Test

Newborn mass screening + SCID, ALD, SMA Lysosomal diseases 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 for babies on an optional basis. (costs borne by the parent/guardian)

Results

In case of no abnormality

We will send your results of the drawn blood in 2-3 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 Disorders

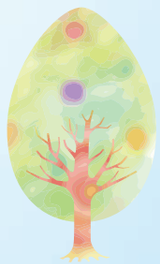
For lysosomal disorders, the enzyme replacement therapy that supplements insufficient enzymes is available.
By supplying an enzyme preparation through an intravenous drip, accumulated abnormal substances may be dissolved to control disease progression. In addition, depending on conditions, the same effect 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 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.

Spinal muscular atrophy (SMA)

New medicines, such as nucleic acid drugs and gene therapy that supplement gene function, have been developed. Decreased muscle strength and breathing difficulties can be alleviated by early diagnosis and treatment.



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 Expanded Screening for Rare Diseases

In addition to general mass screening for newborns, additional screening tests for lysosomal diseases, severe combined immunodeficiencies (SCID), Adrenoleukodystrophy (ALD) and Spinal muscular atrophy (SMA) 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.

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2023.4