

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.

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"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:

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- 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.

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"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."

- 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.

Many parents find that they need support from other families who have children with PKU to feel reassured that their child will be fine¹⁸. 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 occur¹⁹. 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.

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"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

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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 food²⁰. 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 diet²¹. 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.

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

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"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."

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

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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 infant 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 work 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.

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).

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.

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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)."*

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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	<ul style="list-style-type: none"> • 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 	<ul style="list-style-type: none"> • 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 	<ul style="list-style-type: none"> • 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.

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"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.

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.

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!

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.

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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.

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 μ mol/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.

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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.

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.*

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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

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"...[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.

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

"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

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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.