Newborn Screening
Neugeborenen-Screening
Dépistage Néonatal
Screening Neonatale

Prevention: A Simple Method with Lasting Results

What are the tests for?
Metabolic or hormone disorders, which are diagnosed by newborn screening, usually cause serious damage to various organs if left untreated. In particular, brain development can be adversely affected. In the period following birth, these illnesses are without clinical symptoms, meaning that if the newborn has not been tested, typical indicators are often only detected during the first few years of life. Therefore, beginning treatment immediately after birth is extremely important to prevent lasting damage.

State of the art methods used in newborn screening make it possible to identify metabolic and hormonal disorders shortly after birth simply by taking a few drops of blood from the baby's heel on the third or fourth day of life. The blood is transferred to a strip of filter paper and sent to our laboratory where it is tested for the diseases listed below.

Historical Overview

As in most European countries, all newborns in Switzerland, at present circa 80,000 per year, are checked for certain congenital metabolic and hormone diseases. Thanks to the initiative of paediatricians and the enthusiastic cooperation of obstetricians, midwives and nurses in neonatal hospital units since the 1960's, the approach used throughout Switzerland has maintained the current high standard of excellence. The minimal costs for these routine tests are covered by all health insurances.

The Disorders Covered by the Tests

1. Phenylketonuria (PKU)

If left untreated, this metabolic disease usually leads to serious brain damage in the growing child, which will require constant care throughout life. However, if the disorder is identified in the first weeks of life and treated with an appropriate diet, the child will be healthy. PKU prevents the body from processing the amino acid phenylalanine, a normal component of all animal and plant proteins usually absorbed by the body during digestion. Lack of degradation leads to the accumulation of metabolic substances that are poisonous to the brain. A special diet containing low levels of phenylalanine prevents the disease from advancing. Incidence is about one in 8000 newborns, with varying severity.

2. Hypothyroidism

Hypothyroidism, a congenital subfunction of the thyroid gland, slows all metabolic functions due to insufficient thyroid hormones, leading to serious impairment of physical and mental development. If thyroid hormone is administered orally soon after birth on a daily basis, normal metabolic function is established and thus healthy childhood development will ensue.
Hypothyroidism is quite common, affecting one in 3500 newborns.

3. Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)

MCADD is a congenital fatty acid oxidation disorder. This disease becomes apparent when the body has to access fat reserves during prolonged periods of fasting, and causes vomiting, diarrhoea and fever, for example, as a result of viral infections experienced frequently throughout childhood. The effects are drowsiness, nausea, seizures and coma. If undiagnosed, the first onset frequently leads to death and is reminiscent of "sudden infant death".
If appropriate measures are taken – avoidance of long periods of fasting and providing a sufficient intake of carbohydrates, especially during infections and fever - the prognosis is excellent. MCADD affects one in 10,000 newborns.

4. Galactosaemia

Newborns suffering from galactosaemia cannot process lactose. A milk-based diet leads to severe liver, kidney and brain damage, impaired vision, and, in some cases, even death. Yet if the illness is discovered within the first few days of life and treated by means of an appropriate diet, the child will remain healthy.
Treatment consists of a strict avoidance of galactose, a component of lactose, which is present in human breast milk, cow's milk and other milk-based foods.
The disorder affects one in 55,000 newborns. There are two other forms of galactosaemia apart from the classic variety, but these are rarer and less serious.

5. Congenital Adrenal Hyperplasia (CAH)

In cases of CAH, the adrenal glands produce insufficient cortisol an salt-retaining hormones. This leads to dangerous crises involving low blood sugar and blood pressure levels and the loss of salts during infections with fevers. There also is an excessive production of male hormones that causes masculine development of the outer genitalia in affected girls. Boys' genitals develop normally, but puberty sets in early, leading to stunted growth and infertility.

Therapy is simple: The lacking hormones are taken orally. If treatment begins shortly after birth, no damage is caused. The disorder affects one in 9000 newborns.

6. Biotinidase Deficiency

This disorder causes various types of damage, which may be extremely severe, and can even result in the infant's death. The enzyme biotinidase normally extracts biotin from food during digestion, making it available to the organism in its free form. In cases of biotinidase deficiency, the vitamin is lost. However, biotin is vital for various metabolic enzymes. Here too, early diagnosis and immediate therapy can prevent damage. Treatment of biotinidase deficiency is simple, involving the daily intake of one biotin tablet. The disorder is roughly as common as galactosaemia.

7. Cystic Fibrosis (CF)

CF is caused by a malfunction of cellular salt processing leading to mucus build-up in the airways and pancreas. This causes chronic airway infection, stunted growth, and serious illness if it remains untreated. Milder variants are also known, which are sometimes not diagnosed until adulthood.

With the use of intensive inhalations, chest physiotherapy, and an appropriate diet involving the supplementation of digestive enzymes and fat-soluble vitamins, unnecessary hospitalisations can be avoided and a normal development is possible.
CF is the most common congenital metabolic disease, which affects one in 2500 newborns.

8. Glutaric acidemia type 1 (GA1)

In this metabolic disorder, the amino acids, lysine and tryptophan, normal components of all animal and plant proteins, cannot be processed normally in the body. As a consequence, metabolic products can accumulate that are toxic for the brain. Children with the condition tend not to have any symptoms as a newborn. If left untreated, most children with the condition will have a larger head than normal and over time will suffer from developmental delay as well as certain movement disorders.Between the ages of 3 months and 3 years, acute metabolic crises occur, often triggered by minor infections, which can lead to permanent movement difficulties and disability. GA1 can be well-managed with a special diet and by taking l-carnitine supplements. To prevent metabolic crises, minor infections during the first years of life should be always be treated as an emergency.

9. Maple syrup urine disease (MSUD)

Maple syrup urine disease (also known as MSUD) is a hereditary metabolic disease, in which particular protein components (amino acids leucine, isoleucine and valine) cannot be properly processed in the body. As a consequence, toxic substances accumulate,which can lead a rapid decline in newborns. Particularly serious is the occurrence of brain swelling, which can cause a coma. If left untreated, this disease can lead to death. There are also milder forms, which can affect sufferers less significantly.

With a strict diet MSUD can be well-managed. Patients who are diagnosed as part of newborn screening have a good prognosis.

10. Severe Combined Immunodeficiency (SCID) and severe T-cell lymphopenia

In Severe Combined Immunodeficiency (SCID) and severe T-cell lymphopenia, there is a disorder in the formation of T-cells. As a result, life-threatening infections and a pronounced growth disorder can quickly develop. If left untreated, SCID and severe T-cell lymphopenia can lead to death. There are also mild forms known that are less severely affected. With a stem cell transplant SCID and severe T-cell lymphopenia can be cured. Patients discovered in neonatal screening have a good prognosis.

How the Screening is Done

The dried blood samples preserved on filter paper are sent to the Swiss screening laboratory in Zurich. The results are available within a few days. If they are normal, which is the case for the vast majority of children, the parents are not informed and can be assured that their child is free of all of the listed diseases.

If, on the other hand, the results are positive, parents will be informed immediately via the birth unit, the responsible paediatrician or the nearest paediatric clinic, so further tests can be made. A positive result at this stage need not mean that the child is afflicted by one of these illnesses. In many cases, a second test reveals that everything is in order. Only if the diagnosis can be confirmed by intensive analyses, usually taking no more than a few days, treatment can begin immediately. Further steps concerning lifelong treatment are then planned together with the responsible paediatrician or the nearest paediatric clinic and specialists at the relevant centre for metabolic and hormone disorders.

All test results as well as the remaining materials from the blood test are kept at our screening laboratory. If illnesses occur later, which can be clarified via tests on this sample, the responsible physician can request it from us. Part of the remaining material may also be used by the screening laboratory during quality control measures and for the development of new research methods. In this case, the samples will be anonymous, having undergone a procedure to make them unidentifiable.

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