Test

Newborn mass screening + ADA deficiency, ALD, Lysosomal disease test

*This test is carried out using public funds, covering all babies (costs partially borne by the parent/guardian)

Regarding filter paper blood for the newborn mass screening, only a minuscule amount of blood is drawn.

*This test is carried out using public funds, covering all babies (costs partially borne by the patient/guardian)

Results

In case of no abnormality

We will send your results of the drawn blood in 2–3 weeks weeks via the healthcare provider.

In case of abnormality

You will promptly be informed via the healthcare provider that drew the blood. A detailed test is needed.

"Suspected" does not necessarily mean that the baby has a disease.
In some cases, drawing blood a second time may be requested for confirmation.

Treatment

Severe Combined Immunodeficiencies (SCID)

Currently the main method of treatment is the hematopoietic stem cell transplantation that uses HLA (white blood cell type) matched bone marrow (from a family member or bone marrow bank) or umbilical cord blood from a cord blood bank.
Unless the transplantation is carried out promptly, the life of the baby with a severe combined immunodeficiency cannot be saved. Furthermore, after a serious infection has occurred due to a delayed diagnosis, the number of babies who cannot be saved will increase even if a hematopoietic stem cell transplantation is performed.
If a baby with a severe combined immunodeficiency is detected early as a result of the mass screening before any infection is contracted, a better treatment outcome can be expected. Other treatment methods such as gene therapy are currently being studied.

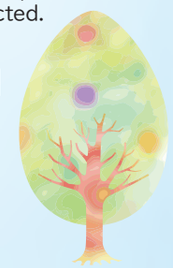
Lysosomal Diseases

For lysosomal diseases, the enzyme replacement therapy that supplements insufficient enzymes is available.
By supplying an enzyme preparation through an intravenous drip, accumulated abnormal substances may be dissolved to control disease progression. In addition, depending on conditions, the same effect may be achieved by oral medication. It is necessary to start treatment immediately depending on the disease and severity. However, in some cases, the time to start treatment may be determined while considering progress in the child's growth and development stages.

Adrenoleukodystrophy (ALD)

As soon as possible after the outbreak, hematopoietic stem cell transplantation should be conducted if any symptoms are experienced to suppress the progress of the symptoms. Once symptoms have appeared, good results cannot be expected, so periodic MRI scans should be performed. It is essential to consider hematopoietic stem cell transplantation the moment abnormalities are detected.

Performing early detection and appropriate treatment is most important.



Q&A

What is the purpose of this test?

A The purpose of this test is to detect serious diseases at an early stage.
Even babies who look healthy may have been born with a disease. By early detection and treatment, possible growth disorders can be minimized.

Is the test mandatory?

A The test is not mandatory.
However, since these are very rare diseases which are difficult to detect and treat, we recommend that all babies undergo screening.

Do I have to pay for the test?

A Since the test is optional, a cost is incurred.
Regarding the details of actual cost, please contact your healthcare provider.

What happens if my baby is suspected of having these diseases?

A You will be contacted through the healthcare provider which drew the baby's blood sample.
Then, you will be introduced and referred to a healthcare provider which can offer a detailed test and treatment. In the event that a disease is detected, preparations will be made in cooperation with the healthcare provider which can carry out a detailed test, an expert medical consultant (physician), and a support organization associated with the healthcare provider, in order to promptly offer specialized treatment to protect your precious baby. Personal information will be strictly protected and managed.

If you would like to receive this test, please contact our hospital.

ARDnet
Aichi Rare Disease Network

Notification of Additional Screening for Rare Diseases

In addition to general mass screening for newborns, additional screening tests for lysosomal disease, severe combined immunodeficiencies due to adenosine deaminase deficiency and Adrenoleukodystrophy are available.

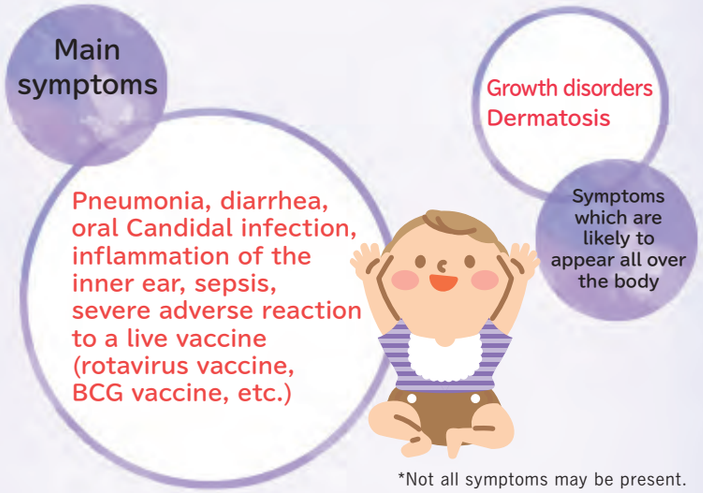
These diseases are difficult to diagnose, and if they are not identified until after symptoms appear, treatment efficacy is sometimes insufficient. If detected early, however, these diseases can be treated with various types of options. For the sound growth of your child, please consider receiving additional screening.

General Supervisor: Tetsuya Ito
(Professor, Fujita Health University Hospital, Department of Pediatrics)
Aichi Rare Disease Network

2024.2

Severe combined immunodeficiency (SCID) due to adenosine deaminase (ADA) deficiency

Severe immunodeficiency is a disease caused by innate immune abnormalities. The cells involved with immunity within the blood cell contain very few T lymphocytes. This prevents B lymphocytes from producing antibodies to pathogens, which makes it impossible to defend the body from the pathogens, leading to repeated infections.



*Not all symptoms may be present.

Specific symptoms and characteristics

- Soon after birth, most babies do not show symptoms, and they cannot be differentiated from healthy babies without a test.
- In early infancy, serious infections such as pneumonia, sepsis, and gastroenteritis occur repeatedly.
- Due to chronic diarrhea and malabsorption, weight gain may be interrupted.
- Serious pneumonia or sepsis can develop, and if diagnosis is too late and the appropriate treatment is not received, there is the possibility of death.
- Before symptoms appear and diagnosis is made, the baby also has a risk of receiving **a live vaccine (rotavirus vaccine, BCG vaccine, etc)**.
- Inoculation with such vaccines may cause serious, life-threatening adverse reactions in babies with a severe combined immunodeficiency.

If a severe combined immunodeficiency is not appropriately treated at an early stage, there is a high risk of death before the baby reaches one year of age. In cases of late detection and inoculation of a live vaccine such as BCG, the situation is extremely dangerous.
ADA deficiency is one of the disorders that cause SCID. However, the national and local governments are likely to fail to detect it when they perform SCID screening as a verification project. We perform tests for this disorder separately using a different testing method.

Early detection makes treatment possible.
Taking a test for ADA deficiency, which is a cause of SCID, is strongly recommended.

Supervisor: Hideki Muramatsu
(Lecturer, Nagoya University Hospital, Department of Pediatrics)

Lysosomal diseases

What are lysosomal disease?
Lysosomes are sac-like structures inside human cells that act to dissolve and remove substances no longer needed in the cell. Inside lysosomes, there are a lot of proteins called “enzymes,” which work to break down any substances, such as fat and carbohydrates, that are no longer needed. If the activities of enzymes inside lysosomes worsen, substances which should be dissolved will accumulate inside the cell. Diseases in which the cell malfunctions are collectively called “lysosomal diseases.” There are approximately 60 known types of lysosomal disease, and some new medical treatments have been developed. Through early detection of the disease and starting treatment as soon after birth as possible, minimizing the progress of symptoms and disease outbreak can be expected. For this reason, we carry out a test on four types of lysosomal diseases—Pompe disease, Fabry disease, and mucopolysaccharidosis type I and type II.

Pompe disease

In this disease, a substance called “glycogen” accumulates in the lysosome. As muscle strength decreases, various disorders appear. In the most severe cases, called the “infantile-onset type,” symptoms such as enlargement of the heart or respiratory disorder appear a few months after birth, which requires an artificial respirator, and life may be lost due to heart failure. In milder cases called the “late-onset type,” disease symptoms such as decreased muscle strength are observed after early childhood. The infantile-onset type is rarer than the late-onset type, but early screening, detection, and treatment soon after birth have been found to prevent or delay the progress of the disease.

Fabry disease

This disease causes abnormal substances to accumulate in blood vessels throughout the body, leading to symptoms such as pain in the hands and feet, kidney, heart, cerebral blood vessels, and eyes. Since the disease is a sex chromosome genetic disorder, symptoms mainly appear in male infants, but can be observed in women after adulthood. Symptoms appear after 5–6 years years of age, so even if identified by mass screening, treatment will not start immediately. When to begin the treatment will be decided after closely observing the symptom conditions. For girls, even if the test results from the mass screening are normal, symptoms may appear after puberty or adulthood. Consequently, only boys are targeted in the mass screening test.

Mucopolysaccharidosis (MPS) type I and type II

This disease occurs when a substance called “mucopolysaccharide” accumulates throughout the body.
This is a progressive disease with symptoms such as the inflammation of the inner ear, joint contracture, peculiar facial features, cataracts (not observed in type II), respiratory disorders, heart valve disease, and growth/ developmental disorders. There are no differences between the sexes for type I, but type II is a sex chromosome genetic disorder that almost exclusively affects male infants. By early detection and enzyme replacement therapy or hematopoietic stem cell transplant, prevention or delay of disease progression can be expected.

Supervisor: Tetsuya Ito
(Fujita Health University Hospital, Department of Pediatrics)

Adrenoleukodystrophy (ALD)

A type of peroxisomal disorder that occurs mostly in male infants. There are several known types of the disease, but the severe pediatric cerebral type causes degeneration of nerve cells and nerve fibers in the brain and other organs, and abnormalities in the adrenal glands, which produce essential hormones. This results in a rapid deterioration of intelligence, behavioral abnormalities, and deterioration of motor skills and vision. Without treatment, the patient will become bedridden within one to two years of the appearance of symptoms, but early treatment such as bone marrow transplantation can prevent or reduce the symptoms.
Since the disease progresses if the symptoms are not diagnosed until after the onset of the disease, the new newborn mass screening test is used to diagnose the disease in advance, and periodical MRI scans are performed to check for the disease’s onset. However, this does not guarantee that abnormalities will appear even if the disease is diagnosed. Therefore, only boys are targeted in the mass screening test.

Cautions

Regardless of the disease, due to the characteristics of the test, even healthy persons who do not need medical treatment or persons with very minor symptoms may be identified to have potential abnormalities. Likewise, even in babies with no abnormalities found in this test, the possibility of the presence of the diseases is not completely ruled out.
On the other hand, for severe cases, even if treatment is started early, the disease may continue to progress.

To protect our precious children, expanded newborn screening is strongly recommended.

Babies cannot explain with words no matter how sick they may feel. What the parents can do for their baby is to detect any diseases as early as possible, before it gets too late.