Screened Diseases

Within the Czech Newborn Screening Programme there are at present 13 diseases being screened for. For most of these diseases an efficient treatment is available – for the remaining diseases early treatment substantially increases the quality of life of the patients.

- **Congenital Hypothyroidism (CH)**: insufficient production of the hormone of the thyroid gland
- **Congenital Adrenal Hyperplasia (CAH)**: insufficient production of the hormones of the adrenal glands
- **Cystic Fibrosis (CF)**: inherited disorder of the production of mucus
- **Inherited Disorders of the Amino Acids Metabolism**: Phenylketonuria; Maple Syrup Urine Disease (MSUD; leucinosis); Glutaric Acidemia type I (GA I); Isovaleric Acidemia (IVA)
- **Inherited Disorders of the Fatty Acids Metabolism**: MCAD deficiency; LCHAD deficiency; VLCHAD deficiency; CPT I deficiency; CPT II deficiency; CACT deficiency

Probability of detecting an affected newborn

The chance that your child will have some of these diseases is very small. Approximately only one child in 1100 newborns suffers from one of those 13 screened diseases (e.g. in the year 2010 there were 107 children diagnosed from 117 163 newborn babies that were screened).

The aim of screening procedure is to find all patients with an increased risk of being affected by one of the diseases. The side effect of the screening procedures may however lead to generating false positive results. In such cases the levels of screened compounds are only temporarily elevated and the original suspicion is ruled out by further testing. The chance of these false positive results varies for individual diseases.

Subsequent care

Treatment

Treatment for each of the screened disease is provided by specialized clinics, treatment varies for individual diseases. It may include an administration of special medications, special diets, administration of hormones which the body is unable to produce, or inhalation and physiotherapy.

Genetic testing and counselling

Most of the screened diseases are genetic in origine, the follow up of the patient may include genetic counselling and genetic testing of close relatives of the child.

For more information please visit the website
www.novorozeneckyscreening.cz

Screening Results

Negative result

If no suspicion for any of the screened diseases arose the screening laboratories do not issue the negative report.

Positive result – increased risk of a disease

If there is any suspicion that your child might suffer from one of the screened diseases, subsequent procedures depend on the type of the disease and the level of suspicion. In such cases either the screening laboratory or child's primary care paediatrician contact the parents, usually within one week from the blood-collection (in case of cystic fibrosis within approx. six weeks). Therefore it is essential to provide correct contact details of the parents or the primary care paediatrician, or both. Ideally these details should include both the full address and the telephone number(s).

Positive result of the screening test however does not mean that your child suffers from the screened conditions. Positive results indicate only increased risk of the baby to be affected by one of the diseases. In most cases it is necessary only to repeat the screening tests from another heel prick. In some cases further laboratory testing is needed (usually after collecting venus blood or/and urine). In rare circumstances there might be need for urgent admission to hospital.

Newborn Screening Programme is a preventive programme intended for detecting newborns with an increased risk for several rare diseases. These diseases if detected early can be treated to prevent serious harm. The screening programme is based on the recommendations of the Ministry of Health of the Czech Republic and it is fully covered by the health insurance.

The newborn screening tests are performed in specialized laboratories using dry blood spots. Within 48 – 72 hours after birth several drops of blood are taken from the heel of the baby using a special screening card.

The screening test is carried out after the informed consent (oral/written) of the parents of the baby has been obtained. The parents should obtain the necessary information from the paediatrician in the maternity ward. For more detailed information the parents can also consult the www.novorozeneckyscreening.cz website. In case of delivery out of a hospital the blood should be taken by the child's primary care paediatrician.

The aim of screening procedure is to find all patients with an increased risk of being affected by one of the diseases. The side effect of the screening procedures may however lead to generating false positive results. In such cases the levels of screened compounds are only temporarily elevated and the original suspicion is ruled out by further testing. The chance of these false positive results varies for individual diseases.

Subsequent care

Treatment

Treatment for each of the screened disease is provided by specialized clinics, treatment varies for individual diseases. It may include an administration of special medications, special diets, administration of hormones which the body is unable to produce, or inhalation and physiotherapy.

Genetic testing and counselling

Most of the screened diseases are genetic in origine, the follow up of the patient may include genetic counselling and genetic testing of close relatives of the child.

For more information please visit the website
www.novorozeneckyscreening.cz

newborn screening

Newborn Screening Programme is a preventive programme intended for detecting newborns with an increased risk for several rare diseases. These diseases if detected early can be treated to prevent serious harm. The screening programme is based on the recommendations of the Ministry of Health of the Czech Republic and it is fully covered by the health insurance.

The newborn screening tests are performed in specialized laboratories using dry blood spots. Within 48 – 72 hours after birth several drops of blood are taken from the heel of the baby using a special screening card.

The screening test is carried out after the informed consent (oral/written) of the parents of the baby has been obtained. The parents should obtain the necessary information from the paediatrician in the maternity ward. For more detailed information the parents can also consult the www.novorozeneckyscreening.cz website. In case of delivery out of a hospital the blood should be taken by the child's primary care paediatrician.

Screened Diseases

Within the Czech Newborn Screening Programme there are at present 13 diseases being screened for. For most of these diseases an efficient treatment is available – for the remaining diseases early treatment substantially increases the quality of life of the patients.

- **Congenital Hypothyroidism (CH)**: insufficient production of the hormone of the thyroid gland
- **Congenital Adrenal Hyperplasia (CAH)**: insufficient production of the hormones of the adrenal glands
- **Cystic Fibrosis (CF)**: inherited disorder of the production of mucus
- **Inherited Disorders of the Amino Acids Metabolism**: Phenylketonuria; Maple Syrup Urine Disease (MSUD; leucinosis); Glutaric Acidemia type I (GA I); Isovaleric Acidemia (IVA)
- **Inherited Disorders of the Fatty Acids Metabolism**: MCAD deficiency; LCHAD deficiency; VLCHAD deficiency; CPT I deficiency; CPT II deficiency; CACT deficiency

Probability of detecting an affected newborn

The chance that your child will have some of these diseases is very small. Approximately only one child in 1100 newborns suffers from one of those 13 screened diseases (e.g. in the year 2010 there were 107 children diagnosed from 117 163 newborn babies that were screened).

Screening Results

Negative result

If no suspicion for any of the screened diseases arose the screening laboratories do not issue the negative report.

Positive result – increased risk of a disease

If there is any suspicion that your child might suffer from one of the screened diseases, subsequent procedures depend on the type of the disease and the level of suspicion. In such cases either the screening laboratory or child's primary care paediatrician contact the parents, usually within one week from the blood-collection (in case of cystic fibrosis within approx. six weeks). Therefore it is essential to provide correct contact details of the parents or the primary care paediatrician, or both. Ideally these details should include both the full address and the telephone number(s).

Positive result of the screening test however does not mean that your child suffers from the screened conditions. Positive results indicate only increased risk of the baby to be affected by one of the diseases. In most cases it is necessary only to repeat the screening tests from another heel prick. In some cases further laboratory testing is needed (usually after collecting venus blood or/and urine). In rare circumstances there might be need for urgent admission to hospital.