



Genetic Fact Sheets for Parents

Amino Acid Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – <http://www.newbornscreening.info>

Disorder name: Phenylketonuria
Acronym: PKU

- What is PKU?
- What causes PKU?
- If PKU is not treated, what problems occur?
- What is the treatment for PKU?
- What happens when PKU is treated?
- What causes the PAH enzyme to be absent or not working correctly?
- How is PKU inherited?
- Is genetic testing available?
- What other testing is available?
- Can you test during pregnancy?
- Can other members of the family have PKU or be carriers?
- Can other family members be tested?
- How many people have PKU?
- Does PKU happen more often in a certain ethnic group?
- Does PKU go by any other names?
- Where can I find more information?

This fact sheet has general information about PKU. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be advised for some children but not others. All children with PKU should be followed by a metabolic doctor in addition to their primary care provider.

What is PKU?

PKU stands for “phenylketonuria”. It is one type of amino acid disorder. People with PKU have problems breaking down an amino acid called phenylalanine from the food they eat.

Amino Acid Disorders:

Amino acid disorders (AAs) are a group of rare inherited conditions. They are caused by enzymes that do not work properly.

Protein is made up of smaller building blocks called amino acids. A number of different enzymes are needed to process these amino acids for use by the body. Because of missing or non-working enzymes, people with amino acid disorders cannot process certain amino acids. These amino acids, along with other toxic substances, then build up in the body and cause problems.

The symptoms and treatment vary between different amino acid disorders. They can also vary from person to person with the same amino acid disorder. See the fact sheets for each specific amino acid disorder.

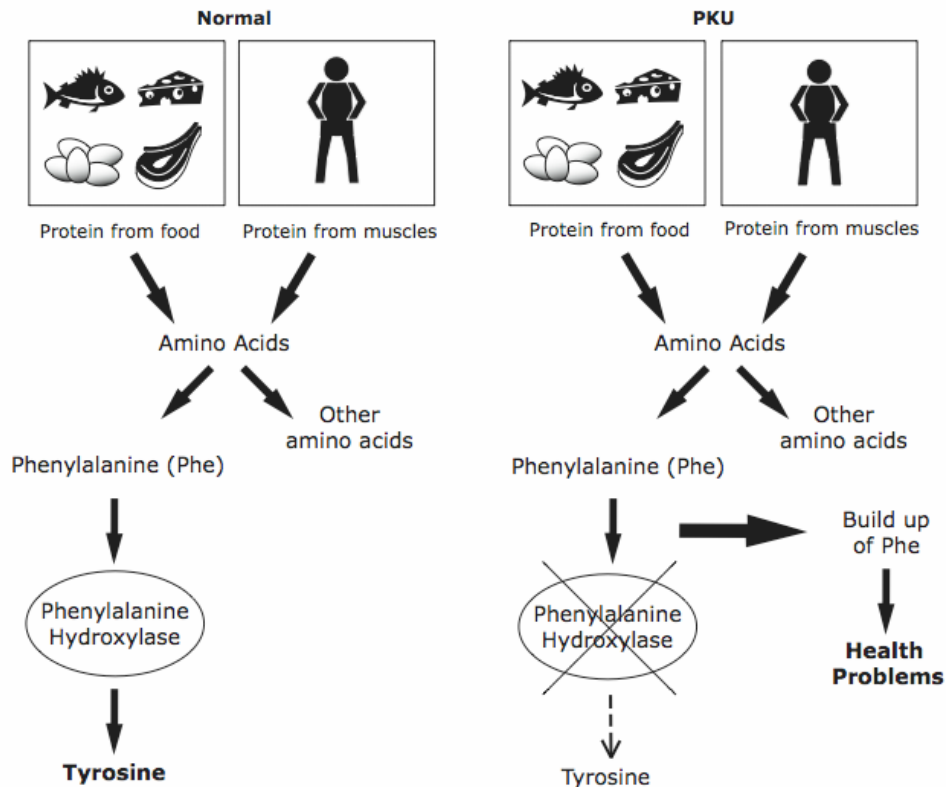
Amino acid disorders are inherited in an autosomal recessive manner and affect both males and females.

What causes PKU?

In order for the body to use protein from the food we eat, it is broken down into smaller parts called amino acids. Special enzymes then make changes to the amino acids so the body can use them.

PKU occurs when an enzyme, called “phenylalanine hydroxylase” (PAH), is either missing or not working properly. This enzyme’s job is to break down the amino acid phenylalanine (Phe – pronounced ‘fee’). When a child with PKU eats food containing Phe, it builds up in the blood and causes problems. Phe is found in almost every food, except pure fat and sugar.

PHENYLKETONURIA (PKU)



If PKU is not treated, what problems occur?

Babies with PKU seem perfectly normal at birth. The first effects are usually seen around 6 months of age. Untreated infants may be late in learning to sit, crawl and stand. They may pay less attention to things around them. Without treatment, a child with PKU will become mentally retarded.

Some of the effects of untreated PKU include:

- mental retardation
- behavior problems
- hyperactivity
- restlessness or irritability
- seizures
- a skin condition called eczema
- a “musty” or “mousy” body odor
- fair hair and skin

What is the treatment for PKU?

Your baby's primary doctor will work with a metabolic doctor and a dietician to care for your child.

Prompt treatment is needed to prevent mental retardation. Newborns need to drink a special medical formula. It is still possible to breastfeed your baby as long as you get help from a dietitian. Babies who are breastfed usually need the medical formula as well.

Most children need to eat a special diet made up of very low-protein foods, special medical foods, and the special formula. You must start the low-Phe diet as soon as you know your child has PKU. Your dietitian will create a food plan that contains the right amount of protein, nutrients, and energy to keep your child healthy. The diet should be continued throughout life.

The following are treatments often advised for children with PKU:

1. Medical formula

Even though they need less Phe, children with PKU still need a certain amount of protein. The medical formula gives babies and children with PKU the nutrients and protein they need while helping keep their Phe levels within a safe range.

Your metabolic doctor and dietitian will tell you what type of formula is best and how much to use. Some states offer help with payment, or require private insurance coverage for the formula and other special medical foods.

2. Low-Phe food plan

The low-Phe diet is made up of foods that are very low in Phe. This means your child must not have cow's milk, regular formula, meat, fish, eggs or cheese. Regular flour, dried beans, nuts, and peanut butter also have Phe and must be avoided or strictly limited.

It is very important that your child avoid the sugar substitute aspartame (sold under the brand names "Equal", Nutrasweet" "Sweetmate", "Canderl"). Aspartame contains high amounts of Phe. It can quickly raise the blood levels of Phe in people with PKU. Your child must not have any diet foods or drinks that contain aspartame. Some medicines and vitamins also contain aspartame. If you're not sure, ask your pharmacist, metabolic doctor or dietitian.

Many vegetables and fruits have only small amounts of Phe and can be eaten in carefully measured amounts. In addition, there are other medical foods such as low-Phe flours, baking mixes, breads, and pastas that are made especially for people with PKU.

Your child's food plan will depend on many things such as his or her age, weight, general health, and blood test results. Your dietician will fine-tune your child's diet over time.

Your child should follow this diet throughout life. Adults who do not stay on the diet and have high levels of Phe in their blood may notice some of the following:

- trouble paying attention
- problems making good decisions
- slow thinking
- irritability
- eczema

Women need to be on the low-Phe diet before becoming pregnant. They need to stay on the diet throughout pregnancy. This will lessen the chance for serious health and learning problems in their babies.

3. Tracking Phe levels

Babies and young children with PKU need to have regular blood tests to measure their Phe levels. If there is too much or too little Phe in the blood, the diet and formula may need to be adjusted.

4. Pregnancy in women with PKU (“Maternal PKU”)

Women with PKU who are not on the low-Phe diet when they become pregnant have a high chance of having babies with birth defects and mental retardation.

Women who are not on the diet usually have high levels of Phe in their blood. The extra Phe gets to the fetus and causes problems with brain and body growth. Babies of untreated mothers may have the following:

- small brains
- mental retardation
- birth defects of the heart
- low birth weight

This condition is called “maternal PKU syndrome”.

Women with PKU who want to have children need to have very low blood Phe levels before they get pregnant. During pregnancy, they need to:

- stay on the low-Phe diet
- visit their PKU clinic on a regular basis
- have their blood Phe levels checked often

What happens when PKU is treated?

Children with PKU who start treatment soon after birth and keep their Phe levels within the suggested range usually have normal growth and intelligence. Some

children, even when treated, have problems with school work and may need extra help.

If treatment is not started until several weeks after birth, delays or learning problems may occur. The level of delay varies from child to child.

Children who start treatment after 6 months of age are often mentally retarded. Treatment is still important, even if started late, because it can help control behavior and mood problems and can prevent further damage to the brain.

What causes the PAH enzyme to be absent or not working correctly?

Genes tell the body to make various enzymes. People with PKU have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the PAH enzyme either does not work properly or is not made at all.

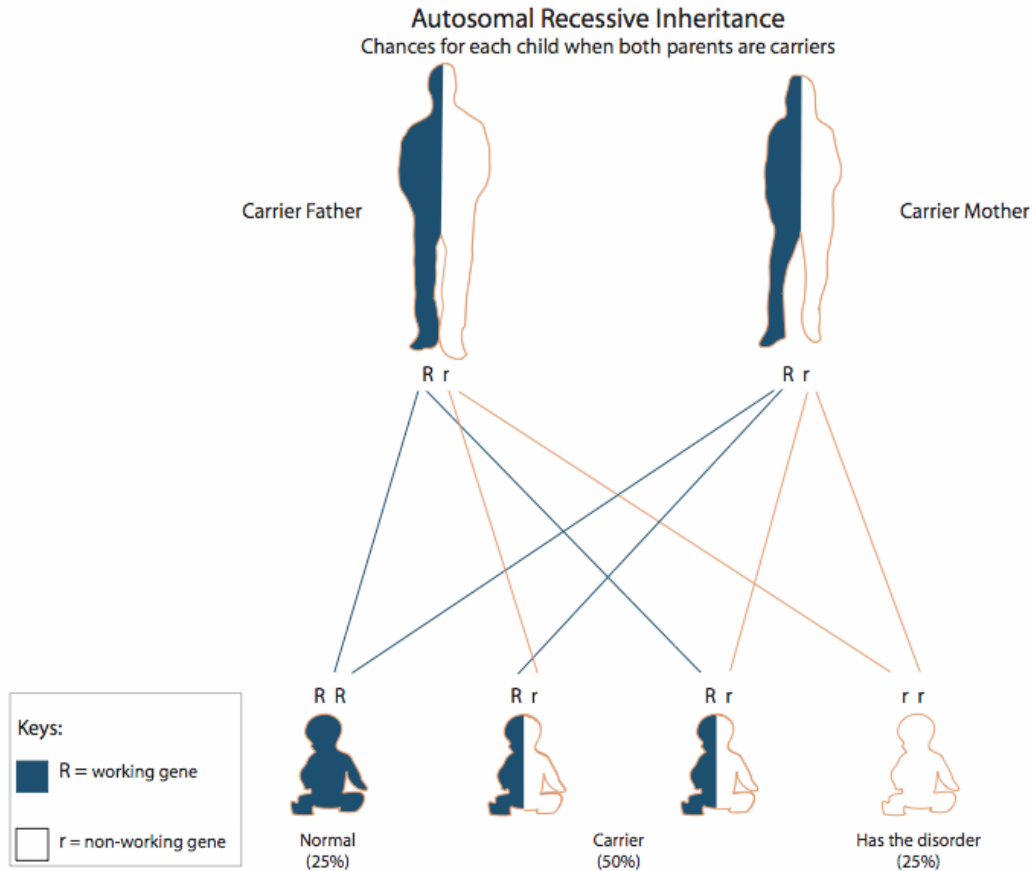
How is PKU inherited?

PKU is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has a pair of genes that make the PAH enzyme. In children with PKU, neither of these genes works correctly. These children inherit one non-working gene for the condition from each parent.

Parents of children with PKU rarely have the condition themselves. Instead, each parent has a single non-working gene for PKU. They are called carriers. Carriers do not have PKU because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have PKU. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



Genetic counseling is available to families who have children with PKU. Genetic counselors can answer your questions about how PKU is inherited, choices during future pregnancies, and how to test other family members. Ask your doctor about a referral to a genetic counselor.

Is genetic testing available?

Genetic testing for PKU can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes in the pair of genes that causes PKU.

DNA testing is not necessary to diagnose your child. It can be helpful for carrier or prenatal testing, discussed below.

What other testing is available?

PKU is confirmed by measuring the amount of Phe in a blood sample. Talk to your doctor or your genetic counselor if you have questions about testing for PKU.

Can you test during pregnancy?

If both gene changes have been found in your child with PKU, DNA testing can be done during future pregnancies. The sample needed for this test is obtained by either CVS or amniocentesis.

Parents may choose to have testing during pregnancy or wait until birth to have the baby tested. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

Can other members of the family have PKU or be carriers?

Having PKU

If they are healthy and developing normally, older brothers and sisters of a baby with PKU are unlikely to have PKU. Talk to your doctor or genetic counselor if you have questions about your other children.

PKU carriers

Brothers and sisters who do not have PKU still have a chance to be carriers like their parents. Except in special cases, carrier testing should only be done in people over 18 years of age.

Each of the parents' brothers and sisters has a 50% chance to be a carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with PKU.

Some states do not offer newborn screening for PKU. However, expanded newborn screening through private labs is available for babies born in states that do not screen for this condition. To learn more about expanded newborn screening, see [How to obtain MS/MS](#).

When both parents are carriers, newborn screening results are not sufficient to rule out the condition in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

Can other family members be tested?

Diagnostic testing

If there is concern about whether they have the condition, your other children can be tested. Talk to your doctor or genetic counselor if you have questions about testing for PKU.

Carrier testing

If both gene changes have been found in your child, other family members can have DNA testing to see if they are carriers.

If DNA testing is not helpful, other methods of carrier testing may be available. If you have questions about carrier testing, ask your genetic counselor or metabolic doctor.

How many people have PKU?

About one in every 10,000 Caucasian babies in the United States is born with PKU.

Does PKU happen more frequently in a certain ethnic group?

PKU happens in people of all ethnic groups around the world. It happens more often in people whose families come from Ireland and other parts of Northern Europe. It is also more common in people from Turkey. About 1 in every 50 Caucasians is a PKU carrier.

Does PKU go by any other names?

PKU is sometimes also called:

- hyperphenylalaninemia
- phenylalanine hydroxylase deficiency

Some variants of PKU not discussed in this fact sheet are:

- hyperphenylalaniemia – mild type
- bipterin deficiency
- dihydropteridine reductase deficiency

Where can I find more information?

Children's PKU network

<http://www.pkunetwork.org/>

National Coalition for PKU and Allied Disorders

<http://www.pku-allieddisorders.org/>

Children Living with Inherited Metabolic Diseases (CLIMB)

<http://www.climb.org.uk>

Genetic Alliance

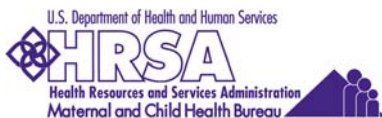
<http://www.geneticalliance.org>

Document Info

Created by: www.newbornscreening.info
Reviewed by: HI, CA, OR, and WA metabolic specialists
Review date: September 31, 2006
Update on: September 31, 2006

DISCLAIMER:

THIS INFORMATION DOES NOT PROVIDE MEDICAL ADVICE. All content ("Content"), including text, graphics, images and information are for general informational purposes only. You are encouraged to confer with your doctor or other health care professional with regard to information contained on this information sheet. After reading this information sheet, you are encouraged to review the information carefully with your doctor or other healthcare provider. The Content is not intended to be a substitute for professional medical advice, diagnosis or treatment. NEVER DISREGARD PROFESSIONAL MEDICAL ADVICE, OR DELAY IN SEEKING IT, BECAUSE OF SOMETHING YOU HAVE READ ON THIS INFORMATION SHEET.



This project is supported by a grant from the Maternal and Child Health Bureau, Health Resources and Service Administration, Genetic Services Branch, MCH Project #:1H46 MC 00189-03 <http://mchb.hrsa.gov>