



NEWBORN SCREENING ONTARIO  
DÉPISTAGE NÉONATAL 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. As a group these diseases affect about 225 out of the approximately 143,000 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 your doctor, midwife or other 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 – 48 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)

Newborn screening also includes Critical Congenital Heart Disease (CCHD).

A quick and painless test called pulse oximetry measures the level of oxygen in the baby's blood. Results are available right away.

NSO, in collaboration with the Ontario Infant Hearing Program, offers screening for risk factors related to hearing loss using the newborn screening sample. This additional testing is performed with a parent or guardian's consent taken at the time of hearing screening.

A full list of diseases is available on the NSO website: [\*\*newbornscreening.on.ca\*\*](http://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. Please also keep in mind 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. If your baby’s HCP’s are concerned about your baby’s Critical Congenital Heart Disease (CCHD) screening results, they will arrange more tests at the time of the screen.

## **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.

Some babies need to have their CCHD screen repeated because it was incomplete. If your baby's CCHD screen was incomplete, you will be contacted by your HCP or the hospital that performed the screen about next steps.

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 some diseases 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**

Newborn Screening Ontario (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 providing health care, 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 needed. PHI is also shared with the Ontario Laboratories Information System (OLIS). If you do not want this information shared, please make your wishes known to your health care provider 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 ensure the quality of the newborn screening tests. This can benefit your baby and all babies in Ontario. Another possible use of the sample is testing as part of the Expanded Hearing Screening collaboration with the Ontario Infant Hearing Program. This would only be done with your consent. Your baby's sample could be 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 newborn screening sample can only be shared if you agree to this 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**

[newbornscreening.on.ca](http://newbornscreening.on.ca)

### **Phone**

Toll Free: 1-877-NBS-8330

(1-877-627-8330)

8:00 AM – 16:00 PM

### **Email**

[NSO@cheo.on.ca](mailto:NSO@cheo.on.ca)

### **Mail**

Newborn Screening Ontario

Children's Hospital of Eastern Ontario

415 Smyth Road

Ottawa, Ontario K1H 8M8

### **YouTube**

[Youtube.com/user/NBSOntario](https://www.youtube.com/user/NBSOntario)

### **Twitter**

[@NBS\\_Ontario](https://twitter.com/NBS_Ontario)