Phenylketonuria (PKU)

Phenylketonuria (PKU) is a hereditary disease that is caused by the lack of a liver enzyme. This enzyme is needed to digest phenylalanine. Phenylalanine is an amino acid that is most commonly found in foods containing protein. Some of these foods are meat, cow's milk, over-the-counter infant formulas (both regular and soy) and breast milk.

CAUSES

The gene defect for phenylketonuria is carried by people who do not know they have it and pass it down to their children as a disease. This faulty gene only shows up when two carriers have children together and pass it to their offspring. For each pregnancy of two such carriers, there is a 1 in 4 chance that the child will be born with the disease. There is a 1 in 2 chance that the child will be a carrier for the gene defect.

SYMPTOMS

Newborns affected by PKU usually do not show any signs of the disease at birth. Within the first few weeks of life, they begin to show neurologic problems such as epilepsy. Children suffering from PKU also may have an unpleasant, musty smell. Almost 90% of affected people have blond hair and blue eyes. Signs also include skeletal changes such as a small head, short stature, and flat feet. People with PKU may also suffer from *eczema* (a skin disorder).

TREATMENT

Treatment of PKU is the removal of phenylalanine from the diet. Babies who are diagnosed with PKU must be immediately be put on a special milk/formula substitute. Later in life, the diet will be mainly vegetarian.

Females who have PKU, become pregnant, and have stopped following the appropriate diet, must return to the diet throughout all pregnancies. Babies of females with PKU who do not stay on the suggested diet during pregnancy can be severely malformed and mentally retarded. Females who are on the suggested diet have normal, healthy infants.

PROGNOSIS

PKU-affected children who are not diagnosed and treated will suffer from irreversible brain damage and mental retardation. Properly treated individuals should live a normal, healthy life.

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