Mild 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 mild PKU have some difficulty using a part of the protein called phenylalanine. Blood phenylalanine levels need to be monitored periodically to determine if treatment is necessary.

Q. Is there only one form of PKU?

A. No, there are several forms of PKU. Mild PKU is a less severe form.

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 mild PKU?

A. In a child with mild PKU, the phenylalanine hydroxylase enzyme does not work very well. This results in higher than normal levels of phenylalanine that build up in the blood.

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

A. A child with mild PKU only requires treatment if blood phenylalanine levels reach a certain point.

Q. What is the treatment for mild PKU?

A. Children with mild PKU have regular testing to make sure the phenylalanine levels in their blood are safe. Treatment with a special diet that is low in phenylalanine is given if levels are outside of the safe range. People with mild PKU should be seen at a clinic with experience in treating this disorder.

Q. Why would a child have mild PKU?

A. Mild PKU is an inherited disorder. It results when a baby receives a double-dose of a phenylalanine hydroxylase gene that does not work well (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. How common is mild PKU?

About one in every 24,000 babies in Washington State is born with mild PKU.

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

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