Newborn Screening
Neugeborenen-Screening
Dépistage Néonatal
Screening Neonatale

Prevention: A Simple Method with Lasting Results

What are these tests for?
If left untreated, the congenital disorders that are diagnosed by newborn screening can cause serious damage to various organs, adversely affecting brain development in particular, or lead to serious infections. 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 months or years of life and treatment is initiated late. However, in order to prevent permanent damage, it is of utmost importance to start treatment for these disorders in the first days of life. State-of-the-art methods used in newborn screening make it possible to detect the covered congenital 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 we test it for the diseases listed below.

Historical Overview

As in most European countries, all newborns in Switzerland, at present around 87,000 per year, are checked for certain congenital diseases. It is thanks to the initiative of paediatricians and the enthusiastic cooperation of obstetricians, midwives and nurses in neonatal hospital wards since the 1960s, that this Switzerland-wide programme has achieved the high standard of qualitative excellence seen today. The low costs of these screenings are covered by all health insurers as basic benefits.

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, who will then 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.

In cases of PKU, the amino acid phenylalanine, a normal component of all animal and plant proteins, usually absorbed during digestion, cannot be processed properly in the body. This leads to the production 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 underactive thyroid, slows all metabolic functions due to insufficient thyroid hormones, leading to serious impairment of physical and mental development. However, if the lacking hormone is administered in tablet form soon after birth on a daily basis, normal metabolic function is established and healthy childhood development is thus guaranteed.

Hypothyroidism is quite common, affecting one in 3500 newborns.

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

MCADD is a congenital disorder affecting the breakdown of fatty acids in adipose tissue. This disease first becomes apparent when the body has to access fat reserves during prolonged periods of fasting, vomiting, diarrhoea or fever – for instance as a result of the minor infections experienced frequently throughout childhood. The effects are drowsiness, nausea, low blood sugar, seizures and coma. If undiagnosed, the first onset frequently leads to death.

However, if appropriate preventative measures are taken (avoiding long periods of fasting and providing a sufficient intake of carbohydrates, especially during infections with fever) the prognosis is excellent.

MCADD affects one in 10,000 newborns.

4. Galactosaemia

Newborns suffering from galactosaemia cannot tolerate lactose. A milk-based diet then leads to severe liver, kidney and brain damage, impaired vision and, in some cases, even death. Yet if the disorder is identified in the first days of life and treated with an appropriate diet, the child will remain healthy.

It is treated by strictly avoiding galactose, a component of lactose, which is present in human breast milk, cow's milk and other foods.

The disorder affects one in 55,000 newborns. There are two other forms of galactosaemia besides the classic variety, but these are rarer and less serious.

5. Congenital Adrenal Hyperplasia (CAH)

In cases of CAH, the adrenal cortex produces insufficient cortisol. This leads to dangerous crises involving low blood sugar and the loss of salts during infections with fever. There also is an excessive production of male hormones that causes masculine development of the outer genitalia in affected girls. Among affected boys, genitals develop normally, but puberty sets in early, leading to stunted growth and infertility. Therapy is simple: The lacking hormones are administered as tablets. If such treatment begins shortly after birth, CAH causes no damage. 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 releases the vitamin biotin from its bound form, making it available to the body in its free form. In cases of this illness though, the vitamin is lost. However, biotin is vital for the functioning of various metabolic enzymes.

Here too, early diagnosis and immediate therapy can prevent damage. Treatment of biotinidase deficiency is simple, consisting of the daily intake of one biotin tablet. The disorder is roughly as common as galactosaemia.

7. Cystic Fibrosis (CF)

CF involves a cellular salt-exchange malfunction, leading to excessively viscous secretions in the airways and pancreas. This causes chronic airway inflammation, stunted growth and serious illness if it remains untreated. Mild variants are also known, which are sometimes not diagnosed until adulthood. With intensive inhalation, chest physiotherapy, and an appropriate diet involving the supplementation of digestive enzymes and fat-soluble vitamins, unnecessary hospitalisation can be avoided and better development is possible. CF is the most common congenital metabolic disease and it affects around one in 3000 newborns.

8. Glutaric Acidemia Type 1 (GA1)

In cases of this metabolic disorder, the amino acids lysine and tryptophan, normal components of all animal and plant proteins, cannot be processed properly in the body. This leads to the production of metabolic substances that are toxic for

the brain. Affected children tend not to have any symptoms as newborns. 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 mild 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 disorders and severe disability.

GA1 can be managed well with a special diet and by taking l-carnitine supplements. To prevent metabolic crises, even minor infections should be treated as emergencies during the first years of life, meaning inpatient emergency treatment as a precautionary measure.

9. Maple Syrup Urine Disease (MSUD)

Maple syrup urine disease (also known as MSUD) is a congenital metabolic disorder, in which particular protein components (the so-called amino acids leucine, isoleucine and valine) cannot be properly processed in the body. As a consequence, toxic substances accumulate, which can lead to a very rapid decline in newborns. The occurrence of brain swelling is particularly serious and can cause a coma. If left untreated, this disease can lead to death. There are also mild forms known, which affect patients less significantly. MSUD can be managed well with a special diet. Patients who are diagnosed during newborn screening have a good prognosis.

10. Severe Combined Immunodeficiency (SCID) and Severe T-Cell Lymphopenia

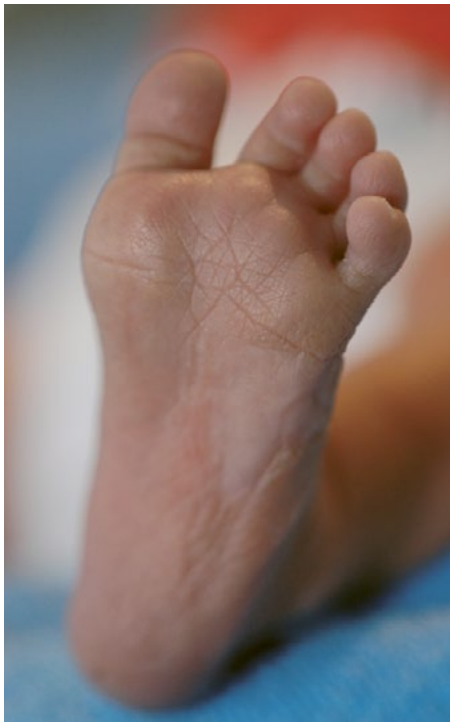
Cases of severe combined immunodeficiency (SCID) and severe T-cell lymphopenia involve a disturbance in the formation of T-cells. As a result, life-threatening infections and a pronounced growth disorder can quickly develop. If left untreated, SCID and severe T-cell lymphopenia can lead to death. There are also mild forms known, which affect patients less significantly. With a stem cell transplant, SCID and severe T-cell lymphopenia can be cured. Patients who are diagnosed during newborn screening have a good prognosis.

11. Spinal muscular atrophy (SMA)

SMA is a rare congenital disease. People living with SMA experience a loss of nerve cells that are necessary for movement and muscle strength in the whole body. This leads to weakness in the muscles of the arms and legs. Breathing and swallowing can be severely affected during the course of the disease. Symptoms often become apparent in the first year of life. Newborn screening can identify almost all newborns (approx. 95%) at risk of SMA. If the test is positive, the parents are invited to a neuromuscular center for an urgent examination of the newborn and additional tests. Diagnosis after newborn screening allows for early monitoring and treatment. Effective therapies are now available that can halt or slow the progression of SMA. If treatment begins before the first symptoms appear, the baby has the best chance of normal motor development.

How the Screening is Done

The dried blood samples on filter paper are sent to the Newborn Screening Switzerland laboratory at University Children's Hospital Zurich. The results are available within a few days. If these are normal, which is the case for the vast majority of children, the parents are not informed and can rest assured that their child is free of all the aforementioned diseases.



If, on the other hand, the results are positive, the parents are contacted either immediately or via the maternity clinic, paediatrician or nearest paediatric clinic, so that further tests can be arranged. However, a positive result at this stage need not mean that the child is afflicted by any of these illnesses. In many cases, a second test reveals that everything is actually in order. Once diagnosis is confirmed by intensive analyses, usually taking no more than a few days, treatment of the child is started without delay. The further course of treatment, which is sometimes necessary for the rest of the child's life, is then planned together with the paediatrician or nearest paediatric clinic and specialists at the relevant centre for metabolic, hormonal, immunological or lung disorders.

Storage of Test Results and Blood Samples

All test results are kept at the screening laboratory for at least 30 years. For quality assurance purposes, the remaining material from the blood sample is kept for 10 years. If, in the event of a newly occurring disease, questions arise later that could be answered by testing this sample (e.g. testing for a congenital cytomegalovirus infection using PCR), the treating physician can, during this period, request that we get the sample tested accordingly, with your consent.

Some of the remaining material, in a non-identifiable anonymised form, can also be used by the screening laboratory for checking the quality of tests and for the development of new testing methods.

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