

## PKU Testing Informed Consent

Metabolic Screening Tests for Newborns, commonly called PKU, screens for a variety of treatable metabolic disorders. There are dozens of inborn errors of metabolism, some of which can be treated with dietary changes and some which may require intervention. Blood testing via a heel prick, using a bacterial inhibition assay (Guthrie Test) is used to detect many of these disorders. Phenylketonuria (PKU) and congenital hypothyroidism are screened in all fifty states. The state of Georgia also tests for Galactosemia, Maple Syrup Urine Disorder, Homocystinuria, Sickle Cell Anemia, Congenital Adrenal Hyperplasia, and Tyrosinemia. Phenylketonuria is an inborn metabolic disorder affecting about one in every 10,000 to 15,000 Caucasian infants. It appears to be less frequent among other races, but racial frequency is not well known. The problem stems from an inability to metabolize phenylalanine, an essential amino acid (protein); unprocessed phenylalanine builds up in the brain, causing damage. Although, some infants with PKU may appear to be normal for months even though problems are developing. These include severe mental retardation, microcephaly, eczematous or oily skin, cerebral palsy, convulsions, dysplasia, projectile vomiting, hyperactivity with purposeless movements, an abnormal EEG and blocked pigmentation.

In bottle fed babies, the test is abnormal within 24 hours in 90% of cases and almost always by 48 hours of birth, if the baby has ingested adequate dietary protein. In a breastfed baby, the test should be done after the milk has been in for 48 hours to allow for sufficient buildup of protein. Breast milk contains less phenylalanine than formula, so it is important to test after the milk supply and nursing are well established, usually about a week after birth. Variant or mild forms of PKU require longer feeding to produce abnormal results.

In a hospital birth these tests are done before discharge and may need to be repeated in several days. In a home birth this test will need to be done by your pediatrician at their office, usually about a week after birth.