



## **The Arc Answers: PKU or Phenylketonuria**

### **How many people have Phenylketonuria?**

Each year, approximately 1 in 15-25,000 babies are born with PKU in the United States. It is more common in people of Northern European and Native American descent.

### **What causes PKU?**

PKU is an inherited disorder. Both parents must pass on a mutated gene on chromosome 12 for their child to inherit PKU. If both parents are carriers, there is a 1 in 4 chance that this will happen. If the child only receives one copy of the gene, they will become a carrier but will not have PKU. When a child inherits PKU, they are unable to break down the amino acid, phenylalanine. This leads to a dangerous build up of phenylalanine (sometimes called “phe”), in the blood stream, central nervous system and brain.

### **How is PKU diagnosed?**

Doctors can screen for PKU prenatally. Newborns do not show any signs or symptoms, but babies born in US hospitals are screened within a day or two of birth using a blood test that detects phenylalanine levels.

### **What are the primary characteristics of PKU?**

Infants with PKU often have lighter skin, eyes, and hair color than their siblings as phenylalanine affects the body's production of melanine. Children who are undiagnosed or untreated can develop irreversible brain damage and intellectual disabilities within the first year of life. By age 3-6 months, untreated infants may show decreased interest in their surroundings and lower activity levels. By age 1, children may exhibit developmental delays, irritability and restlessness.

Children and adults with untreated PKU may also be at greater risk for experiencing various behavioral and mental health concerns due to the way that phenylalanine affects brain chemistry. These include: attention deficit hyperactivity disorder (ADHD), anxiety, phobias, depression, psychosis, and obsessive-compulsive disorder.

### **What are common medical concerns?**

Children and adults may experience seizures, rashes similar to eczema, and dry skin.

### **What is maternal PKU?**

Maternal PKU occurs when a pregnant woman with PKU does not follow a low-phenylalanine diet. The high levels of phenylalanine in her system can cause her child to have intellectual disabilities, a small head size, heart defects, and a low birth weight, even if the child doesn't inherit PKU.

### **What interventions have proven helpful to people with PKU?**

Doctors recommend that people with PKU follow a strict, life-long diet that limits their intake of phenylalanine, which is mostly found in foods high in protein. Examples of such foods include: milk, eggs, cheese, nuts, chicken, beef, products with aspartame, and chocolate, among others. Children and adults may need to drink a daily supplemental formula to get necessary nutrients. Doctors and dieticians work with each person individually to determine a safe level of phenylalanine and an appropriate diet for them. A medication that helps regulate phenylalanine, taken in conjunction with a PKU diet, has recently come onto the market and may be an option for some people.

### **Where can I find other resources?**

Children's PKU Network, a non-profit organization: <http://www.pkunetwork.org/>

Mayo Clinic: <http://www.mayoclinic.com/health/phenylketonuria/DS00514>

The Mid-Atlantic Connection for PKU and Allied Disorders: <http://www.macpad.org>

National PKU Alliance: <http://npkua.org/>

National PKU News: <http://pkunews.org/>

The Arc of NC  
343 East Six Forks Rd. Ste.320  
Raleigh, NC 27609  
1-800-662-8706  
[www.arcnc.org](http://www.arcnc.org)