
Groupe
National du
Screening
Néonatal
Luxembourg
Newborn Screening in the Grand Duchy of Luxembourg
On the third day of life!

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Dear future mum, Dear future dad,

In the first days after your baby's birth, your physician or midwife will inform you about the national newborn screening programme. This programme was introduced in 1968 and concerns all newborns.

Newborn screening enables the identification of children affected by certain serious diseases, often of genetic origin.

It is important to diagnose these diseases as early as possible, since effective treatments exist. Treatments must begin in the first days of life in order to prevent certain serious consequences for your child's health, severe deficiencies and even early death.

In the Grand Duchy of Luxembourg, the four following disorders are screened:
- Phenylketonuria,
- Congenital hypothyroidism,
- Congenital adrenal hyperplasia,
- Medium Chain Acyl-CoA Dehydrogenase Deficiency - MCADD.

Since 1989, more than 100,000 babies have been tested. Forty-two affected children have been identified and treatment started within a fortnight.

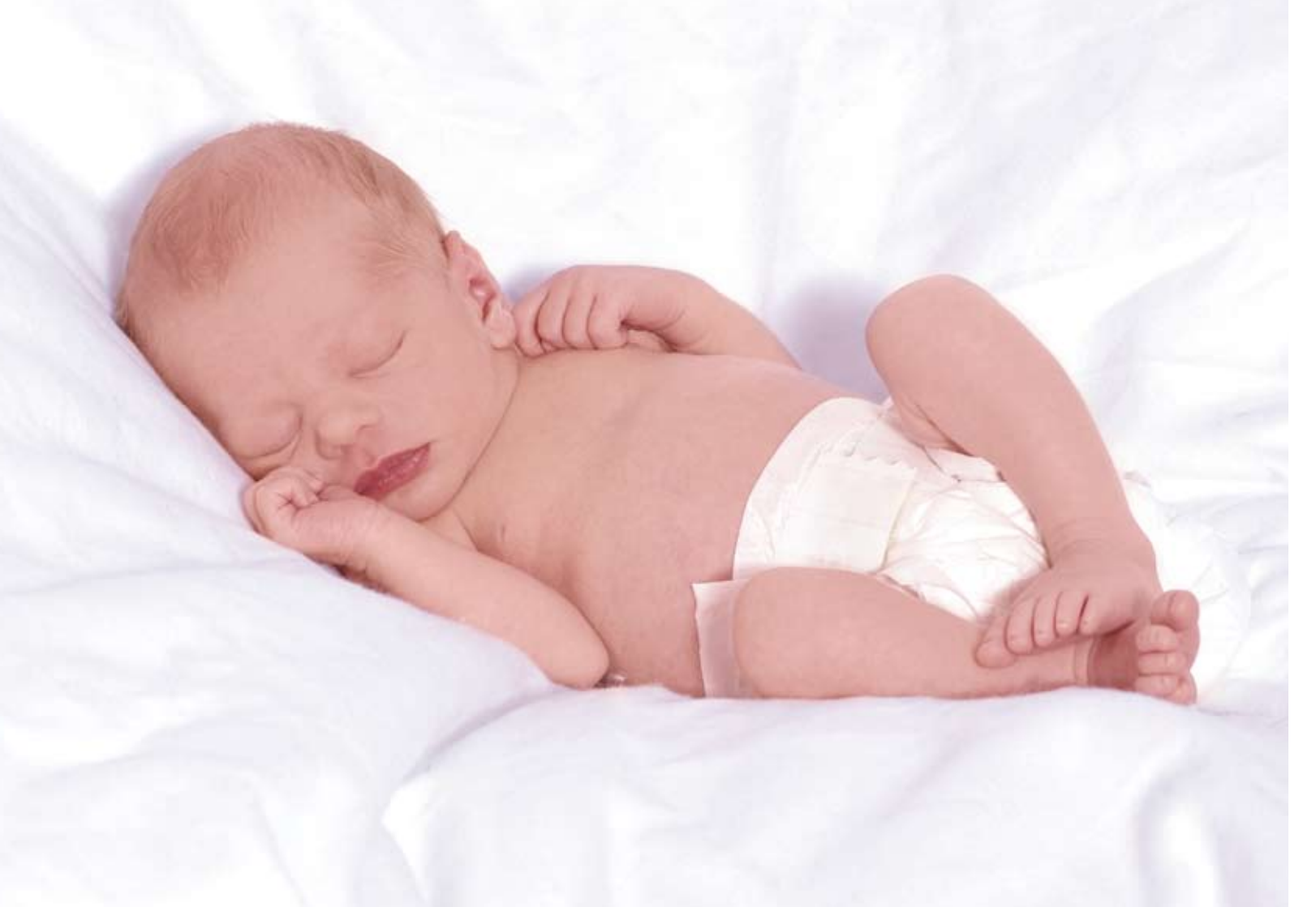
This leaflet aims to inform you about the newborn screening programme in the Grand Duchy of Luxembourg, the diseases detected, their treatment and the services you can contact, should you have any further questions.

Newborn screening is of great benefit to your baby's health.

Mars Di Bartolomeo
Ministre de la Santé

The National Group for Newborn Screening of the Grand Duchy of Luxembourg is responsible for implementing and supervising the efficient organization of the newborn screening program.

This Group is composed of representatives of:
- the National Laboratory of Health,
- the Ministry of Health
- the Luxembourg Paediatric Society,
- specialist paediatricians
- the Luxembourg Midwives Association
- the Luxembourg task force for rare diseases.



Why screen certain disorders early?

The newborn screening tests allow certain diseases that have serious health consequences if not treated rapidly to be detected in babies.

It is for this reason that the newborn screening takes place in the first days of your child's life.

Why test all newborns?

At birth, affected children appear to be in good health. A simple blood spot test enables these diseases to be detected. Therefore, it is important to test all babies. These tests present no danger to your child's health.

The disorders currently screened are rare; meaning that only one child in several thousand is affected. The probability that your baby is affected is therefore extremely low.

When do the tests take place?

Since it is very important to diagnose the diseases as soon as possible, the tests will be carried out at the 3rd day of life.

How is it done?

Your baby's heel will be pricked using a special device to collect some drops of blood onto a card.

This blood spot test is transferred to the National Laboratory of Health. They analyse the blood samples using specific methods for detecting each of the different diseases.

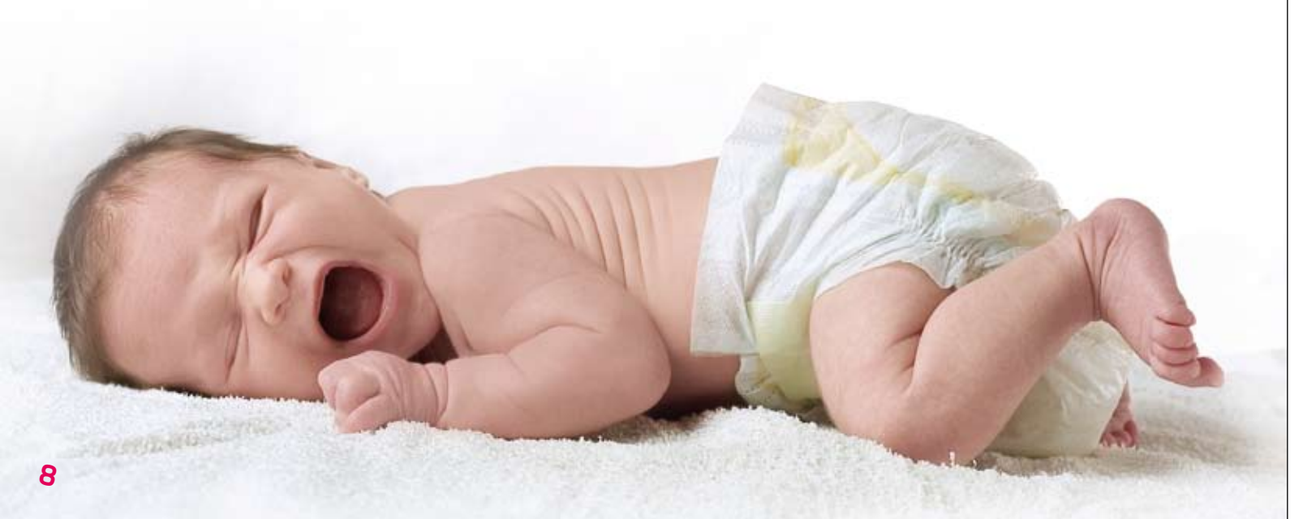
How will I hear about the results?

If results are normal, your child is not affected by one of these disorders. The results will not be directly addressed to you, but will be available in the maternity or in the paediatric unit where the blood spot test has been performed.

If one of the tests shows an abnormal result, you will be informed directly. A second blood spot test will be necessary. This verification, carried out as soon as possible, will confirm whether your child is really affected by the disease.

Since effective treatment exists for each of the screened disorders, it can be initiated quickly. Appropriate and early treatment enables normal development of the child.

Diseases detected by the newborn screening programme of the Grand Duchy of Luxembourg



Phenylketonuria

Phenylketonuria is caused by a defect in the transformation of phenylalanine into tyrosine. Phenylalanine is one of the food components that we eat every day.

A specific analysis makes it possible to detect in the blood of the affected baby an increase of phenylalanine that affects brain development if the patient is not adequately treated. **Without treatment, affected children develop serious and irreversible neurological conditions, such as mental disabilities.**

By following a lifelong diet low in foods naturally rich in phenylalanine (such as milk, meat, fish, eggs, etc.) started within the first weeks of life, affected children can develop and grow normally.

Systematic screening for this disorder was introduced in the Grand Duchy of Luxembourg in 1968.

Frequency:
≃ 1/13.500 newborns

Congenital hypothyroidism

Babies with congenital hypothyroidism produce an insufficient amount of the hormone thyroxine, normally produced by the thyroid gland. This hormone is essential for growth and the normal development of the child's brain.

If thyroxine is insufficiently secreted, the pituitary gland produces an excess of TSH (Thyroid Stimulating Hormone). The TSH level is measured to detect this disorder.

The lifelong treatment consists of the daily intake of thyroxine.

The test for detecting this disorder exists in the Grand Duchy of Luxembourg since 1978.

Frequency ≃ 1/3.500 newborns

Congenital adrenal hyperplasia

Congenital adrenal hyperplasia is a genetic disorder resulting in the abnormal production of hormones secreted by the adrenal cortex.

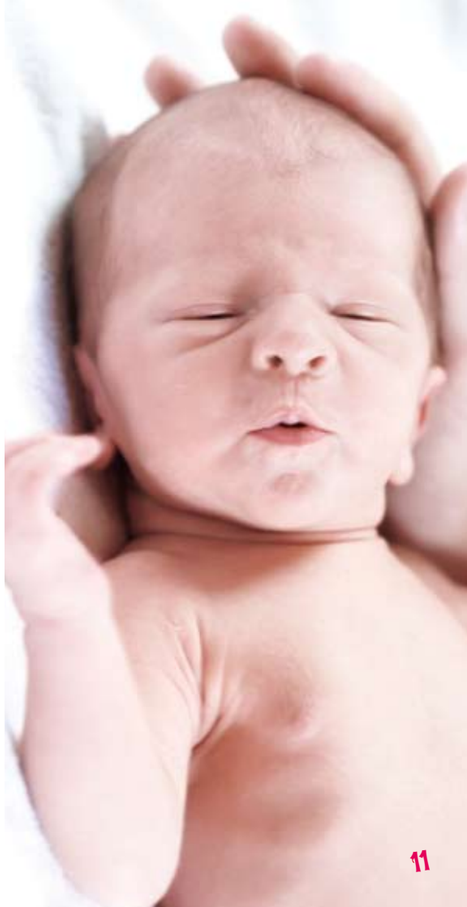
This disorder can cause severe dehydration, growth problems and the masculinisation of girls.

The newborn screening test focuses on the concentration of 17-OH progesterone.

Testing for this disorder was introduced in the Grand Duchy of Luxembourg in 2001.

Treatment consists in the daily administration of the lacking hormones and must be continued throughout life.

Frequency ≃ 1/14.000 newborns



MCADD
Medium Chain Acyl CoA Dehydrogenase Deficiency

People with Medium Chain Acyl-CoA Dehydrogenase Deficiency suffer from a metabolic disorder that prevents fatty acids being broken down and used as an energy source.

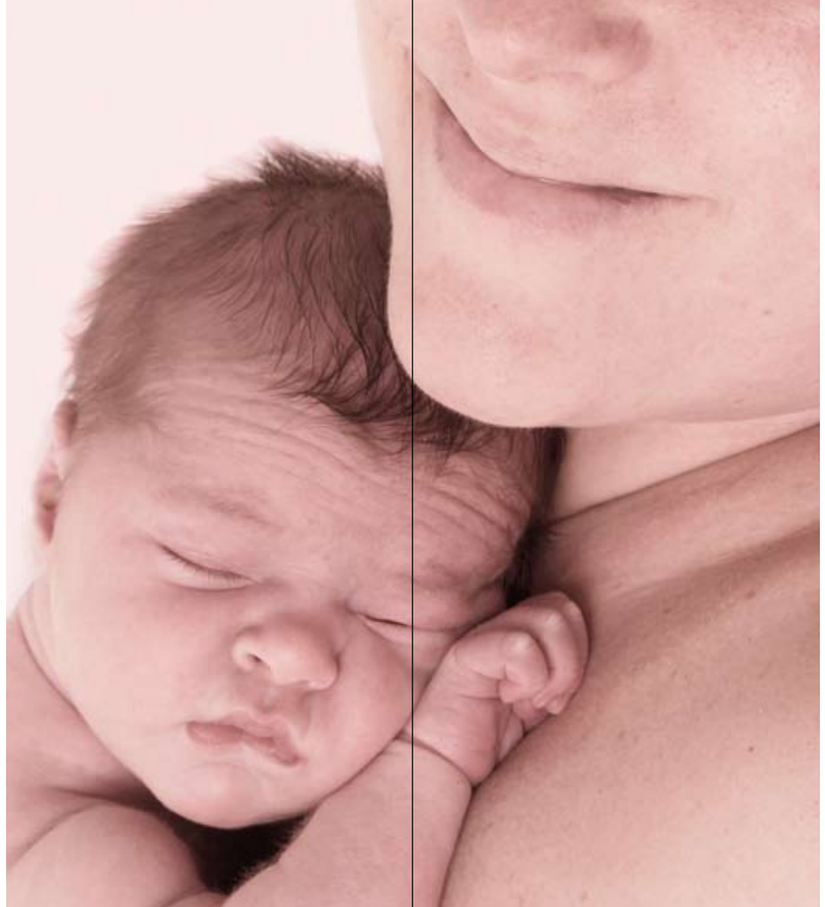
In times of fasting, infections, febrile illnesses, diarrhoea and vomiting, affected individuals may experience serious metabolic crises.

The technique used to detect this condition is the measurement of the level of acylcarnitines.

Screening for this disorder was introduced in the Grand Duchy of Luxembourg in 2008.

Treatment consists essentially in avoiding periods of fasting and in ensuring sufficient intake of glucose during illnesses such as infections, fever, vomiting, diarrhoea, etc. These measures must be maintained throughout life.

Frequency
≃ 1/10.000 newborns



If you have further questions

This leaflet aims to answer certain questions about the newborn screening programme and the disorders it targets.

Nevertheless, if you require further information, your paediatrician, physician or other health professionals caring for you and your child will be able to provide it.

Useful addresses

Groupe National du Screening Néonatal
Ministère de la Santé
Villa Louvigny, Allée Marconi
L-2120 Luxembourg

Laboratoire National de Santé
Service de Dépistage Néonatal
42, rue du Laboratoire
L-1911 Luxembourg

Parental consent

Dear Parents,

The newborn screening programme concerns all newborns in the Grand Duchy of Luxembourg.

Screening your baby for all these conditions is strongly recommended.

If you do not wish your baby to benefit from the screening programme, after having read this leaflet, please discuss your decision with your gynaecologist or your paediatrician.

Should you refuse screening for your baby, you will be required to sign a «non-consent» form, to exclude your child from the programme.

If you think that your baby might not have been screened, please discuss your concern with your paediatrician.

Groupe National du Screening Néonatal Luxembourg

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