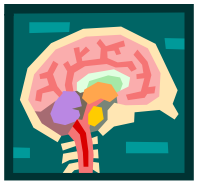

Disability Awareness Begins With You:

Phenylketonuria (PKU)

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What is phenylketonuria (PKU)?

Phenylketonuria (pronounced fee-nill-key-toe-NURR-ee-uh) or PKU is an inherited disorder of metabolism that can cause mental retardation if not treated.

In PKU, the body can't process a portion of the protein called phenylalanine (Phe), which is in almost all foods. If the Phe level gets too high, the brain can become damaged.

All babies born in U.S. hospitals are now routinely tested for PKU soon after birth, making it easier to diagnose and treat them early.

What are the symptoms of PKU?

Children with untreated PKU may appear normal at birth. By age three to six months, they begin to lose interest in their surroundings. By age one year, they are developmentally delayed and their skin has less pigmentation than someone without the condition. If Phe is not restricted in the diet, those with PKU develop severe mental retardation.

What are the treatments for PKU?

The most effective treatment for PKU is a special diet of foods that help control the amount of Phe consumed (some Phe is needed for normal growth and development). People with PKU who are on this diet from birth or shortly thereafter develop normally and often have no symptoms of PKU.

The PKU diet includes fruits, vegetables, and some low-protein breads, pastas, and cereals. There is also a special formula, made without Phe, that people with PKU drink to help them get the vitamins and minerals they can't get from their food.

Generally, people with PKU can't eat high-protein foods, such as meat, milk, eggs, and nuts. An NIH Consensus Panel recently recommended that people with PKU stay on the diet for life to promote overall health and to prevent decline in mental function.

Are there other concerns for those with PKU?

Pregnant women who know they have PKU need to keep good control of the level of Phe in their diets beginning before and continuing throughout pregnancy. High levels of Phe in the blood can cause developmental problems and birth defects (such as small brain size and heart defects) in the fetus.

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