NEWBORN SCREENING

Newfoundland & Labrador – Patient Information

NL Health Services

What is newborn screening?

Newborn screening identifies babies at risk of having rare, but serious medical conditions and is offered to all babies in Newfoundland and Labrador (NL) and across Canada. Screening is important because:

- affected babies may not show any signs or symptoms of illness at birth.
- early detection allows for early treatment and better health.
- without screening, a condition could remain undetected. By the time symptoms appear, health and development may already be impaired.

How is newborn screening done?

A small blood sample is collected 24-72 hours after birth. Your baby's heel is pricked, and the blood is placed on special paper. The sample is tested in Ottawa and results are sent to your local health care team. If the result is positive, you would be contacted within 1-2 weeks.

What is being screened for?

- **Metabolic Disorders**: Babies with metabolic disorders are unable to break down certain substances in foods, like fats, proteins, or sugars. This can cause serious health problems if not treated.
- **Congenital Hypothyroidism (CH)**: Babies with CH have a thyroid that is too small, in the wrong place, not working normally, or is missing. If untreated, CH can lead to intellectual disability and abnormal growth.
- **Cystic Fibrosis (CF)**: CF causes damage to the lungs and digestive system. Symptoms may include frequent lung infections, cough, and poor weight gain.

What are the possible screening results?

The screening results will show if your baby is at high or low risk for certain conditions:

- **Screen Negative**: This is the most common screening result. This means that there is a low chance that your baby has one of these conditions. No follow-up testing is needed. You will not be contacted.

- **Screen Positive**: This result usually does not mean that your baby has a condition. It means that your baby has a higher chance of having one of these conditions. Further testing is usually required. You will be contacted to discuss the result and follow-up testing. If a positive result is confirmed and a diagnosis is made, the Provincial Medical Genetics Program will provide support, information, and will help to arrange ongoing follow-up for your child.
- You may be asked to bring your baby back for repeat testing. This could be for a number of reasons including if the sample was poor quality, collected at the wrong time, or your baby was premature. It is important to get a repeat sample as soon as possible so that your baby gets the full benefit of newborn screening.

Will newborn screening find anything else?

This testing may show that a baby has a risk for other diseases. If this happens, you will be contacted to discuss possible follow-up testing. Newborn screening may also detect if your baby is a carrier. Babies who are carriers are healthy and do not need any special medical treatment. Please keep in mind that newborn screening does not test for all serious medical problems.

Can I refuse?

Newborn screening is the standard of care for every baby. It is recommended as it may help prevent serious health problems. However, it is your right to decline newborn screening for your baby. You may wish to discuss your choice with your health care provider. Newborn screening is the only way to find babies with these conditions early enough to prevent serious health problems.

Use and storage of samples

Samples are sent back to NL and kept indefinitely. Samples can be used to develop new or better screening tests. Samples could also be used for anonymous research approved by a research ethics board.

More questions?

If you have any questions about newborn screening in NL, please talk to your healthcare provider. The Provincial Medical Genetics Program and newborn screening coordinator can be contacted at 709-777-4363.