



Newborn Screening: A healthy start leads to a healthier life

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Getting the best start

As a new or expecting parent, your baby's health is important to you. To help your baby get the best start in life and stay healthy, your newborn – and every other newborn in Ontario – will be screened for at least 28 different disorders. Although most babies with these disorders look healthy at birth, they may be at risk of having serious health problems – including developmental disabilities, recurrent sickness and even death – if their disorder is not detected and treated. Early identification could save your baby's life, and is the key to effective treatment.

Early detection leads to early treatment

Individually, these disorders are rare. As a group, they will affect about 150 out of approximately 140,000 babies born each year in Ontario. Even if there is no family history of these disorders, your baby is still at risk. By testing newborns within the first days of their lives, disorders can be treated early, reducing the chance of serious health problems later in life.

This pamphlet will provide you with information about the Ontario Newborn Screening Program (ONSP), and answer questions that you may have about the newborn screening test.



A small test, producing big benefits

In order to perform the screening test, a small sample of blood is taken from your baby by pricking the heel. The blood is collected on a special paper card and then sent to the ONSP for testing. You should be given an information letter that includes a reference number in the top right-hand corner. This number can be used to link to your baby's sample. Blood samples can be taken anytime between one day (24 hours) and seven days after your baby is born. The best time to collect the blood sample is when your baby is between two days (48 hours) and three days (72 hours) old. If your baby is tested before one day (24 hours) of age, your baby's health care provider should repeat the test within five days, at the baby's first checkup.

Screening results: high risk and low risk

A screening test only shows whether there is a high or low risk that your baby has a disorder. It is important to understand that the test does not make a diagnosis of a disorder, but only identifies babies who need to be referred for further testing. Once the laboratory has received and analyzed your baby's blood sample, one of the following will occur:

1. Your baby screens negative for all the disorders.

A report will be sent by mail to the hospital or health care provider that sent in your baby's sample. It will be filed in your baby's medical records. More than 99 per cent of babies screened will receive a "screen negative" result. This means there is a very low risk that your baby has one of these rare disorders. On very rare occasions, the newborn screening test may miss a baby with one of these disorders.

2. The laboratory may need another blood sample.

It may be that the first sample was not taken properly, there wasn't enough blood to complete the testing, or there was some other problem with the sample. In this case, the ONSP will contact the



hospital or health care provider that sent in the sample and ask them for a new sample. Your health care provider will call you to let you know that your baby's test needs to be repeated and arrange for another blood sample to be taken as soon as possible.

3. Your baby screens positive for one of the conditions. A screen positive result **does not necessarily** mean that your baby has a disorder; however, it does mean that your baby has an increased chance to have the disorder. A doctor, genetic counsellor or nurse from the ONSP may contact your health care provider or you directly and ONSP doctors will refer your baby to doctors at a regional treatment centre for follow-up diagnostic testing to find out if your baby has the disorder. In some cases, the ONSP's health care providers work directly with local health care providers and families to arrange testing. If a diagnosis of a disorder is made, your baby will be provided with treatment and your family with counselling and advice.

The treatment centre doctors will provide the ONSP doctors with follow-up information about your baby. This includes medical information, which tests were done, the results

of those tests, and whether or not your baby has the disorder. This feedback allows the ONSP to make sure that your baby receives appropriate and timely care. It also helps to ensure that the ONSP is able to provide quality service to your baby, and all babies born in Ontario. The health care providers at the ONSP review this information and may contact you, your health care provider, or the regional treatment centre if they have questions about your baby's care. You may choose not to share this information, in which case, please discuss this with your health care provider, a member of the health care team at your treatment centre, and/or contact the ONSP.

The ONSP also issues a report by mail to the hospital or health care provider that sent in your baby's sample, which will be filed in your baby's medical records. It is important to remember that less than 1 per cent of babies tested will receive a "screen positive" result.

What does the testing look for?

The testing done for the ONSP looks for at least 28 disorders that can cause health problems in babies and children.

These disorders include:

Metabolic disorders: When the body is not able to break down (metabolise) certain substances in food like fats, proteins or sugars, they can accumulate in the body and cause serious health problems.

Endocrine disorders: The endocrine system, which is responsible for producing hormones, can sometimes produce too much or too little of some hormones, leading to sickness or developmental disabilities.

Blood disorders: Oxygen is carried through the body in the hemoglobin found in red blood cells. If hemoglobin is not formed or not working properly, red blood cells break down, leading to health problems including severe anemia and serious infections.

Cystic Fibrosis: A condition that causes mucous to build up in the lungs, digestive system and other organs.

What is ONSP?

The Ontario Newborn Screening Program (ONSP) is administered by the Children's Hospital of Eastern Ontario (CHEO) under the stewardship of the Ontario Ministry of Health and Long-Term Care. ONSP is committed to providing the best possible screening for newborn babies across the province and to do this we need to collect and use a small blood sample from babies as well as some information about them and their families. ONSP is committed to keeping your information safe and confidential, and follows rules set out in law about collection of health information, uses of the information for your care, analysis and research, and limited disclosures of that information.

Want to learn more?

For more information about the Ontario Newborn Screening Program, speak to your doctor or midwife, or visit the ONSP website at www.newbornscreening.on.ca, or call **1-866-532-3161** (TTY 1-800-387-5559).