NEWBORN SCREENING

Saving Babies' Lives

FOR ADDITIONAL INFORMATION

Visit with your health care provider

Call the South Dakota Department of Health Newborn Screening Program at **1-800-738-2301**

Scan the QR code below for additional resources

SOUTH DAKOTA DEPARTMENT OF HEALTH
DOH.SD.GOV

SOUTH DAKOTA DEPARTMENT OF HEALTH
Newborn Screening Program
email: dohnewbornscreening@state.sd.us

NBS-002

QUESTIONS REGARDING SCREENING

What is Newborn Screening?

All newborns in South Dakota are required by law to have a blood test shortly after birth to screen for metabolic and other inherited disorders. The newborn screening helps identify babies who may have one of these disorders.

Why should my baby be screened?

Even if your baby looks healthy, he/she may have one of these disorders. Although these disorders are rare, they are usually serious. With early diagnosis and medical treatment, complications may be prevented. Without treatment babies with these disorders could suffer intellectual disabilities, seizures, or even death.

How will my baby be tested?

A few drops of blood from your baby's heel is all that is needed to test your baby. It is recommended that the test be performed 24-48 hours after birth.

When do I get the screening results?

Generally, parents are notified only if retesting or further testing is needed. You can ask about the results when you take your baby in for a regular check-up.

What if my baby needs to be re-tested?

If your baby's doctor asks you to bring your baby in for retesting or further testing, do so as soon as possible.

There are 3 main reasons why a repeat screening test may be needed: (1) There was a problem with the sample, (2) The test was done prior to 24 hours of age, or (3) The test result was abnormal. Generally, if the results of the repeat screening test are also abnormal, the doctor will discuss the need for further treatment or testing. It is important that your hospital and doctor have your correct address and phone number to contact you.

What happens to the blood spots after screening?

Most of the dried blood is used up during testing. The leftover blood spots are destroyed within 60 days. They are not used for research.

Can I refuse screening for my baby?

South Dakota law requires hospitals, doctors, and midwives to tell you about newborn screening. Because screening is so important, all babies should have these screens. If you don't want your baby screened, you must sign the refusal form. Ask your midwife or doctor for the form. Newborn screening can save your baby's life. The possible health risks of not screening are serious.

KREW'S STORY

After Krew was born, we were told about the newborn blood spot screening test. It's a simple heel prick for a spot of blood that is sent to a lab to check for metabolic and genetic disorders. Things were going great on our first full day at home until we got a call from the hospital. Something had come back abnormal on Krew's newborn screening. They told us it was a genetic condition called MCAD (Medium-chain acyl-CoA dehydrogenase). If this inherited condition is not found and treated quickly, it can cause low blood sugar, seizures, breathing difficulties, coma, and even death.

> *The newborn screening truly changed our lives and saved our baby's life.*

The hospital had us come back and Krew was admitted for 24 hours to monitor her blood sugar. Her diagnosis was confirmed. As first-time parents, we were overwhelmed and terrified of what could have happened had this not been caught by newborn screening. Our genetics team was fantastic giving us all the information and resources we needed to ensure that Krew would continue to live a healthy and happy life. The newborn screening truly changed our lives and saved our baby's life. As parents we are the best advocates for our children and choosing to do the newborn screening was easily one of the best decisions we have ever made. It truly has been lifesaving.

— MANDY S. MOM OF KREW KAY

Disorder	What is it?	What happens if not treated?	Treatment
Expanded Screening • **Amino Acid (AA) disorders** *(includes Homocystinuria, PKU testing, & Maple Syrup Urine Disease)*	These disorders are conditions that limit the body's ability to break down certain proteins.	These disorders are conditions that limit the body's ability to break down certain proteins.	Special diet.
• **Fatty Acid Oxidation (FAO) disorders** *(includes MCAD-medium chain acyl CoA dehydrogenase deficiency)*	These disorders affect the body's ability to make energy and to use the body's stored energy.	Can cause seizures, coma, and even death.	Making sure infants and children are eating and drinking regularly, especially when they are ill.
• **Organic Acid (OA) disorders**	These disorders lead to high levels of acids in blood and urine.	Can cause seizures, intellectual disability, and death.	Special diet and medicine.
Biotinidase Deficiency	The baby is unable to use the vitamin, biotin.	Babies grow very slowly and can become intellectually disabled.	Daily biotin supplement.
Congenital Adrenal Hyperplasia	The body cannot make enough of certain types of hormones.	Affects growth and development. The body has trouble maintaining a balance of salt causing vomiting, dehydration, and heart problems. If left untreated, death.	Special medicine.
Congenital Hypothyroidism	The thyroid gland doesn't produce enough of the hormone thyroxine.	The thyroid gland doesn't produce enough of the hormone thyroxine.	Medication – thyroxine.
Cystic Fibrosis (CF)	Causes mucus to build up in the baby's lungs and intestines.	Causes mucus to build up in the baby's lungs and intestines.	Regular medical care and a good diet.
Galactosemia	The body cannot use a certain sugar (galactose) found in dairy products.	Babies become very sick, have liver and eye damage, become intellectually disabled or even die.	Special diet.
Hemoglobinpathies (Sickle Cell Anemia)	Red blood cells tend to change from the normal round shape to an abnormal sickle shape, which may cause blockage of blood flow.	Babies with sickle cell disease can get very sick and even die from common infections.	Antibiotic treatment may lessen problems.
Severe Combined Immunodeficiency (SCID)	Affects the function of infection-fighting cells causing very little or no immune system.	It can be difficult or impossible to fight infections. If left untreated, it almost always causes death within the first year of life.	Bone marrow transplant.
Disorder Spinal Muscular Atrophy (SMA)	Spinal muscular atrophy (SMA) is a group of inherited neuromuscular disorders characterized by the loss of nerve cells in the spinal cord.	Progressive muscle weakness, including the muscles that affect feeding, swallowing, breathing and even death.	Medications and gene therapy.
Lysosomal Storage Diseases • **POMPE – glycogen storage disease II**	Genetic enzyme mutation. The body can't break down glycogen in the muscles for fuel. It causes progressive weakness to heart and skeletal muscles.	Infantile onset results in death within the first year.	Enzyme replacement therapy.
X-ALD (X-Linked Adrenoleukodystrophy)	X-ALD is a rare genetic condition that can cause problems in the brain and adrenal glands. Babies born with X-ALD may not show any signs at birth.	Without treatment, these problems can worsen quickly and may become fatal.	Treatable if caught early. Regular visits with a team of specialists to monitor disease progression is crucial to determining best treatment options.
Deafness or Hard of Hearing *This can range from slight to profound deafness in one or both ears.*	A hearing loss happens when any part of the ear is not working in the usual way. This includes the outer ear, middle ear, inner ear, hearing nerve, and auditory system.	Hearing loss can affect a child's ability to develop communication, language, and social skills. It can cause delays in their learning and behavior.	People can use devices like hearing aids or cochlear implants that make sounds louder. They can also learn to communicate using sign language, like American Sign Language (ASL).