Test

Newborn Mass Screening Test
All babies are eligible for this test and the cost is subsidized by the government.
(Partially paid by the parent/guardian)

Additional Screening Test

Primary immunodeficiency diseases
&
Lysosomal diseases

The test is conducted for babies upon request.
(The total cost of the test is borne by the parent/guardian.)

Since this test is performed in conjunction with the Newborn Mass Screening test, there is no additional burden on your baby, such as drawing another blood sample.

Test procedure

Blood sampling
Blood is collected on filter paper from the heel at 4-6 days of age.

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Test
The results will be available within a week to 10 days after the blood is drawn.

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Notification of test results

In case of no abnormality

2-3 weeks after the blood is drawn,
the results will be mailed to the medical institution where you gave birth.
The result sheet will be given to you at your baby's 1-month checkup.

In case that an abnormality is suspected

You will be contacted immediately
by phone by your maternity medical institution.

Retest

If the first test cannot reliably determine that there is no abnormality, another blood sample will be drawn and tested to confirm the result with certainty.

Detailed examination

Your maternity medical institution will call you and refer you to another medical institution that can perform a detailed examination to see if your baby is carrying a particular disease.

Even if any of the diseases are detected, there is a system in place where the medical institution that performs a detailed examination, a medical consultant (physician), and the testing organization cooperate with each other to ensure that appropriate treatment is promptly provided.

The most important thing is early detection with this test to provide proper treatment.

Contact Information
Hokkaido Pharmaceutical Association Public Health Examination Center
Phone: 011-824-1348 Fax: 011-824-1627

Q & A

What is the purpose of this test?

Even babies who appear healthy may have been born with a disease. Early detection and treatment can minimize the onset of disease and developmental problems.

Does my baby have to take the test?

Only one in tens to hundreds of thousands of babies are diagnosed with these diseases. However, because of the difficulty in diagnosis and treatment, we recommend that all babies undergo this test.

Do I have to pay for the test?

Since the test is optional, you will have to pay for it. Please contact your maternity institution.

Does the need for "retest" or "detailed examination" mean my baby is sick?

No. The need for retest or detailed examination does not necessarily mean that the baby is sick. In some cases, due to the condition of the baby, it may not be possible to make an accurate diagnosis and the test may be repeated. A detailed examination will be done at a specialized hospital to determine if the baby definitely carries the disease.

How will I pay for the treatment of my baby's illness?

Primary immunodeficiency diseases and lysosomal diseases are designated as specific diseases (intractable diseases) and specific pediatric chronic diseases by the Japanese government, and are therefore covered by the medical subsidy programs of the national and local governments.

Hokkaido Early Diagnosis Network for Rare Diseases

This test is conducted in cooperation with the following organizations

Diagnosis and Treatment Organization
Department of Pediatrics, Hokkaido University Graduate School of Medicine
National Hospital Organization Hokkaido Medical Center
Department of Pediatrics, Asahikawa Medical University

Testing organization
Hokkaido Pharmaceutical Association Public Health Examination Center

If you would like to know more about this test, please visit the following website.
https://www.douyakken.or.jp/HEDNet-RD/

If you would like to take the test, please contact our hospital.

2021.4

Primary immunodeficiency diseases Lysosomal diseases

Information on Additional Screening

Along with Newborn Mass Screening, please have your baby tested!

Do everything possible to protect your precious child.

Additional Screening Test

Newborn Mass Screening Test

Primary immunodeficiency diseases
&
Lysosomal diseases

The sooner the disease is identified with this test, the sooner the baby can be treated.

There is no additional burden on the baby such as another blood sampling.

Hokkaido Early Diagnosis Network for Rare Diseases

Primary immunodeficiency diseases (PID)

What kind of diseases?

These are diseases that cause repeated infections and become severe, due to an innate abnormality in the immune system that fights infections. The two most common types are **severe combined immunodeficiency (SCID),** in which there are no T lymphocytes, and **B cell deficiency,** in which there are no B lymphocytes. Both of these diseases cause repeated severe infections from early infancy, resulting in pneumonia, otitis media, meningitis, and sepsis.
In the US, Taiwan, and some parts of Japan, this test is already available, and many babies have been diagnosed early and saved.

So this test is important.

Supervisor: Masafumi Yamada, Associate Professor, Department of Pediatrics, Hokkaido University Graduate School of Medicine

Severe combined immunodeficiency (SCID)

Severe combined immunodeficiency is a disease that carries a high risk of death if it is not treated properly. Even if a baby appears healthy, the disease can be detected early by testing. **It is important to diagnose and start treatment before an infection occurs.**

Major symptoms

- **Persistent diarrhea and coughing**
- **Inability to gain weight or loss of weight**
- **Severe viral infection**
- **Oral candida infection**

If left untreated and the child is later inoculated with a live vaccine such as a rotavirus vaccine, a BCG vaccine, etc., the child could die.

Treatment

Hematopoietic stem cell transplantation can restore the immune system and provide a complete cure. Bone marrow or umbilical cord blood (blood from the umbilical cord) is transplanted.

B cell deficiency (X-linked agammaglobulinemia, etc.)

The disease occurs mainly in boys and develops at the age of around 3 to 4 months when the immunoglobulin received in the womb from the mother starts to decrease. Early diagnosis and treatment can prevent the infection from becoming severe and recurring.

Major symptoms

- **Repeated otitis media and sinusitis**
- **Pneumonia, meningitis, and sepsis**
- **Bronchiectasis**

Treatment

Immunoglobulin preparations are given throughout life to compensate for the lack of antibodies and help prevent infections.

Get your baby tested for primary immunodeficiency and lysosomal diseases to ensure his/her healthy growth!

Lysosomal diseases

What kind of diseases?

The enzymes contained in the lysosomes in the cells are either absent or malfunctioning from birth, resulting in the accumulation of fat and carbohydrates and the appearance of various symptoms. There are about 60 known types of lysosomal diseases, and this test can detect five of them: mucopolysaccharidosis type I and II, Pompe disease, Fabry disease, and Gaucher disease. **Detecting the diseases as early as possible after birth and providing enzyme replacement therapy, etc. can control the progression of symptoms.**

So this test is important.

Supervisor: Masayoshi Nagao, Vice Director, National Hospital Organization Hokkaido Medical Center

Treatment

The main treatment for lysosomal diseases is enzyme replacement therapy. By replenishing the body with the missing enzymes through intravenous infusion, the accumulated substances are broken down and the progression of symptoms can be suppressed. In some cases, treatment is given immediately, while in other cases, the timing of the start of treatment is based on stages of the child's growth and development.

Mucopolysaccharidosis type I
Mucopolysaccharidosis type II

Symptoms may not be seen immediately after birth, but gradually become more apparent, and symptoms may be noticed, and a diagnosis is made around one or two years of age.

Major symptoms

- **Short stature**
- **Delayed development**
- **Herniated intestine/Protruding navel**
- **Repeated otitis media**
- **Swelling of the liver and spleen**
- **Joint pain and difficulty moving joints**
- **Heart diseases**
- **Corneal clouding**

Pompe disease

The time of onset differs depending on the type of the disease, ranging from symptoms appearing soon after birth, to those noticed in adulthood.

Major symptoms

- **Loss of muscle strength**
- **Breathing disorders**
- **Heart failure**

Fabry disease

Once believed to be a higher risk for men, but now recognized as affecting both sexes. In girls, the disease may occur from puberty to adulthood, even if the Newborn Mass Screening test had shown no abnormality.

Major symptoms

Children

- **Pain in the hands and feet**
- **Reduced ability to sweat**
- **Heat intolerance**
- **Fever**
- **Abdominal pain**
- **Diarrhea**
- **Skin rash**

Adult

- **Renal disorder**
- **Cardiac disorder**
- **Cerebrovascular disorder**

Gaucher disease

There are three types of Gaucher disease, which differ in the presence or absence of neurological symptoms such as convulsions, the time of onset, and the progression of the disease.

Major symptoms

- **Abdominal distension**
- **Swelling of the liver and spleen**
- **Blood abnormalities (anemia, thrombocytopenia)**
- **Bone pain, deformity, and fracture**
- **Strabismus, difficulty opening the mouth**
- **Convulsions**