Blood and Urine

Newborn Screening

UN QUÉBEC POUR TOUS

Québec

The aim of newborn screening is to detect, as early as possible, certain diseases that are present at birth but not yet clinically apparent. These diseases may require immediate medical attention in order to prevent serious and permanent damage to children affected by them. Many of the children who benefit from early treatment will develop normally into healthy adults.

Every child born in Québec qualifies for screening. Two different tests are available in hospitals and birthing centres. The first uses a blood sample, while the second uses a urine sample. These two tests are complementary, since illnesses of the blood and those found in urine are different.

SCREENING TYPE	WHEN?	HOW?	WHERE?	WHO?
BLOOD TEST	24-48 hours after birth	Some drops of blood are taken from the baby's heel	Hospital or birthing centre	Nurse or midwife
URINE TEST	21 days after birth	A small quantity of urine is taken from the baby's diaper	At home	Parents You will receive a testing kit and instructions on how to use it.

Diseases detected by screening tests

Diseases detected are rare, but serious. If left untreated, they may have major effects on the baby's organs and brain. They can even threaten the baby's life.

There are 3 groups of detectable diseases:

Metabolic diseases:
These diseases interfere with normal food digestion and cause toxic waste to accumulate in the body. Examples are Phenylketonuria (PKU), Type 1 Tyrosinemia and urea cycle disorders.

Endocrine diseases:
These diseases, for example congenital hypothyroidism, prevent normal hormone function and may be responsible for stunted growth and mental retardation.

Hæmoglobin diseases:
These disorders, for example sickle cell anemia which causes deformed red blood cells, may be responsible for pain crisis, higher risk of infection and various other complications.

The diseases targeted in the screening process can take various forms. Only the most serious and the most common forms are identified.

Screening for sickle cell anemia gene carriers

The sickle cell anemia screening test is special in that it also enables detection of genetic carriers of the disease. Carrier status information **is not automatically provided**.

Carriers of the gene are not sick and are no more likely to become sick than any other children. However, knowing his or her carrier status will be useful to your child in later life. At that time, if the child's spouse is also a carrier, they may have a child affected with sickle cell disease.

If you wish to receive this information, you may apply directly by filling out the request form on the Web site of the ministère de la Santé et des Services sociaux: **www.msss.gouv.qc.ca/depistage-neonatal**. Alternatively, you may ask your doctor to apply on your behalf.

Screening results

The diseases detected in the screening process are rare. Most often, screening results are normal, in which case parents are not contacted.
No news is good news!

If any of the diseases being screened are detected in your child's blood or urine, you will be contacted 2 or 3 weeks after the test. An appointment will then be immediately set at a specialized hospital, where additional testing will be done. If the additional tests confirm that your baby has the disease, all needed care will be provided.

You may also be contacted in the event that a second blood or urine sample is needed. There is no reason to be concerned if this happens, since it can simply mean that the first sample could not be used. If you are contacted, follow the instructions without delay.

Types of treatment

Early identification of these diseases in newborns enables treatment to begin soon after birth. In most cases, the treatment received by the child will improve his quality of life even if the disease cannot be completely cured.

Here are some of the treatments that your child could receive:

- A special diet
- Preventive treatment with antibiotics medications and/or vitamins
- Specialized medical monitoring

Limits and drawbacks of screening

While highly effective, screening does have some limits. In rare situations:

- The screening test might not detected the targeted disease in your child
- The diagnosis may be difficult to confirm and the doctors may need to examine your child several times
- A preventive treatment could be started but subsequently abandoned if further analysis reveals it to be unnecessary
- The treatment may not produce the desired benefits and your child could continue to be affected by the disease

Participation in screening tests

Blood and urine screening is available to all Québec newborn babies.

Participation is on a voluntary basis but is recommended.

If you are unsure about blood screening, talk about it with your health professional. If you prefer that your child not be screened, tell the nurse before a blood sample is taken. You will need to sign a form confirming your decision to decline the screening test. With respect to urine screening, your wish to participate will be confirmed by sending your baby's sample to the test centre.

Sample retention and privacy protection

Personal information gathered during the screening process as well as blood and urine sample results are retained on a confidential basis. They will only be used for quality assurance and ongoing service improvement needs.

Learn more

Speak to your health professional or go to the Web site of the ministère de la Santé et des Services sociaux:
www.msss.gouv.qc.ca/depistage-neonatal.

Tests information

Blood screening
Centre hospitalier universitaire de Québec
1 855 654-2103

Urine screening
Centre hospitalier universitaire de Sherbrooke
1 855 905-5253

13-918-05A

Santé et Services sociaux Québec