Phenylketonuria (PKU)
General Overview

Q. What is PKU?

A. Phenylketonuria (PKU) is a treatable disorder that affects the way the body processes protein. Children with PKU cannot use a part of the protein called phenylalanine. If left untreated, phenylalanine builds up in the bloodstream and causes brain damage.

Q. How does the body normally process phenylalanine?

A. The body normally converts phenylalanine into tyrosine, which is used by the body in other metabolic functions. This conversion is made possible by an enzyme called phenylalanine hydroxylase.

Q. What happens to phenylalanine in a child with PKU?

A. In a child with PKU, phenylalanine cannot be converted to tyrosine because the phenylalanine hydroxylase enzyme does not work properly. This results in dangerously high levels of phenylalanine that build up in the blood and become toxic to the brain and nervous system.

Q. What are the effects of having PKU if it is not treated?

A. Untreated PKU results in severe mental retardation and other problems of the nervous system.

Q. What is the treatment for PKU?

A. Phenylketonuria is treated with a special diet that is low in phenyalanine. To prevent mental retardation, treatment must begin shortly after birth. Since protein is essential for normal growth and development, the child must continue on a special formula that provides protein and essential nutrients, but contains little or no phenylalanine. People with PKU require specialized treatment through a clinic with experience in treating this disorder.

Q. Why would a child have PKU?

A. Phenylketonuria is an inherited disorder. It results when a baby receives a double-dose of a non-working phenylalanine hydroxylase gene (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. How common is PKU?

About one in every 15,000 babies in Washington State is born with PKU.

For more information about PKU, please see the Disorders section of our website: www.doh.wa.gov/nbs.

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