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Editorial Note on Phenylketonuria

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Editorial

PKU, or phenylketonuria, is a rare inherited disorder that causes an amino acid called phenylalanine to accumulate in the body. PKU is caused by a mutation in the gene that produces the enzyme required to break down phenylalanine. Without the enzyme required to process phenylalanine, a dangerous buildup can form when a person with PKU consumes protein-containing foods or aspartame, an artificial sweetener. This can lead to serious health issues in the long run. PKU babies, children, and adults must eat a diet low in phenylalanine, which is found primarily in protein-containing foods, for the rest of their lives. In the United States and many other countries, babies are screened for PKU shortly after birth.

Women who have PKU and become pregnant are at risk of developing a different form of the condition known as maternal PKU. If women do not adhere to the special PKU diet before and during pregnancy, blood phenylalanine levels can rise, endangering the developing foetus or resulting in a miscarriage. Even women with less severe forms of PKU may endanger their unborn children if they do not adhere to the PKU diet. PKU is rarely inherited by babies born to mothers with high phenylalanine levels. However, if the mother's blood phenylalanine level is high during pregnancy, they can have serious consequences [1-3].

PKU is caused by a faulty gene (genetic mutation), which can be mild, moderate, or severe. This defective gene results in a lack or deficiency of the enzyme required to process phenylalanine, an amino acid, in a person with PKU. When a person with PKU consumes protein-rich foods such as milk, cheese, nuts, or meat, as well as grains such as bread and pasta, a dangerous buildup of phenylalanine can occur, as can the consumption of aspartame, an artificial sweetener. This phenylalanine buildup causes nerve cell damage in the brain.

In order for a child to inherit PKU, both the mother and father must have the defective gene and pass it on to the child. This inheritance pattern is known as autosomal recessive. A parent can be a carrier that is they have the defective gene that causes PKU but do not have the disease. There is no risk of passing PKU to a child if only one parent has the defective gene, but the child may be a carrier. PKU is most commonly passed down to children by two parents who are carriers of the disorder but are unaware of it. Having a defective gene in both parents, which causes PKU. In order for their child to develop the condition, both parents must pass on a copy of the defective gene [4,5]. Being of a certain ethnicity. PKU is caused by a gene defect that varies by ethnic group, and it is less common in African-Americans than in other ethnic groups. Untreated PKU can lead to complications in infants, children, and adults. Fetal birth defects or miscarriage can occur when mothers with PKU have high blood phenylalanine levels during pregnancy.

Almost all cases of phenylketonuria are detected by newborn blood testing. In the United States, all states require newborns to be screened for PKU. Many other countries routinely screen infants for PKU as well. If you have PKU or a family history of it, your doctor may advise you to have screening tests before getting pregnant or giving birth. A blood test can be used to identify PKU carriers. A PKU test is performed a few days after your baby is born. To ensure accurate results, the test is performed after your baby is 24 hours old and has consumed some protein in his or her diet. A nurse or laboratory technician takes a few drops of blood from your baby's heel or the bend in his or her arm. Because protein-containing foods contain phenylalanine, a lifetime diet with very limited protein intake is recommended.

Taking a PKU formula a special nutritional supplement for the rest of your life to ensure you get enough essential protein (without phenylalanine) and nutrients needed for growth and overall health. A safe amount of phenylalanine varies from person to person with PKU and can change over time. In general, the goal is to consume no more phenylalanine than is required for normal growth and body processes. Your doctor can determine a safe amount by reviewing diet records, growth charts, and phenylalanine levels in your blood on a regular basis. Frequent blood tests to track phenylalanine levels as they fluctuate over time, particularly during childhood growth spurts and pregnancy.

Conflict of Interest

None.

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