The Institute of Mother and Child

Department of Screening and Metabolic Diagnostics, Warsaw

http://przesiew.imid.med.pl

NEWBORN SCREENING FOR CONGENITAL DISEASES

Newborn screening for congenital diseases is an important preventive test aimed at early detection of inborn conditions. These diseases do not produce visible symptoms at the neonatal age but can have serious consequences in terms of delayed physical development and/or permanent mental retardation.

Only early identification of the affected child and prompt initiation of treatment can prevent irreversible complications and improve the child's quality of life.

In Poland, screening tests are performed free of charge for all newborns as part of the state health policy programme commissioned by the Minister of Health. Currently, 30 congenital diseases are covered by the screening programme. Annually, around 400 children are born with such diseases in Poland.

The tests are carried out exclusively in specialised diagnostic laboratories (at screening centres) and the procedure is coordinated by the Institute of Mother and Child in Warsaw.

Screening tests pose no risk to the child.

? When is the test performed?

Between 48 and 72 hours after birth of each newborn in the neonatal unit a few drops of blood are taken from the heel onto a special blotting paper. Once the blood has dried, the blotting paper is sent to the screening centre.

? How to check if blood was collected for the screening test? - IMPORTANT!

Before leaving the neonatal unit, check whether in the Baby Health Booklet there is a label showing the test identification number and blood collection date.

Badanie Przesiewowe Noworodka	AB1234
Pobrano dnia:	

? Should the blood draw be repeated and when?

The lab may send you a letter with blotting paper and a code, asking to repeat the blood draw, in case:

- ✓ the result is inconclusive and the test needs to be repeated;
- ✓ your child had a blood transfusion before the first blood draw;
- ✓ the volume of blood collected was insufficient;
- ✓ your baby's birth weight was very low birth.

? Who will receive the results of the screening test?

In the majority of children, test results are normal, meaning that there is no suspicion of any of the diagnosed diseases. Information on the normal result is not sent to parents.

- NO NEWS IS GOOD NEWS -

In case the test was performed from a repeated blood draw, information on the result is sent to the parents or legal guardians at the address entered on the blotting paper.

In case the baby is suspected of having a congenital disease the parents or legal guardians are notified (by letter or phone, if a phone number has been provided on the blotting paper) that their baby should be brought to a specialist clinic or hospital ward. At the same time, the doctor at the specialist clinic or hospital ward is notified of the screening test result.

? Does the test ensure detection of all sick newborns?

Screening tests cannot guarantee 100% disease detection. However, it is very rare that a sick newborn is not detected in screening tests.

? Does molecular testing require my consent?

Newborn screening is based on biochemical and genetic tests: spinal muscular atrophy (SMA), cystic fibrosis (CF). However, as most of the diseases diagnosed by these tests are genetically determined, molecular tests are needed for definite confirmation or subtype differentiation, e.g. phenylketonuria (PKU).

According to the law, such a test in a newborn requires parent's or legal guardian's consent. The consent is given by signing the reverse of the blood collection blotting paper (specimen overleaf). ***In the absence of a signature – which indicates a lack of consent - genetic testing is not performed***.

? What diseases are detected?

All newborns in Poland are screened for: phenylketonuria (PKU), congenital hypothyroidism (CH), cystic fibrosis (CF), congenital adrenal hyperplasia (CAH), biotinidase deficiency (BIOT), spinal muscular atrophy (SMA) and 25 other rare inborn errors of metabolism (IEMs).

ⓘ Cystic fibrosis (CF)

Cystic fibrosis causes chronic lung disease and impaired food absorption. Most children with cystic

fibrosis have poor weight gain and are at risk of frequent lung and bronchial infections posing serious health risks.

Appropriate treatment significantly increases the chances of affected child's survival by alleviating the course of the disease.

Phenylketonuria (PKU)

Phenylketonuria causes a build-up of phenylalanine in the child's blood, leading to central nervous system damage, including disruption of normal brain development. Lack of early treatment often leads to severe, irreversible mental retardation. Treatment consists of a diet low in phenylalanine that must be started in the first weeks of life and continued over the following years.

Congenital hypothyroidism (CH)

Congenital hypothyroidism is a syndrome resulting from a deficiency of thyroid hormones. The absence of this hormone in a child causes developmental abnormalities and can lead to severe, permanent physical and mental impairment. Screening makes it possible to start early treatment with thyroxine, which prevents permanent changes and ensures normal development of the child.

Rare inborn errors of metabolism (IEMs)

The test detects 25 rare inborn defects of protein or fat metabolism. As a result of an enzyme block, substances accumulate in the blood of the affected child and are not further metabolised. This often leads to poisoning-like symptoms, vomiting, flaccidity, apnoea, dysfunction of various organs, up to coma and death. Treatment is complex and consists mainly of a special diet specific to the inborn error of metabolism.

Congenital adrenal hyperplasia (CAH)

Congenital adrenal hyperplasia is a disease caused by a deficiency of one of the enzymes responsible for production of adrenal hormones: cortisol and aldosterone. Deficiency of these hormones can result in life-threatening dehydration (salt loss syndrome). In addition, in girls, the disease causes abnormal development of the external genital organs already in the foetal period, which can make it difficult to determine the sex of the child at birth. Early initiation of appropriate hormonal treatment ensures the normal development of the child.

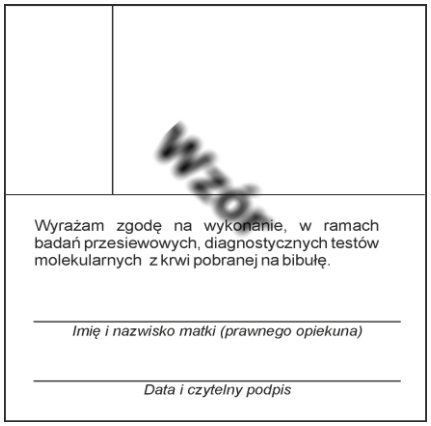
Biotinidase deficiency (BIOT)

Biotinidase deficiency symptoms include convulsions, difficulty breathing, hypotonia, skin rash, alopecia, hearing loss and developmental delay. They usually appear in the first few months of life but may also occur at a later stage. Detection of the disease at screening allows treatment through the supply of free biotin and ensures normal development.

Spinal muscular atrophy (SMA)

SMA is a severe congenital disease that manifests as progressive muscle weakening and atrophy that can cause respiratory failure. Half of children develop symptoms of the disease in the first six months of life, and the majority before the age of two. The SMA treatment is most effective when the diagnosis is made in the newborn and therapy is introduced before the onset of clinical symptoms. Newborn screening for SMA is based on genetic testing ***performed ONLY after signing the consent at the back of the blotting paper***. In case a mutation is found and thus spinal muscular atrophy is confirmed, treatment is implemented.

Drawing of blotting paper (overleaf)



Personal data processing

INFORMATION CLAUSE

Pursuant to Article 13(1) and (2) of Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 (hereinafter: GDPR), we would like to inform you that the Administrator of your personal data (your child's data) is the Institute of Mother and Child, Kasprzaka 17A, 01-211 Warsaw, phone: +48 22 32 77 000. In all matters related to the processing of your personal data, you can contact the Data Protection Officer (DPO), email: iod@imid.med.pl. Personal data is necessary for the performance of a task carried out in the public interest and for the fulfilment of a legal obligation incumbent on the administrator and contained in medical records.

Detailed information on personal data processing can be found on the Institute's website **www.imid.med.pl** under: About us – GDPR.

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Ministerstwo Zdrowia

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