

BC Newborn Screening Program

Parent Information Sheet

A Simple Blood Test Could Save Your Baby's Life



Why is my baby screened?

A small spot of your baby's blood can be used to get important information about his or her health. A newborn baby can look healthy but have a rare and serious disorder that you and

your doctor or midwife may not know about. Newborn screening finds babies who may have one of a number of these rare disorders. When these disorders are found and treated early, the chances of serious health problems are prevented or reduced later in life. If not treated, these disorders can cause severe mental handicap, growth problems, health problems and sudden infant death.

In British Columbia there are about 40 babies born each year (1 out of every 1,000) who are found to have one of these rare disorders.

How is my baby screened?

Your baby's heel is pricked and a few drops of blood are taken and put onto a special card. Your baby may cry, but taking the blood sample does not harm your baby. You can help your baby by holding and breastfeeding her or him while the blood is being taken. The blood sample is sent to the laboratory at BC Children's Hospital for testing. The same blood sample is used to screen for all disorders.

How soon after birth will my baby be screened?

The blood sample is usually taken between 24 and 48 hours after birth. This will be done before your baby leaves the hospital or, if a home birth, by your midwife at home.

What if my baby goes home before 24 hours old?

A blood sample will be taken in the hospital before leaving. Over 80% of disorders can be screened using this blood sample. You will be given instructions on how to have the sample repeated within 2 weeks. The purpose of the second sample is to double check the few disorders that can be missed on the first (early) screen.

Can I wait and have my baby tested later?

The earlier these treatable disorders are found, the better the outcome for babies with these disorders. It is strongly advised that your baby not leave the hospital without a blood sample being taken. If you decide you do not want your baby to have a blood sample taken before he or she leaves the hospital, you will be asked to sign a form to show you understand the reasons for the test and the possible outcome for your baby if your baby is not tested and has one of these disorders.

If your baby is under the care of a Registered Midwife, the midwife may review options with you to have the blood sample drawn at home.

How do I find out the results of the screening?

Your baby's screening results are reported to the hospital where your baby was born and your baby's doctor or midwife.

What does it mean if the screen is negative?

A negative screen means that the chance that your baby has one of these disorders is very low. Very rarely, the test may miss a baby with one of these disorders.

What does it mean if the screen is positive and what happens next?

A positive screen tells that there might be a problem. It does not mean that your baby has one of these disorders, but it is possible. More tests are needed.

Will screening for these disorders find anything else?

Screening for sickle cell disease and cystic fibrosis may also tell if your baby is a carrier for one of these disorders. Babies who are carriers are healthy and no more likely to get sick than any other baby. If your baby is a carrier, you will be provided with more information to find out what this means for your baby, yourself and your family.

Which disorders are included in the Newborn Screening?

In British Columbia, babies are screened for 22 rare but treatable disorders. These include:

Metabolic disorders. These occur when the body is not able to break down (metabolize) certain substances in food like fats, proteins or sugars. These substances can build up in the body and cause serious health problems. Serious health problems can usually be prevented with early treatment.

Endocrine disorders. Babies with endocrine disorders of either the thyroid or adrenal glands make too little of certain hormones. Babies with these disorders can receive hormones to replace the ones their bodies cannot make. Replacement of thyroid hormone prevents growth problems and mental handicap. Replacement of adrenal gland hormones can prevent serious health problems such as shock or unexpected death.

Blood disorders. Blood disorders happen when the part of the red blood cell that carries oxygen (hemoglobin) throughout the body is changed. Hemoglobin is important because it picks up oxygen in the lungs and carries it to the other parts of the body. Serious health problems can be prevented through medicines and special treatments.

Cystic Fibrosis. Cystic fibrosis is an inherited life-limiting disorder. It causes thick mucus to build up in the lungs, digestive system (and pancreas) and other organs. Most people with CF get chest infections. They also have problems digesting their food and, as a result, they may not gain weight as well as they should. Early treatment can be started with medicines and physical therapy that help babies with cystic fibrosis digest food and keep their lungs clear of mucus. CF affects about 1 in every 3,600 babies in BC.

What if the results show that my baby has one of the disorders after all the tests are done?

Your baby will need treatment from a doctor who specializes in the disorder. You will be referred to a specialist right away. Treatment can start in a few days.

What happens to my baby's blood spot card when the testing is complete?

Your baby's card with the leftover blood will be kept for 10 years in secure storage by the BC Newborn Screening Program. Occasionally, the dried blood spot samples may be used for other purposes after the testing is finished. These include 1) re-running a test if the first test result was not clear; (2) trying to find the reason for a health problem that has developed later in a child's life or trying to find the cause of an unexplained illness or death of a child; (3) checking the quality of testing done by the laboratory to make sure that results are accurate; and (4) developing better tests for screening of disorders. Samples may also be used for health research if the research has been approved by a Clinical Research Ethics Board. In these cases, all information that may identify the baby is removed.

If you do not wish your baby's stored blood spot card to be used for these purposes, you may fill out a form called a Directive to Destroy Leftover Newborn Screening Blood Samples and send to the BC Newborn Screening Program. See website for details.

If you need more information:

Talk to your doctor or midwife. Visit the Newborn Screening website at www.newbornscreeningbc.ca

We are committed to protecting the privacy of personal information:

The BC Newborn Screening Program collects, uses and discloses personal information only as authorized under section 26 (c) of the BC Freedom of Information and Protection of Privacy Act and other legislation. We take all reasonable steps to make sure that personal information is treated confidentially, is only used for the intended purpose and is securely stored. Should you have any questions regarding the collection, use or disclosure of your baby's personal information, please contact the BC Newborn Screening Program at (604) 875-2148.

Disorders Screened and Test Accuracy

1. What disorders does the newborn screen detect?

- BC's Newborn Screening Program screens for 22 disorders.

Table 1: Disorders Screened

Grouping	Metabolites Measured	Disorder	Abbrev.
Metabolic Disorders			
Amino Acid Disorders	Amino Acids	Phenylketonuria	PKU
		Maple Syrup Urine Disease	MSUD
		Citrullinemia	CIT
		Argininosuccinic Acidemia	ASA
		Homocystinuria	Hcy
		Tyrosinemia, Type I	Tyr I
Fatty Acid Oxidation Disorders	Acylcarnitines	Medium-chain Acyl-CoA Dehydrogenase Deficiency	MCAD
		Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD
		Trifunctional Protein Deficiency	TFP
		Very-long Chain AcylCoA Dehydrogenase Deficiency	VLCAD
Organic Acid Disorders	Acylcarnitines	Propionic Acidemia	PROP
		Methylmalonic Acidemia	MUT
		Cobalamin Disorders	Cbl A, B
		Glutaric Aciduria, Type I	GA I
		Isovaleric Acidemia	IVA
Galactosemia	GALT enzyme activity	Galactosemia	GALT
Endocrine Disorders	Thyroid stimulating hormone (TSH)	Congenital Hypothyroidism	CH
	17OH-progesterone (1 st tier) Steroid panel (2 nd tier)	Congenital Adrenal Hyperplasia	CAH
Hemoglobinopathies	Hemoglobin HPLC	Sickle Cell Disease	HbSS
		Sickle Cell/Hemoglobin C	HSC
		Sickle Cell/ β -thalassemia	HbS/ β -thal
Cystic Fibrosis	Immunoreactive trypsinogen (IRT) (1 st tier) CFTR mutation panel (2 nd tier)	Cystic Fibrosis	CF