Newborn Metabolic Screening Information Sheet

What is the Newborn Metabolic Screening Test?
The Virginia Newborn Screening Program is a state mandated special blood test that looks for almost 28 hidden birth anomalies. Hidden birth anomalies are problems in the body's ability to make and use hormones, proteins, sugars or blood cells. These rare anomalies (affecting 1 in 2,000 infants) are very difficult or impossible to detect by physical exam alone. Often times, when signs and symptoms of these anomalies appear, severe damage to your baby or death has already occurred. The Newborn Metabolic Screen is the earliest way to detect a hidden birth anomaly. The State of Oregon requires all infants be screen unless the parent declines in writing.

What birth defects are screened for?
Each newborn metabolic screening test looks anomalies in the follow categories

1. Endocrine (hormone) anomalies: congenital hypothyroidism and congenital adrenal hyperplasia.
2. Hemogloinopathies (blood anomalies): sickle cell anemia and three other defects
3. Biochemical or metabolism anomalies:
    a. Amino acid (protein) defects: phenylketonuria, maple syrup urine disease, and five other amino acid defects
    b. Carbohydrate (sugar): galactosemia
    c. Fatty acid anomalies: medium chain acyl-CoA dehydrogenese deficiency (MCADD) and seven other defects
    d. Organic acid anomalies: six defects
    e. Biotinidase deficiency

How is the test preformed?
In Virginia, it is required for all infants to have a newborn screening test. The screening occurs after the baby is 24 hours old. It is not done before that time, because the baby need to have had a chance to intake nourishment. The anomalies are screened for in the baby's blood. During the test, blood samples are obtained by pricking along the side of the baby's heel. Enough blood must be obtained to saturate five separate filters. Within 24 hours of collection, these filters are sent to the lab for analysis. The test cost approximately $54.

The parents are responsible to ensure that their baby is tested. Your midwife or the baby's pediatrician will be responsible of collecting the sample and receiving the test results.

What are the risks of the blood test?
The blood sample will be obtained by pricking the baby's heel. All precautions will be taken to insure minimal discomfort and chance of infection. Despite these efforts, your baby my feel pain, and experience slight redness and swelling at the puncture sight.

What if the test result is abnormal?
All test results will be sent to your midwife or noted follow up care provider. If your baby receives an abnormal test result, this does not mean that your baby has an anomaly. Instead, it means that further testing needs to be preformed. It is very important that an abnormal test result be further investigated by a specialist. Treatment of each anomaly is different. Some are treated with a special diet and others with medication. With attention to their particular case, people with these anomalies lead normal lives.

Where can I get further information?
You can go to the following websites for more information
Virginia State Public Health Laboratories
www.vahealth.org/vnsp/

Newborn Screen and Genetics Resource Center
http://genes-r-us.uthscsa.edu