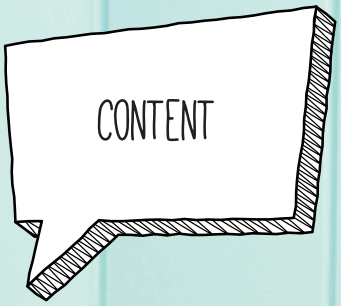
LNS
LUXEMBOURG

LE GOUVERNEMENT
DU GRAND-DUCHÉ DE LUXEMBOURG
Ministère de la Santé
et de la Sécurité sociale

Direction de la santé



WHY IS THE EARLY DETECTION OF CERTAIN DISEASES IMPORTANT?

Neonatal screening tests can detect certain diseases in babies that can have serious consequences if they are not treated quickly, and which can stall the physical and psychological development of the child. This is why newborn screening tests are carried out in the first few days of your baby's life.

WHY TEST ALL NEWBORNS?

Affected children can look healthy when they are born. There is nothing to indicate that they are suffering from these diseases. It is therefore important to test all babies. These tests do not pose any danger to your child's health.
The diseases that we screen for are rare – they only affect one child in several thousand. The probability of your baby being affected is low.

WHEN ARE THE TESTS DONE?

Between the second and third day of life after birth, your child will have a blood sample taken. All the tests are carried out on a few drops of blood taken at the heel and collected on blotting paper.

The blotter card is sent to the National Health Laboratory, which analyses it using the appropriate methods for each disease.
Parental consent is required for screening. This is done by signing the Guthrie test card.

HOW WILL YOU RECEIVE THE RESULTS?

If the results are normal, your child is not suffering from one of the diseases tested. The results will not be returned directly to you, but will be available from the maternity or paediatric ward where the sample was taken.
If any of the tests show an abnormal result, you will be promptly informed. A second blood sample will be needed. This control, which is carried out as soon as possible, will allow us to know whether your child is really affected by the disease.

Appropriate care exists for each disease that we screen for. These measures can quickly be implemented, allowing the child's development to proceed in the best possible way.


DISEASES THAT WE SCREEN FOR

PHENYLKETONURIA

Phenylketonuria is a disease resulting from a fault in the way that phenylalanine in transformed into tyrosine. Phenylalanine is a component of our daily diet. Specific analysis can detect an abnormal increase in phenylalanine in the baby's blood, which prevents its brain from developing normally. Without treatment, children develop severe and irreversible neurological disorders, including mental disability.

A low-phenylalanine diet that begins as soon as possible and continues throughout the person's life allows affected children to develop and grow normally.

Screening for this disease has been systematically carried out in the Grand Duchy of Luxembourg since 1968.

FREQUENCY:
1/13,500 newborns

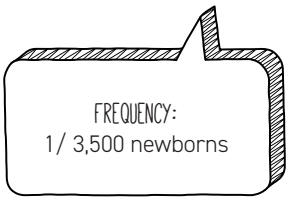
CONGENITAL HYPOTHYROIDISM

Children with this disease do not produce enough thyroxine – a hormone produced by the thyroid that is essential for the proper development of the baby's brain.

In cases of thyroxine deficiency, the pituitary gland produces an excess of TSH (thyreostimuline), which is measured during screening.

Treatment consists of the daily oral administration of thyroxine, which allows the child to develop normally.

Screening has been in place since 1978 in the Grand Duchy of Luxembourg.

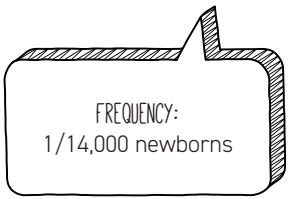


CONGENITAL ADRENAL HYPERPLASIA

Congenital adrenal hyperplasia is a genetic disorder that causes the abnormal production of hormones secreted by the adrenal glands. This disease can lead to severe dehydration and problems with genital development. Screening is based on the concentration of the hormone 17-OH Progesterone.

Treatment involves the daily oral administration of the missing hormones. This continues throughout the person's life, and allows for good growth and a normal development.

Screening for this disease has been in place in the Grand Duchy of Luxembourg since 2001.



MCAD DEFICIENCY

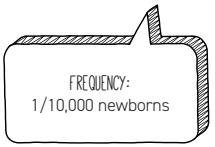
(Medium-chain acyl-CoA dehydrogenase)

In children with MCAD deficiency, fat metabolism is disrupted. This means that fat cannot be used as a source of energy. At times when energy requirements are increased (fasts, infections, fever, diarrhoea, surgery, etc.), children can have metabolic crises involving severe hypoglycaemia.

Detection involves examining the levels of acylcarnitine that accumulate in the blood of the sick baby.

The disease is primarily managed by avoiding periods of fasting and ensuring adequate glucose intake at times of increased energy requirements. These measures must continue throughout the person's life.

Screening for this disease has been in place in the Grand Duchy of Luxembourg since 2008.



CYSTIC FIBROSIS

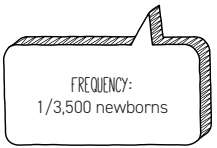
Cystic fibrosis is a genetic disease that causes excessive viscosity of secretions that affect the function of various organs, including the lungs and the pancreas.

The disease is responsible for nutritional disorders, and progressive lung disease is a serious associated complication. Clinical diagnosis is often made late, which is harmful for the patient.

A trypsin test is used to identify newborns suspected of carrying the disease. Its interpretation may require the study of the cystic fibrosis gene using molecular biology techniques.

Early management reduces the frequency of clinical symptoms and ensures a better quality of life.

This screening process was initiated in the Grand Duchy of Luxembourg in 2018



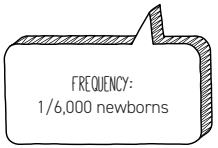
SPINAL MUSCULAR ATROPHY (SMA)

SMA is a rare genetic neuromuscular disease characterised by weakness and progressive reduction in muscle volume caused by the degeneration and loss of motor neurons, the nerve cells that receive movement information and relay it to the muscles.

The result is progressive paralysis, particularly of the lower limbs, including the respiratory muscles.

Screening is based on the search for an anomaly in the SMN1 gene, the absence of which is the cause of the disease.
Early treatment of infants (in the pre-symptomatic phase) can limit or even prevent the development of the disease.

This screening was introduced in the Grand Duchy of Luxembourg in 2024.



DO YOU STILL HAVE QUESTIONS?

This booklet can answer some of your questions. However, it does not take the place of information provided by a paediatrician, GP, or any of the other health professionals who care for your baby.

Screening can be essential for your baby's health and future. While it is not obligatory to carry out these tests, it is strongly recommended that you do so.

Screening is free, and fully funded by the Ministry of Health.

DATA PROTECTION

Your personal data and that of your child will be processed in order to carry out the neonatal screening tests and to inform you in the event of a positive result. The data relating to these tests is kept by the Laboratoire Nationale de Santé for a period of 10 years, in compliance with medical confidentiality.

To find out more about the management of your personal data and to exercise your rights, please refer to the LNS data protection policy at the following address: https://lns.lu/donnees-personnelles/.

In order to ensure the follow-up of screening tests, the data collected as part of these tests may only be communicated to healthcare professionals, in particular the maternity doctor.

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et de la Sécurité sociale

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