UW Medicine
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Appendix D

Screening Tests for Newborns

What you should know

What is newborn screening?

Newborn screening is a way to find out if a baby is at risk for serious health problems that may not be clearly seen at birth. State law requires that a blood sample be collected from every baby born in Washington state within 48 hours of birth. This sample is used to test for potentially life-threatening disorders.

Why is this screening important?

A newborn can look very healthy but still have a serious health problem. This screening finds problems that, if not found and treated early, can result in developmental delays, severe illness, or even death. Finding and treating these problems early can save a baby's life.

How is screening done?

A few drops of blood will be taken from your baby's heel. The sample is sent to the Newborn Screening Program at the State Public Health Laboratories in Shoreline. The hospital or provider who submitted the sample will receive the results within a few days.

A few drops of blood will be taken from your baby's heel for newborn screening.

When is it done?

The 1st blood sample should be taken when your baby is between 18 and 48 hours old. Early testing allows infants to be treated as soon as possible, if needed. The 2nd sample should be collected between 7 and 14 days old, but may be done when your baby is older. Your baby's provider may also ask for other tests.

Mother Baby Unit | Box 356078 | 1959 N.E. Pacific St., Seattle, WA 98195

Screening Tests

Amino Acid Disorders

- Argininosuccinic acidemia (ASA)
- Citrullinemia (CIT)
- Homocystinuria (HCY)
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)
- Tyrosinemiatype 1 (TYR-I)

Fatty Acid Disorders

- Carnitine uptake deficiency (CUD)
- Long-chain L-3-hydroxy acyl-CoA dehydrogenase (LCHAD) deficiency
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- Trifunctional protein (TFP) deficiency
- Very-long chain acyl-CoA dehydrogenase (VLCAD) deficiency

Organic Acid Disorders

- 3-hydroxy-3-methylglutaric aciduria (HMG)
- Beta-ketothiolase deficiency (BKT)
- Glutaric acidemia type 1 (GA-I)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemias (CblA,B and MUT)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PROP)

Other Disorders

- Biotinidase deficiency (BIOT)
- Congenital adrenal hyperplasia (CAH)
- Congenital hypothyroidism (CH)
- Cystic fibrosis (CF)
- Galactosemia (GALT)
- Hemoglobinopathies (Hb)
- Severe combined immunodeficiency (SCID)

Why are 2 blood samples needed?

Most problems can be found from the sample taken before the baby is 48 hours old. But, some may not be found until the baby is a little older.

Sometimes more than 2 samples may be needed. This does not mean your baby has a problem. The most common reason for asking for an extra sample is that the first results were unclear.

There is just 1 fee for the screening for each child. If more samples are needed, you will not need to pay more. But, your provider may charge a fee to collect the blood sample. If diagnostic tests are needed, they will involve extra costs.

What problems will the screening show?

For a complete list of health issues, please see the list at left.

What happens if a problem is found?

If the screening shows there may be a problem, your baby's provider will be contacted right away. Diagnostic tests will be advised so treatment can be started without delay.

How can I find out the results?

If you have questions about the results of your baby's screening, please talk with your provider. If your provider does not have the results, they should contact the Newborn Screening Program.

As a parent, may I refuse to do this screening?

Parents may refuse the screening tests for their baby only if this testing conflicts with their religious beliefs or practices. If this is true for you, be sure to tell the hospital staff or your provider.

Can my baby be screened for other problems?

The Newborn Screening Program only checks for the problems listed at left. But, there are other critical problems that can be found when your child is very young. These include *critical congenital heart disease* (CCHD) and hearing loss. All birthing hospitals offer these screenings for newborns.

Where can I learn more?

Talk with your provider or call the Newborn Screening Program at 206.418.5410 (toll free: 866.660.9050).

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