



PKU (Phenylketonuria)

What is it?

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.

What causes it?

In order for the body to use protein from food, 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.

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:

- 1) Mental retardation.
- 2) Behavior problems.
- 3) Hyperactivity.
- 4) Restlessness or irritability.

- 5) Seizures.
- 6) A skin condition called eczema.
- 7) A musty or mousy body odor.
- 8) 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 dietician. 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 dietician can 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:

Medical Formula

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

Your metabolic doctor and dietician will tell you what type of formula is best and how much to use.

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 and Canderol). 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 dietician.

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:

- 1) Trouble paying attention
- 2) Problems making good decisions
- 3) Slow thinking
- 4) Irritability
- 5) Eczema

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.

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 with PKU 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 the brain and body growth. Babies of untreated mothers may have the following:

- 1) Small brains
- 2) Mental retardation
- 3) Birth defects of the heart
- 4) 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:

- 1) Stay on a low-Phe diet.
- 2) Visit their PKU clinic on a regular basis.
- 3) 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 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 PKU enzymes 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 does not work properly or is not made at all.

Is PKU inherited?

PKU is inherited and 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 a nonworking gene for the condition from each parent.

Parents of children with PKU rarely have the condition themselves. Instead, each parent has a single nonworking 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 percent chance in each pregnancy for the child to have PKU. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

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

Having PKU

If they are healthy and growing 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 be done only in people older than 18.

Each of the parents' brothers and sisters has a 50 percent 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.

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 10,000 to 15,000 people of all ethnic groups 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 one in every 50 Caucasians is a PKU carrier.

Does PKU go by any other names?

PKU also is called:

- 1) Hyperphenylalaninemia.
- 2) Phenylalanine hydroxylase deficiency.

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

- 1) Hyperphenylalaninemia - mild type.
- 2) Biopterin deficiency.
- 3) Dihydropteridine reductase deficiency.

Where can I find more information?

Children's PKU network

www.pkunetwork.org

Children Living with Inherited Metabolic Diseases (CLIMB)

www.climb.org.uk

National Coalition for PKU and Allied Disorders

www.pku-allieddisorders.org

Genetic Alliance

www.geneticalliance.org

Children's Special Health Services (CSHS)

State Capitol Judicial Wing
600 E. Boulevard Ave., Department 301
Bismarck, ND 58505-0269

Toll Free: 800.755.2714

701.328.2436

Relay TDD: 701.328.3975

CSHS website: www.ndhealth.gov/CSHS

North Dakota Department of Health website:

www.health.gov

Family support resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

Family Resources

Family to Family Network

Center for Rural Health

University of North Dakota

School of Medicine and Health

PO Box 9037

Grand Forks, ND 58202-9037

Toll Free: 888 434.7436

701.777.2359

Fax: 701.777.2353

E-mail: NDF2F@medicine.nodak.edu

www.medicine.nodak.edu/crh

Pathfinder Services of ND

Pathfinder Family Center

1600 2nd Ave. SW, Ste. 19

Minot, ND 58701

Toll Free: 800.245.5840

701.837.7500

Relay TDD: 701.837.7501

E-mail: ndpath01@ndak.net

www.pathfinder.minot.com

Family Voices of North Dakota, Inc.

P.O. Box 163

Edgeley, ND 58433

Toll Free: 888.522.9654

701.493.2634

Fax: 701.493.2635

www.geocities.com/ndfv

**This fact sheet has general information.

Every child is different and some of these facts may not apply to your child specifically.

Certain treatments may be recommended for some children but not others. All children should be followed by a metabolic doctor in addition to their primary care provider.

Acknowledgement

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Disclaimer

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