homebirth & women's health

WomanWise

Informed Choice: Newborn Metabolic Screen

What is the Newborn Metabolic Screen?

Newborn screening is the process of testing young babies for certain potentially severe metabolic disorders. The newborn screen is also known as the "PKU test," as it was first used to screen for phenylketonuria (PKU). Screening now includes several other metabolic disorders – the exact list varies by state. Metabolic disorders are rare but they are usually serious. Some disorders may be life threatening. Others may cause delayed physical development, mental retardation, or other problems. All of these disorders can be managed or treated (usually by changing the child's diet) if detected early, although some have better treatments available than others.

In Maryland, Pennsylvania, Delaware and New Jersey, newborn screening is mandated by state law. The laboratory test is conducted on a few drops of blood taken by pricking your baby's heel. In addition, parents have the option of using the same blood sample to test for several other metabolic disorders (called supplemental screening).

What disorders are tested?

Six disorders are included in the PENNSYLVANIA newborn screen: congenital adrenal hyperplasia, congenital hypothyroidism, galactosemia, maple syrup urine disease, phenylketonuria, and hemoglobin disorders (such as sickle cell disease). The expanded screen also includes: organic acid disorders, fatty acid oxidation disorders, amino acid disorders, cystic fibrosis, biotinidase deficiency.

The NEW JERSEY newborn screen checks for 20 metabolic disorders: congenital adrenal hyperplasia, congenital hypothyroidism, galactosemia, hemoglobin disorders (such as sickle cell disease), phenylketonuria, biotinidase deficiency, cystic fibrosis, maple syrup urine disease, four types of fatty acid oxidation disorders, two types of urea cycle disorders, and six types of organic acidemias.

The DELAWARE newborn screen checks for 37 metabolic disorders: congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, hemoglobinopathies, biotinidase deficiency, cystic fibrosis, 11 amino acid/urea cycle disorders (including phenylketonuria), 10 organic acid disorders, and 10 types of fatty acid oxidation disorders.

The MARYLAND newborn screen checks for 39 meatabolic disorders: biotinidase deficiency, congenital adrenal hyperplasia, congenital hypothyroidism, cystic fibrosis, galactosemia, acylcarnitine abnormalities, and amino acid abnormalities (including phenylketonuria).

What are the benefits of screening?

All of these disorders can be tested for by using a single blood sample from your baby. This screening may be the only way to know if your baby has one of these rare disorders before he/she becomes sick. Early treatment can help your baby grow up as healthy as possible.

What are the risks of screening?

A test result may suggest a disorder when a baby does not have the disorder, which can cause parents to worry. An abnormal result does not mean that your baby has a disorder. Further testing will be required if a test result is abnormal. Furthermore, these screening tests may not catch all cases of these disorders. Even if the results of these screening tests are normal, there may be other medical problems that cannot be detected by these methods.

Refusing the screen could result in no or late detection of one of these disorders if your baby has one. Some of these disorders can lead to brain damage or death.

How is screening done?

Only one blood sample is required to test for all of these disorders. A few drops of blood will be obtained by pricking your baby's heel, then allowing the blood to dry on a piece of special filter paper, which is then sent to a screening laboratory for testing. The heel-prick procedure is generally uncomfortable for your baby, but you may hold your baby, or even nurse your baby, during the procedure.

Resources:

- American Academy of Pediatrics: http://www.medicalhomeinfo.org/screening/newborn.html
- March of Dimes: http://www.marchofdimes.com
- NJ Department of Health Newborn Screening Program: http://www.state.nj.us/health/fhs/nbs.
- PA Department of Health Newborn Screening Program: http://www.dsf.health.state.pa.us/health/cwp/view.asp?a=179&q=232592
- MD Department of Health and Mental Hygiene: http://dhmh.maryland.gov/labs/html/nbs.html
- DE Newborn Screening Program: http://dhss.delaware.gov/dph/chca/dphnsp1.html
- Save Babies Through Screening Foundation: http://www.savebabies.org

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I have been provided with written information about Newborn Metabolic Screening and have had the opportunity to ask questions. I understand the benefits and risks associated with newborn metabolic screening. I believe that my midwife has honored my right to make my own informed decision. I understand that Newborn Metabolic Screening is state-mandated and believe in my right to accept or decline any test or treatment for my child.

I take full responsibility for the health of my child, and I will ensure that if my infant displays any symptoms of a metabolic disorder, I will immediately have my infant checked by a healthcare provider with pediatric expertise. I understand that symptoms may not become immediately apparent and that the newborn metabolic screen is the best way to identify inborn errors of metabolism before they cause harm.

My choice for testing is indicated below.

_____ I choose to have my child tested for metabolic disorders using the state-mandated newborn metabolic screen at 7-10 days after birth.

_____ I choose not have my child tested for metabolic disorders using the newborn metabolic screen.

_____ Other: _____

Signed (mother): _____ Date: _____

Signed (partner): _____ Date: _____

Signed (midwife)_____ Date:_____