Phenylketonuria (PKU)

What is PKU?

PKU is an inherited inborn metabolism error in which affected individuals cannot use the aminoacid phenylalanine appropriately leading it to accumulate in the blood potentially causing serious brain damage.

What causes the disease?

Mutations in the phenylalanine hydroxylase (PAH) gene produce a defective enzyme that is unable to metabolize or breakdown phenylalanine properly**.**

What are the clinical features of PKU?

Babies with PKU are clinically indistinguishable from healthy babies for the first few weeks of life. Without treatment, however, phenylalanine accumulation will cause severe and irreversible mental retardation, eczema, and other problems.Children with untreated PKU may also have a distinctive “mousy” odor. Phenylalanine levels start rising within 24 hours of birth. Newborns with elevated levels on blood spot analysis are investigated further.

How is the diagnosis confirmed?

The diagnosis is confirmed by measuring the levels of plasma phenylalanine and tyrosine in the blood. Other tests may also be done to rule out other causes of higher phenylalanine levels. A diagnosis of PKU can also be confirmed by a genetic analysis of the PAH gene. Diagnostic testing is arranged by specialists at BC Children’s Hospital.

What is the treatment of the disease?

A low protein diet with low phenylalanine intake should be started as soon as possible to prevent mental retardation and other problems. Some phenylalanine is required by the body for normal growth and development, thus it should not be eliminated from the diet. Frequent monitoring of plasma amino acid levels, weight gain, and development are recommended. Adult women with PKU are at risk to have babies with microcephaly, poor growth, and/or mental retardation if their phenylalanine levels are persistently elevated during pregnancy. Therefore, pregnant women with PKU should also be monitored closely. A Biochemical genetics specialist and a Metabolic Genetics dietitian should coordinate the treatment.