these questions by role playing and practicing so when he or she is asked, your child will be ready for it.

Q.Why can't you eat what we are eating?
A. "My body does not break down protein, so I have certain foods I cannot eat. My formula and diet are all a part of that. It keeps me healthy and I feel much better when I stick to it. Protein is kind of [bad for] me, so I have to plan my meals in advance. It takes some time, but in the long run it keeps me happy and healthy." (Patient Perspective)

Q. Are you allergic?
A. Sort of, but if I eat something that I shouldn't, it won't make me sick right away. But later, it could make me very sick.

Q. What happens if you eat something you aren't supposed to?
A. It might not do anything right away, but it might make me grumpy and have a hard time listening and paying attention in class later.

Q. What are you drinking? Why do you always drink that?
A. That's my special drink that gives me the good stuff you would get from food I can't eat. I drink it a lot so I can be strong and healthy.

Q. What kind of treats can you have?
A. I can have lollipops, popsicles and some gum.

Q. Are you sad that you can't eat anything you want?
A. Sometimes I wish I could, and sometimes it's hard not to, but I know it's better for me if I don't.

Resources

Educational Games

RED LIGHT!! GREEN LIGHT!!
Many parents use the Traffic Lights example to teach children about the PKU diet. To create a Traffic Light, draw three circles resembling traffic lights on a large poster board or piece of paper and color them green, yellow, and red. Then cut out pictures of many different types of foods from magazines or websites.

To start the game, explain to your child that there are three kinds of foods: "red", "yellow", and "green". These colors are defined as:

- Green = foods that are low in protein/Phe
- Yellow = foods that are only OK in limited quantities
- Red = foods that are high in protein and not on the PKU diet

Work with your child to organize foods according to each color. Once your child understands these ideas, you can create Traffic Light games to improve or test knowledge. New food pictures can create a fun challenge for your child as he or she figures out where each new food belongs on the traffic light, or you can test your child's recognition by placing a "red" food on the green light, or vice versa, and asking which of the foods does not belong.

The Traffic Light may also help you talk about diet choices with your child. Children familiar with the Traffic Light will readily understand what a "green" food or "red" food is, and this offers a way for parents to say no to foods without using the word "no" constantly. Some parents may also choose to refer to foods as "low Phe" and "high Phe" foods for this same reason.

Low Protein Grocery Shopping Tips

Low protein food offerings have also expanded significantly. Specially formulated breads, pastas, non-meat burgers, cheeses, muffins and cookies are now available, among others, and low protein food companies continue to offer new options for people with PKU.

There are also less expensive low protein foods that are available off the shelves such as those listed below.

PRODUCT	PRICE	PHE
Sunbelt fruit & grain cereal bars (blueberry/strawberry)	$2.00 per box	45-55 mg per bar
Sensible Portions Veggie Straws	$6.00 per 7oz bag	32 mg per serving (38 straws)
Sandwich Mate cheese slices	$1.50 per pack (16 slices)	24 mg per slice
Sun Luck Rice Sticks	~$2.00 per bag	35 mg per serving (1/4 of bag)
KAME bean threads	~$1 per bag	3 mg per 56 gram serving (uncooked)
Pepperidge Farm Very-thin sliced white bread	~$4.20 per loaf	67 mg per slice
Turtle Mountain So Delicious Coconut Milk Yogurt	~1.80 per 6oz	28 mg per 6oz

Resources

The NPKUA website (www.npkua.org) also has a list of low Phe foods available at Trader Joe's and Whole Foods under the PKU Management Tab at Living with PKU.

Support and Informational Resources

National PKU Alliance – www.npkua.org; 1-877-NPKUA-22
The National PKU Alliance (NPKUA) works to improve the lives of individuals and families associated with PKU through research, support, education and advocacy, while ultimately seeking a cure. The NPKUA is the first national non-profit organization to unite adults, families, statewide organizations, the medical community and PKU-friendly businesses under one umbrella organization.

The NPKUA website has links to resources that can help PKU patients and their families living with and managing their PKU, including information about PKU-friendly restaurants, travel destinations and every day information that can support you and your child manage PKU.

State and Regional PKU Organizations
For more information on PKU organizations by state, visit the NPKUA website at www.npkua.org. You can find state organizations listed in the PKU Resources brochure, under the Living with PKU tab.

NORD (National Organization for Rare Diseases) — www.rarediseases.org; 1-800-999-6673
NORD is a not-for-profit health agency dedicated to the identification, treatment, and cure of rare "orphan diseases" such as PKU through education, advocacy, research, and patient services programs.

Patient Power – www.patientpower.info; 1-877-232-5445
Patient Power® is a series of online radio and video programs for patients hosted by Andrew Schorr, a 13-year Leukemia survivor and patient advocate. The programs feature renowned medical experts and inspiring patients discussing an array of chronic medical conditions including PKU. The end goal is patient empowerment.

PKU Listserv - Listserv@Listserve.Emory.Edu
The PKU Listserv is a great place for families of a child with PKU, persons with PKU, and professionals involved in PKU treatment to share their ideas and concerns with others. This list provides an easy way for people all over the world to come together and communicate with one another about PKU. If you are interested in joining the listserv, send an email request including your name and email
address to: macpku@verizon.net.

PKU News – www.pkunews.org; (206) 525-8140
National PKU News is a non-profit organization located in Seattle, Washington dedicated to providing up-to-date, accurate news and information to families and professionals dealing with PKU. Since 1989, it has provided a 16-page newsletter three times yearly, and also has available widely used resources for PKU families including a food list, 2 cookbooks, and a children's book.

Social Networking Sites
Social networking sites such as **Facebook** can offer people with PKU the chance to connect with other PKU patients across the world for tips and support. In addition, local PKU organizations may have Facebook groups that you can join to stay up to date on what is going on in your area. To join, go to www.facebook.com.

Resources

PKU.com – www.pku.com
Offered by BioMarin, PKU.com provides comprehensive information about PKU and a place to meet others with PKU. Log on today to join the discussion and to make new PKU friends.
My PKU Toolkit – www.myPKUtoolkit.com
A website created by Applied Nutrition Corp. and Children's Hospital Boston that includes diet tips and model forms for self-management and advocacy for downloading.

PKU Online – www.citt.ufl.edu/team/PKU/beta/index.html
This website was developed by staff members from the Division of Genetics at the University of Florida to teach children about PKU management through interactive games and stories. A section for parents is available as well.

Your Genes Your Health – www.ygyh.org
The Your Genes Your Health webpage focuses on several genetic conditions (including PKU) with explanations and illustrations of the genetics of the condition, incidence, inheritance, symptoms, testing/screening, living with the condition, and treatment.

Wrightslaw – www.wrightslaw.com
This website is devoted to providing information about education law and how to advocate for individuals with disabilities.

Formula and Low-Protein Food Companies and Retailers

Abbott Nutrition – abbottnutrition.com; 1-800-227-5767
Abbott Nutrition provides medical nutritional products for the management of PKU, including Phenex-1 for infants and toddlers and Phenex-2 for children and adults.

Applied Nutrition – www.dietforlife.com; 1-800-605-0410
Applied Nutrition provides medical food products as well as Maddy's low protein foods available to the metabolic community.

Cambrooke Foods – www.cambrookefoods.com; 1-866 4 LOW PRO
Cambrooke Foods provides low protein food and metabolic formula products; products are available to be ordered online and delivered directly to your home.

Dietary Specialties – www.dietspec.com; 1-888-640-2800
Dietary Specialties provides low protein food products for the dietary management of metabolic disorders and medical conditions requiring low protein diets.

Ener-G Foods – www.ener-g.com; 1-800-331-5222
Ener-G Foods provides low protein food products for the dietary management of metabolic disorders and medical conditions requiring low protein diets.

Lil's Dietary Specialty Shop: www.lilsdietary.com; 773-239-0355
Lil's Dietary Specialty Shop is a Chicago-area dietary food store that carries low-protein products. Products can be ordered online and delivered directly to your home.

Resources

Mead Johnson Nutrition: www.mjn.com; 1-800-BABY123
Mead Johnson offers medical formulas for inborn errors of metabolism, such as PKU, including the Phenyl-Free line of medical formulas.

Med-Diet: www.med-diet.com; 1-800-MED-DIET
Med-Diet offers direct to home delivery of medical foods for conditions such as PKU, including their own line of broths and sauces.

Nutricia – www.myspecialdiet.com; 1-800-365-7354
Nutricia created a website devoted to the management of a metabolic diet. It includes an on-line store to purchase a range of medical foods, formulas and low protein foods, as well as cookbooks and software for diet management.
PKU Perspectives – www.pkuperspectives.com; 1-866-PKU-FOOD
PKU Perspectives provides low protein food products for the dietary management of metabolic disorders and medical conditions requiring low protein diets.

Taste Connections – www.tasteconnections.com; 310-371-8861
Taste Connections provides low protein food products for the dietary management of metabolic disorders and medical conditions requiring low protein diets.

Vitaflo – www.vitaflousa.com; 1-888-VITAFLO
Vitaflo provides a range of flexible medical food products for those with PKU, including ready-to-drink medical formula and other convenience optiosn.

Pharmaceutical Company Resources

KUVAN®

BioMarin – www.bmrn.com, www.kuvan.com; 1-866-906-6100
BioMarin commercializes KUVAN®, which is approved to reduce blood PHE levels in patients with hyperphenylalaninemia (HPA) due to tetra-hydrobiopterin- (BH4-) responsive PKU. Call BioMarin Patient and Physician Support (BPPS) to find out about Kuvan's free 30-day trial: 1-877-MY-KUVAN (1-877-695-8826), or email bpps@bmrn.com.

Large Neutral Amino Acids (LNAAS)

The companies below provide Large Neutral Amino Acid (LNAA) formulas for patients with PKU by clinic authorization:

Applied Nutrition: PheBLOC
www.medicalfood.com; 1-800-605-0410

Nutricia: Lanaflex
www.shsna.com; 1-800-365-7354

Solace Nutrition: PreKUnil® and NeoPhe®
www.solacenutrition.com; 1-888-8-SOLACE

Resources

Phe/Protein Food Reference Guides

Emory University's PKU Booklet is a pocket-sized comprehensive food list on the Phe exchange system that includes information about the Phe content in foods, household measurement information, serving sizes, calories, protein and exchanges. There is also room for a favorite food log and to keep notes. Order form is available at http://genetics.emory.edu/docs/Nutrition%20Docs/PKUorderform9-29-05.pdf

Virginia Schuett's "Low Protein Food List"
This food list is available in a 186-page book as well as for computers and handheld devices such as iPods and iPhones. Either format provides Phe, protein and calorie content of more than 3,000 foods. It includes tabbed chapters for Medical Foods, Baby Foods, Fruits & Vegetables, Beverages, Soups, Breakfast Foods, Low Protein Products, Sauces, Fats & Condiments, Grain Products, Crackers & Snacks, Sweets, Baking Ingredients, Convenience Foods, and Very High Protein Foods. Data sources include USDA and manufacturer's data. An index for the book can be downloaded from the National PKU News website, pkunews.org, in the Diet-Related Information section. Additions and corrections to the book are posted in the same section of the website. It is available to order at www.myspecialdiet.com and http://www.pkunews.org/forms/lowproFoodList.htm.

DietWell Application for iPhone and iPads
DietWell™ for PKU app for the iPhone®, iPod touch®, and iPad™ is designed to assist families and individuals in the dietary management of PKU. This app guides you through your meals, snacks, and formula intake by offering phenylalanine (phe), dietary protein, formula protein equivalent (P.E.), and calorie information for over 7500 food items, specially manufactured low protein food products, metabolic formula products, and Cambrooke recipes along with your Kuvan® intake, blood levels and wellness – all at the touch of your fingertips.

Cookbooks and Recipe Resources

WEBSITES FOR PKU RECIPES

Cook For Love: www.cookforlove.org
Cook for Love is a culinary non-for-profit dedicated to the PKU community. Its website and resources provide people with step-by-step instructions and videos for making low protein recipes. Cook for Love's mission is to empower members of the PKU community to improve their health through cooking and education.

Minnesota PKU Foundation: www.mnpku.org/dietrecipe
The Minnesota PKU Foundation is a nonprofit organization that promotes research and the welfare of PKU patients and their families. The website offers recipes for low protein cooking.

PKU of Illinois Foundation: The PKU Organization of Illinois is committed to the support of appropriate research initiatives to better understand PKU and eventually find a cure. Find tasty recipes at on their website at www.pkuil.org/recipes.

Resources

COOKBOOKS FOR PKU RECIPES

Virginia Schuett: Author Virginia Schuett has three PKU-friendly cookbooks available from Nutricia North America via www.myspecialdiet.com**.** They are

- "Apples to Zucchini: A Collection of Favorite Low Protein Recipes"
  This cookbook contains 562 healthy and delicious recipes in 12 chapters: Salad Celebrations; Soup's On; Bread and Beyond; Vegetables for All Seasons; Rice from East to West; Pasta, Please!; Hot Off the Grill; Where's the Beef?; The Adventurous Cook; Company's Coming; Sweet Delights; And Everything Else. The book's focus is on using naturally low protein fruits and vegetables, with minimal use of special brand-name low protein products.

- "Low Protein Cookery for PKU"
  This "classic" cookbook with over 450 recipes dates from 1977, but is still as relevant today as it was when it was first published. Hundreds of family-favorite recipes grace its pages, utilizing only "basic" special low protein ingredients and easy to find grocery store ingredients. Extensive Helpful Hints and Everyday Tips sections emphasize simple ideas to make the diet more easily managed, especially for younger children (which was the focus of treatment at the time the book was created). The recipes will appeal to a wide age range, from young children to adults and facilitate integration of the diet into normal family eating routines.

Gina Valente:

- "Gina Cooks Low Pro"
  Gina's cookbook gives suggestions and ideas for easy-to-make, quick and healthy low protein recipes. Example recipes include ice cream, pizza, burgers, mac and cheese, meatless balls, tortillas, nacho chips, pasta salad, pizzelles, chocolate cake, puddings and much more. Gina creates her recipes right from the grocery store using a few low pro foods (such as low pro pasta and low pro mixes). She suggests how to incorpo rate fiber, flax, omegas and other nutrients to the PKU diet that can otherwise be deficient. Futhermore, all of Gina's recipes have been reviewed and calculated by a registered dietitian for protein, PHE and calorie counts. A special offer of $16.99 plus S&H has been extended to those who use the code: NPKUA.ginacookslopro. T o order, email: ginacooks@optonline.net

MACPAD: The Mid-Atlantic Connection for PKU and Allied Disorders (MACPAD) is a non-profit organization dedicated to improving the health and well being of individuals and families affected by PKU and related metabolic disorders. MACPAD offers two cookbooks special for PKU diets:

- "Creative Family Cooking"
  Blood, sweat and possibly tears have gone into each creation in this collection of delicious recipes from people from the PKU community. Some people are blessed with the ability to create new recipes, others can adapt existing recipes and others have the skill to know what recipes will appeal to those with PKU. Some of the recipes are basic and others more involved, but each brings an individual accomplishment and adds to the knowledge of dealing with the sometimes

challenging dietary regime. The name of each chef is featured along with the recipe. Available at www.MACPAD.org for $10.

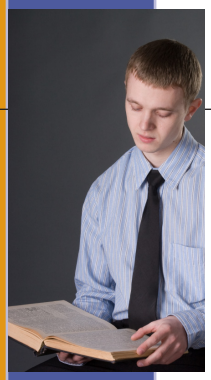
- "Family Friendly PKU Recipes"
  The driving force behind this, their second cookbook, was to find and adapt recipes that can be shared with the entire family, including family friendly recipes to compliment the low protein components. Each recipe was tasted and tested by MACPAD members and friends. Inside you will find sections like Best Baked Goods; Soups, Salads and Salsas; Sensational Salads; Enticing Entrees; Vivacious Vegetables; Fantastic Fruits and Sweets and Snacks. This cookbook is also available at www.MACPAD.org for $18.

Ordering Information

Scales
Most practitioners and PKU patients recommend gram scales made by Ohaus or Tanita. To purchase scales, many PKU patients recommend the website www.oldwillknottscales.com or eBay, but if purchasing from eBay, make sure the scale is new, not used.

Any scale you purchase should have the following features:
- The ability to weigh in grams in 1 gram units
- The ability to weigh up to 1,000 grams
- A tare function that allows you to zero out the weight of any container placed on the scale (before you add any food or formula) to get an accurate measurement of only the food or formula

Lancet Device
You should maintain an adequate number of lancet devices for blood sampling. Using a 21-gauge, trigger loaded lancet pen with settings for different depth options for the stick is recommended for PKU patients. These can be ordered through your local drug store.



Chapter 16: Glossary

Term	Definition
Blood Phe monitoring	Individuals with PKU must monitor their blood Phe levels to ensure that their levels remain in a healthy range. Blood samples are taken at home and at the PKU clinic to be analyzed, with the food record to help the PKU clinic dietitian adjust the PKU diet appropriately.
Classical PKU	The most common form of PKU. It results when there are two severe mutations of the PAH gene and as a result there is little or no PAH enzyme activity to convert phenylalanine to tyrosine. These are the most severely affected patients. Phe levels are above 20 mg/dl (>1200µmol/L).
Consensus Statement	When related to the NIH, a consensus statement is developed by a panel of experts in the field that presents a "best practices" for treating the disease or disorder being discussed. See http://consensus.nih.gov/ABOUTCDP.html for more information.
Food Record/Diet Record	A record of what foods have been consumed with date and time consumed and amount.
Food Reference Guide	A book that shows how much protein common foods contain, which helps you figure out and plan how much protein you will eat at each meal or snack. It also lists "free foods", i.e. foods that contain little or no protein, which do not need to be counted. The dietitian at your PKU clinic will give you a reference guide and help you use it.
Glycomacropeptide (GMP)	A whey-based protein produced when making cheese. It is the only known dietary protein that contains a minimal amount of PHE. Foods made with GMP provide an alternative to the amino acid medical foods currently required in the PKU diet that some find to be more palatable than other medical foods.
Home Blood Phe Monitor	A monitor that works similarly to a diabetes monitor, the Home Blood Phe Monitor is currently under development by BioMarin Pharmaceutical Inc., Novato, C., This would allow PKU patients to monitor their Phe levels at home.
Hyperphenylalaneia (HyperPhe)	Associated with high Phe levels, but potentially not high enough to require treatment. Phe level in blood: less than 6 mg/dl (<360 µmol/L)

Glossary

KUVAN® (sapropterin dihydrochloride)	A FDA approved medication that is used as a treatment method for some patients with PKU. KUVAN works by helping the phenylalanine hydroxylase (PAH) enzyme work more effectively to break down PHE in the body.
Large Neutral Amino Acid (LNAA)	LNAA is a treatment option that is mostly used for adults who have difficulty in maintaining the recommended PHE levels. LNAA is considered a medical food product, and comes in a powder or pill form containing essential amino acids (not including PHE) and large neutral amino acids. LNAA contains amino acids similar to those that are found in PKU formulas, but in more concentrated amounts.
Maternal PKU Syndrome	A syndrome that affects babies born to women with PKU whose Phe levels were not well controlled during pregnancy. Symptoms can include heart problems, slow development, small head size and brain damage.
Medical food	Medical Foods include both medical formula and foods modified to be low in protein. The medical formula provides all of the essential amino acids found in protein (except for Phe), as well as tyrosine, vitamins, minerals and trace elements that your body needs that most people who do not have PKU get from their diet. The foods modified to be low in protein provide an essential energy source and satiety with less than 1 gram of protein.
Moderate/Mild PKU	Is associated with elevated Phe levels, but not as high as for those patients with Classical PKU. Phe level in blood: Above 6 mg/dl but less than 20 mg/dl (360- 1200µmol/L).
National Institutes of Health (NIH)	The United States' medical research organization.
Newborn Screening	All babies born in the United States are tested for many diseases and disorders (including phenylketonuria) approximately 24 hours after birth through the state newborn screening test. A blood sample is taken from a needle prick on a baby's heel, and the blood is analyzed in a laboratory.
Phenylalanine (Phe)	An amino acid found in protein that PKU patients cannot break down.
Phenylalanine hydroxylase (PAH)	The enzyme in the liver that isdeficient in PKU.

Glossary

Phenylketonuria (PKU)	An inherited genetic disorder that affects approximately one in 14,000 babies born in the United States. A person with PKU does not produce enough of an enzyme in their liver called phenylalanine hydroxylase (PAH), which is needed to process the amino acid, phenylalanine (Phe).
PKU Clinic	A clinic that has a group of healthcare professionals trained to support individuals with PKU. Team members often include geneticists, dietitians, genetic counselors, social workers, nurses and sometimes psychologists.
PKU Diet/Low Protein Diet	The PKU diet consists of low protein foods such as fruits, vegetables, fats, sugars, special low protein food such as low protein pasta and medical formula.
Therapeutic Liver Repopulation	Aims to replace the cells that are deficient in PAH (PAH negative cells) with cells that are not PAH-deficient (PAH positive cells). PAH positive cells will then restore function of the liver, theoretically curing PKU.