



NEWBORN SCREENING
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ONTARIO



NEWBORN SCREENING AND YOUR BABY:

**A healthy start
leads to a healthier life**

EARLY DETECTION LEADS TO EARLY TREATMENT

As a new or expecting parent, your baby's health is important to you. Although most babies look healthy at birth, they may be at risk of having serious health problems if they have a disease that is not detected and treated early. To help your baby get the best start in life, your newborn – and every other newborn in Ontario – will be offered screening for rare, serious diseases. While these diseases are rare, as a group they affect about 200 out of the approximately 140,000 babies born each year in Ontario. The goal of screening is early detection – so that treatment can be started early and better health can be achieved.

Newborn screening is not mandatory. It is considered the standard of care for every baby and is highly recommended. You have the right to choose to accept or decline newborn screening for your baby. You may wish to discuss this decision with a health care provider. Newborn screening is the only way to find babies with these diseases early enough to prevent serious, long-term health problems.

A small test, producing big benefits

In order to perform the screening test, a small sample of blood is taken from your baby. It is usually taken between 24–72 hours after birth by pricking the heel and placing the blood on a special paper card. You should be given an information letter that includes a reference number at the top. This number can be used to link to your baby's sample. The sample is then sent to Newborn Screening Ontario (NSO) where it is tested for rare diseases including:

- Metabolic diseases
- Endocrine diseases
- Sickle Cell Disease (SCD)
- Cystic Fibrosis (CF)
- Severe Combined Immune Deficiency (SCID)

A full list of diseases is available on the NSO website www.newbornscreening.on.ca

Screening results: high risk and low risk

The screening results will show if your baby is at higher or lower risk for the diseases, but they are not yes or no tests (also known as “diagnostic” tests). If your baby ever develops symptoms of a disease, your baby’s doctor should do the appropriate diagnostic testing. As well, it is important to remember that newborn screening does not test for all serious medical problems.

A screen negative result means that the chance your baby has one of the diseases is very low and no follow-up testing is needed. More than 99% of babies screened will have a screen negative result.

A screen positive result means that your baby has a higher chance of having one of the diseases and needs further testing. It does not necessarily mean that your baby has a disease. In this case, Newborn Screening Ontario (NSO) doctors will refer your baby to specialists for follow-up testing. You will be contacted by your health care provider (HCP) or a specialist if your baby has a screen positive result.

You may be asked to bring your baby back for a repeat screening sample.

This will happen if:

- Your baby’s first sample was taken before 24 hours of age
- Not enough blood was taken
- The sample was of poor quality

Your hospital or midwife will contact you if a repeat sample is needed. It is important that the repeat sample is taken as soon as possible so that your baby gets the full benefit of newborn screening. Needing a repeat sample does not mean there is anything wrong with your baby.

Results go to the hospital or HCP that did the test, by mail or electronically through the Ontario Laboratories Information System (OLIS). Your baby's HCP may also be able to get the results through OLIS. NSO does not release results directly to parents/guardians.

Will screening for these diseases find anything else?

Sometimes screening will show that a baby has a disease other than the targeted diseases. If something like this is found, a specialist will discuss this with you.



Screening for Sickle Cell Disease may also detect if your baby is a carrier (also known as trait). Babies who are carriers are healthy and do not need any special medical treatment. Carrier results are available by request. Information on how to obtain your baby's carrier results is on the NSO website – or, ask your baby's health care provider.

Protecting your baby's privacy and confidentiality

NSO is committed to keeping your baby's blood sample and information safe and confidential, following the rules set out in law about their collection and use. They can be used for health care, analysis, quality assurance and research. Personal health information (PHI) is shared between health care providers involved in newborn screening and diagnosis to make sure that your baby gets the care and follow-up he or she needs. PHI is also shared with OLIS. You may not want this information shared, in which case, please make your wishes known to your HCP and/or contact NSO.

After testing is finished, your baby's sample is stored in a secure facility as part of your baby's medical record. It is stored for 19 years and then destroyed. Samples are stored so they can be used to make sure that the newborn screening tests are working properly. This can benefit your baby and all babies in Ontario. Sometimes a baby's sample is needed in the future by his or her



doctor to run extra tests. Storing the sample means it is available if needed. Other possible uses for stored samples include testing by other laboratories at your request, the development of new or improved NSO tests, uses for which a legal warrant or court order is issued, and research approved by a research ethics board. In general, information that can connect your baby to the bloodspot sample can only be shared if you okay it in writing or if it is required by law.

If you would prefer that your baby's sample not be stored, you can ask NSO to destroy the sample or release it to you. For more information please contact NSO.

For more information

If you have any questions about newborn screening in Ontario, please talk to your health care provider or contact *Newborn Screening Ontario* directly.

Website

www.newbornscreening.on.ca

Phone

Toll Free: 1-877-NBS-8330

(1-877-627-8330)

8:00 AM – 16:00 PM

Email

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