

**Newborn Metabolic Screening (PKU)**

**What is newborn screening?**

Newborn screening is a way to identify babies who are at risk for serious disorders that are treatable, but not apparent at birth. State law requires that a blood-spot specimen be collected from every baby born in Washington within 48 hours of birth. This specimen is used to test for potentially life threatening disorders. (Chapter 70.83 RCW and Chapter 246-650 WAC)

**Why is screening important?**

This screening identifies disorders that, if not detected and treated early, can result in developmental delays, severe illness or even death. A newborn baby may look perfectly healthy, but still have a serious disorder. Finding these problems early and treating them can prevent many serious complications. Fortunately, treatment is available to prevent or greatly reduce the effects of these disorders. Newborn screening tests are one important way to provide your baby with the best possible health care. A simple blood test can give you and your baby's health care provider information about your baby's health that you may not otherwise know.

**How is screening done?**

All tests are done from a few drops of blood taken from your baby's heel. The blood is collected on a special absorbent paper and sent to the Newborn Screening Program at the State Public Health Laboratories in Shoreline for testing. The hospital or health care provider who submitted the specimen is notified of the results within a few days.

**When should screening be done?**

Generally the first screening specimen should be collected when the baby is between 18 and 48 hours of age. This allows affected infants to be treated as soon as possible. The routine second specimen should be collected between 7 and 14 days of age, but it is still beneficial for older babies. Additional testing should also be done when requested by your baby's health care provider (pediatrician).

**Why are two specimens recommended?**

Most of the disorders will be detected on the first specimen, even if taken on the day of birth. The second specimen is recommended because some disorders may not be detected until the baby is slightly older.

Sometimes more than two specimens may be requested. This does not mean your baby has one of the disorders. The most common reason for requesting an additional specimen isthat the previous results were inconclusive.

There is only one charge per infant for the screening. Additional specimens are tested at no additional charge. However, your health care provider may charge a fee to collect the specimen. Diagnostic testing, if needed, will involve additional costs.

**What happens if a disorder is suspected?**

If the newborn screening test indicates a possible problem, your baby's health care provider will be contacted immediately. Diagnostic testing will be recommended so treatment can be started without delay if your baby is affected with one of the disorders.

**As a parent, may I refuse to have newborn screening done?**

The law gives parents the right to 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 health care provider.

**How can I find out the results?**

If you have questions about the results from your baby's screening tests, please contact your health care provider. If your health care provider does not have the results, he or she should contact the Newborn Screening Program to obtain a copy.

**What disorders are detected?**

**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)

For more information regarding the Newborn Metabolic Screen contact the Newborn Screening Program of Florida

**Phone:** 850-245-4201

**Email:** CMS.NBS@FLHealth.gov

**Address:** Newborn Screening Program

4052 Bald Cypress Way, Bin A06

Tallahassee, FL 32399

**Website:** <http://www.floridahealth.gov/programs-and-services/childrens-health/newborn-screening/>

**Consent**: I have read and understand the importance of the newborn metabolic screen and understand why screening is recommended. I have been provided resources for further research and education regarding the newborn metabolic screening. I have read and understand this information and have had an opportunity to ask questions. I will in no way hold Growing Families liable for my decision. I am fully aware of the risks of refusing the newborn metabolic screen and have freely chosen to take the following action:

**Initial** next to your decision(s):

\_\_\_\_\_\_ I consent to the initial Metabolic Screen and would like to do a newborn heel prick within 48 hours after birth.

\_\_\_\_\_\_ I refuse to do the Newborn Metabolic Screen for religious reasons

Date of Consent:\_\_\_\_\_\_\_\_/\_\_\_\_\_\_\_\_\_/\_\_\_\_\_\_\_\_

Clients Printed Name:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Clients Signature:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Midwifes Signature: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_