

Screening Tests

Amino acid disorders

argininosuccinic acidemia (ASA)
citrullinemia (CIT)
homocystinuria (HCY)
maple syrup urine disease (MSUD)
phenylketonuria (PKU)
tyrosinemia type 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)

Can my baby be screened for other disorders?

Yes. While our laboratory only screens for the disorders listed above, there are other dangerous disorders that can be detected in the newborn period such as critical congenital heart disease (CCHD). Also, early detection of hearing loss can lead to improved speech and language skills. All birthing hospitals offer this screening for newborns. For more information, speak with your baby's health care provider.

Privacy Practices

The Department of Health is required by law to protect the privacy of newborns and their families and assure that all specimen/information forms submitted for screening are protected from inappropriate use or access. Specific requirements are described in Section 246-650-050 WAC.

Copies are available upon request or on our website www.doh.wa.gov/nbs. Newborn screening specimen forms are kept in secured storage for 21 years in accordance with Chapter 70.41.190 RCW. After that time, specimens are destroyed. The Department of Health will destroy a specimen prior to 21 years upon receiving a written request from a parent or guardian and after all required testing has been performed.

Access to newborn screening specimens is restricted to Department of Health personnel and approved individuals who agree to strict confidentiality requirements. Written parental consent is required for any research involving identifiable information. Any release of specimens or other information must comply with the State's Uniform Health Information Act (Chapter 70.02 RCW) and the privacy and security provisions of WAC Chapter 246-650 Newborn Screening.

This pamphlet is available in: Cambodian, Chinese, Korean, Laotian, Russian, Somali, Spanish, and Vietnamese.

For people with disabilities, this pamphlet is available in an alternative format on request. To submit a request please call 1-866-660-9050.

Newborn Screening Tests & Your Baby



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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.



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 is that 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 disorders are detected?

For a complete list of conditions, please see the back the of this pamphlet

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.

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.

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.

Where can I get more information about newborn screening?

For more information, speak with your health care provider or contact the Newborn Screening Program using the information provided on the front of this pamphlet.