Newborn screening – the Guthrie test for Inborn Errors of Metabolism in Infancy

The Guthrie tests is a neonatal metabolic screening test, for an array of disorders and is now used worldwide for neonatal screening.

What is Newborn screening: It identifies healthy babies who are at risk for a disease at a later stage.

What happens with *Inborn Errors of Metabolism*: Most babies with the metabolic disorders screened by this test show no obvious signs or symptoms immediately after birth. For each of these disorders there is a problem in one of the biochemicals which are produced naturally in the baby's body. These conditions involve metabolic pathways leading to build up of toxic substances, resulting in damage to organs. Without a blood test these problems are hard to identify, as these disorders often do not show any symptoms until damage has occurred. Screening can detect these disorders before symptoms appear, and early treatment can be available to your baby. Some disorders are treated with special diets, and others with medications. If treated early, infants may grow up to lead a normal, healthy life. In a few cases, the disorders may not be completely treatable. The early diagnosis and treatment of the disorder will allow your baby the best chance of normal growth and development.

List of diseases include early detection of phenylketonuria, congenital primary hypothyroidism, galactosaemia, cystic fibrosis, urea cycle defects, and a range of disorders of amino, organic and fatty acid metabolism. Many of the inborn errors of metabolism present in the young infant with symptoms of an acute or chronic metabolic encephalopathy.

Best time to screen: do a heel prick blood test 48-72 hours after the first feed.

When a baby with a metabolic disorder is in the womb, the placenta clears away any abnormal biochemical levels as the baby's system produces them. When the baby is born, the baby's own system takes over and if the baby has a metabolic disorder, the biochemicals can accumulate to toxic levels or cause other problems. The baby's own system should be working properly at 48 hours of age, and this is therefore the best time to test the baby for metabolic disorders: when your baby's system is working independently, and before your baby gets sick.

Who should have a newborn metabolic screening test? It is strongly recommended that screening is done for all newborns. It is a voluntary test, but recommended due to severity of the illnesses.

About 1-2% of newborns screened will require follow up testing, whilst 0.1% of newborns screened will result in being diagnosed with a particular condition. The screening test does not replace investigation of symptoms, as screening does not detect all cases.

The medical aids currently do not cover the cost for this test (but this may change in the near future) and will cost around R900/test for 20 conditions (+/- R50/condition).

Created Dr L Smith 2015