Birthwise Midwifery Care
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Newborn Screening

What Is Newborn Screening?

This is a screening test used to provide early detection of a number of rare but serious metabolic diseases:

Phenylketonuria (PKU): incidence 1 in 10,000 to 25,000. If not treated, PKU results in brain and nerve cell damage. It is caused by the body's inability to break down and use phenylalanine. A special diet low in phenylalanine prevents the mental retardation that would occur if left untreated.

Congenital Hypothyroidism: incidence 1 in 3,500. If not treated, this disorder results in mental retardation and poor growth. It is caused by the absence or abnormal development of the thyroid gland. Treatment with daily thyroid medication supplies the needed hormones for normal brain and body development.

Congenital Adrenal Hyperplasia (CAH): incidence 1 in 18,000. If not treated, this disorder can result in severe illness or even death. It is caused by the inability of the adrenal glands to produce normal amount of certain hormones. Treatment with the missing hormones allows the infant to be healthy by preventing large amounts of salt from being lost from the baby's body.

Sickle Cell Disease: incidence 1 in 10,000 in the general population, 1 in 400 in persons of African ancestry. Sickle Cell disease can cause many health problems beginning in infancy that can result in severe illness or even death. It results when hemoglobin (red blood cells) doesn't form or develop normally. Treatment with antibiotics reduces the serious infections that threaten affected infants and young children.

Other Significant Hemoglobinopathies: incidence 1 in 14,000 including hemoglobin C, D, E, and Alpha Thalassemias—These are abnormalities of the red blood cells that cause severe anemia which can result in organ damage, infection and death. Treatment may require periodic transfusions to treat anemia.

Biotinidase Deficiency: incidence 1 in 60,000. If not treated, this disorder can lead to neurological disorders, metabolic crisis, and death. It results from an inability to recycle the vitamin biotin. Treatment with oral biotin will prevent all damage.

Galactosemia: incidence 1 in 50,000. If not treated, this disorder leads to widespread damage to organ systems, developmental damages and possibly death. It is caused by an inability to metabolize a natural sugar called galactose, found in milk products including breast milk. Treatment is based on eliminating galactose and lactose sugars from the diet, which prevents the damage.

Homocystinuria: incidence 1 in 200,000. If not treated, this disorder leads to mental retardation, eye problems, bone problems and early death. It is caused by a defect in the metabolism of an amino acid, methionine, found in many foods including cheddar cheese, chicken and beef. Treatment includes a specific diet to limit methionine and related amino acids, which eliminates the damage.

Maple Syrup Urine Disease (MSUD): incidence 1 in 200,000. There are variable forms of MSUD which can cause problems ranging from mental retardation to death. There is a characteristic smell to the urine similar to maple syrup. This is caused by an inability to metabolize certain amino acids; leucine, isoleucine and valine, found in many foods. Treatment consists of a special diet low in these amino acids.

Medium Chain acyl-co Dehydrogenase (MCADD): incidence 1 in 20,000. This disorder causes severe problems with fasting or low blood sugar and can cause liver damage and death. It is caused by an inability to produce enough of an enzyme involved in the metabolism of medium chain fatty acids. Treatment involves careful avoidance of fasting, reducing fat in the diet, and supplementing with carnitine. Additional tests are occasionally added into the Washington State Standard Screening. If a family is interested in obtaining expanded newborn screening beyond what is offered above, there are laboratories that will screen for over 30 additional metabolic disorders for a small fee.

What are the Benefits of Screening?

The common thread among all of these rare disorders is that ***each can be treated*** **and** ***each must be treated early*** for babies with these diseases to live a normal life, avoid death, serious disease, or mental retardation.

When Should Testing Be Done?

The Washington State Department of Health requires midwives and birth centers obtain a blood sample test on baby between day 1 and 3 after birth (no later than five days of life) and strongly recommend a second test be done between days 10-14. (DOH screening pamphlet Pub #304-007 rev, 1/05)

How is the Testing Done?

A few drops of blood are taken from your baby's heel. These drops of blood are put on a special filter paper card that is dried and mailed to the Office of Newborn Screening for testing.

Are there Risks to Testing?

The risks relate to the puncture site; pain, a small risk of infection, excessive bleeding or bruising at puncture site are possible.

What Happens if a Test is Positive?

You will be notified and asked to bring the baby in to a specialty clinic for additional testing and to establish early treatment if needed.

For More Information:

Colorado State Department of Heath, www.cdphe.state.co.us/release/2006/061306.html
Another helpful web site is, www.uchsc.edu/newbornscreening/state.htm
March of Dimes, Newborn screening recommendations. http://www.marchofdimes.com