PHENYLKETONURIA

What is Phenylketonuria?

Phenylketonuria or PKU for is one of the conditions that can be detected through newborn screening. Untreated children may have mental retardation. Early detection can prevent brain damage.

What causes Phenylketonuria?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme (the phenylalanine hydroxylase (PAH) enzyme), phenylalanine, an amino acid which is one of the building blocks of protein cannot be broken down. The accumulation of phenylalanine causes the signs and symptoms of PKU.

PKU is an inherited condition. The PAH gene is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the PAH gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with PKU have one working and one non-working gene coding for PAH. They do not manifest the disease but can pass them on to their children. They are known as carriers.
If the child inherits the non-working gene from both parents, he or she will have PKU. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of PKU?

Children with PKU may seem normal at birth. They begin to manifest at about 6 months of age with delayed development (late in learning to sit, stand etc). Untreated children may also have decreased attention span, mental retardation, irritability, convulsions, hyperactivity and behavioral problems.

How is PKU diagnosed?

Newborn screening by dried blood spot on a filter paper can detect presence of PKU in newborns. A blood test called amino acid chromatography is diagnostic of PKU. A urine test called organic acid chromatography is supportive to amino acid chromatography.

What is the treatment of PKU?

The main treatment of PKU is through control of the diet. If the child is well or does not have any illness, he/she should continue taking the special milk formula which does not have phenylalanine and be on low protein diet as ordered by pediatrician or metabolic

specialist. It is important that the diet be followed to avoid any complications.

What should I do when my baby is sick?

When children with PKU are sick, phenylalanine may accumulate in the blood. Child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage. Once these signs and symptoms are present, child should be admitted to the hospital for management and alert your pediatrician or metabolic physician.