Newborn blood screening

For more information on screening

Talk to your healthcare professional or go to

Québec.ca/screening

For information on the test

CHU de Québec – Université Laval
1 855 654-2103 (toll free)

25-918-01A

Santé et Services sociaux Québec

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Québec

Newborn screening is designed to detect certain rare diseases as early as possible, since they are not apparent at birth. These diseases must be treated quickly to prevent serious and permanent damage. Early treatment will also help improve the health and quality of life of most of the affected children.

Blood screening test is available to **all newborns in Québec**.

Newborn screening is recommended but voluntary. If you are unsure about screening, talk to your healthcare professional.

Since April 28, 2025, screening for rare diseases is conducted exclusively by taking a blood sample from the baby's heel at birth. Urine screening with a blotting paper placed in the diaper is no longer performed for babies born after this date.

Screening tests

Blood sampling is a simple, safe test that can detect a number of diseases.
It is performed as follows:

- During pregnancy follow-up, health professionals inform parents about screening to help them make an informed decision about whether or not to participate.
- When the baby is born, nurses or midwives check that the parents have received all the information about screening.
  - **If the parents refuse screening**, they sign the sample collection form after checking the box confirming their refusal.
- **If the parents agree**, the nurse or midwife takes a few drops of blood from the baby's heel, ideally at 24-48 hours of life.
- The laboratory analyzes the sample. A call is made to the parents in the event of a positive result for a detected disease, or if the sample needs to be repeated.

Which deseases are screened for?

Newborn screening is designed to detect rare diseases. If left untreated, these diseases prevent the body from functioning normally and may lead to organ damage, including the brain. Some may even be life-threatening.

Metabolic diseases:

These diseases prevent the normal use of fats, glucose or proteins provided by food and cause toxic waste to build up in the body. These diseases can lead to problems affecting the brain, liver or heart, and can threaten the child's life.

Congenital hypothyroidism:

It prevents some hormones from functioning normally and causes stunted growth or intellectual deficiencies.

Hemoglobin diseases:

These diseases cause abnormally shaped red blood cells and prevent normal blood flow. This may lead to painful crises and a greater risk for infection.

Cystic fibrosis:

This disease produces thick, sticky secretions that prevent the lungs and pancreas from functioning normally. This can cause repeated infections and poor food absorption, which slows the child's growth.

Spinal muscular atrophy:

This disease affects the nerve cells that control voluntary muscle movement, hindering the child's ability to hold up their head, sit, walk, eat, and eventually to breathe. This disease can be life-threatening.

Severe combined immunodeficiency (SCID):

This disease affects the immune system, which is responsible for defending the body against infections. The body is unable to effectively fight off germs (bacteria, viruses and fungi), which can be life-threatening.

Test results

No disease detected

Newborn screening is designed to detect rare diseases. Most newborn screening test results are normal. In this case, you will not be contacted.

Repeat sampling

You may be contacted if another sample is required because the first was not usable or the results need to be double-checked. There is no need to worry; just follow the instructions that you are given to collect a new sample right away.

Positive result

If your child's test results indicate they may have one of the screened diseases, you will be contacted within a few weeks. You will be quickly referred to a specialist for further testing. If these test results confirm that your child has the disease, they will receive the care they need.

Unexpected result

Sometimes, a disease not targeted by the screening is found in a child. If this happens, the child will be referred to the appropriate resource for medical care.

Care and treatments

Early screening for these diseases ensures that newborns receive treatment soon after birth. For some diseases, treatment may lead to a cure, while for others, it aims to improve the health and quality of life of the majority of affected children.

These treatments may include:

- a special diet;
- medications or vitamins;
- a specific therapy for the identified disease;
- specialized medical care.

Screening limitations and drawbacks

While highly effective, screening does have its limitations. Although rare, the following situations may occur:

- Treatment may be started as a preventive measure, pending the results of further testing. If the tests show that your child does not have the disease, the treatment will be stopped;
- The diagnosis may be difficult to confirm and doctors may need to see your child several times to conduct other tests before confirming whether or not they have the disease;
- Your child may still experience some effects of the disease despite the screening and the treatment offered;
- Your child may have one of the screened diseases but it may not be detected during screening.

Furthermore, the screening process may cause you to worry about your child's health. Throughout the screening process, the care team will be there to answer your questions and offer you any support you may need.

Carriers of faulty genes associated with certain diseases

For hemoglobin diseases (such as sickle cell anemia), information about your child's carrier status is available upon request. **Carriers are not sick and are not at risk of developing the disease.**

To obtain this information, fill out the form available on the website **Québec.ca/sickle-cell-anemia**

You can also request the information from your doctor.

Sample retention and privacy protection

Personal information collected during the screening process, and blood sample, are kept strictly confidential. Some personal information may be shared with healthcare professionals so that they can provide the child with the necessary care or for quality assurance purposes. Samples yielding normal test results are kept for five years by the laboratory that conducted the tests. Samples yielding abnormal results are kept for a longer time.