Q&A

Why is the test needed?

Even if babies are born healthy, they may have inherited diseases. Early detection and treatment can minimize the development of disease and developmental disturbance.

This test is optional. A diagnosis is difficult to make for diseases examined by an optional screening test even if it occurs. A delay in the detection of disease prevents appropriate treatment in a timely manner, possibly leading to a serious residual impairment. So please consider taking the optional screening test.

Do I have to pay for the test?

The optional screening test is available for a fee.
Please ask your medical institution about the fee.

How about costs for the treatment of disease?

All diseases to be examined by the optional screening test are designated as specified intractable diseases or specific pediatric chronic diseases, which means that the national or local medical expenses subsidy system is applicable.

What should I do if my baby is suspected of having a disease?

Even if the results of the optional screening test suspect an abnormality, it does not always mean that your baby has a disease. You should understand that a screening test is not intended to be a “diagnosis” but is a test to check the probability of suspected diseases.

If your baby is suggested to have a disease, a specialist with an abundance of experience with the diagnosis and treatment of the disease, the medical institution to which you applied for the test, and testing laboratories will collaborate with each other and introduce you to an institution where a specialist is available and your baby can undergo a detailed examination.

If you wish to have the test, please ask us.

Testing laboratories

ACRL by AnGes

AnGes Clinical Research Laboratory (ACRL)

ACRL is a clinical laboratory managed by AnGes, Inc.
Address: Life Science & Environment Research Center
3-25-13 Tonomachi, Kawasaki-ku, Kawasaki, Kanagawa
TEL: 044-223-6484
HP: https://www.acrl.jp

★ If you wish you have the test, please ask a medical institution.
★ The optional screening is different from newborn mass screening covered by public funds. For questions about the optional screening, please contact ACRL.

Optional Screening

Information about the test

Detect hidden disease in your sweet baby early and treat it appropriately

AnGes, Inc.
Version 1.0 (01-Apr-2025)

What is the optional screening test?

It is a test to check whether or not a baby may have diseases that are not tested in the “newborn mass screening” that is performed in all babies by public funds.

Diseases to be examined by the optional screening test

- **Lysosomal storage diseases**
  Mucopolysaccharidosis I, II, IVA, VI, and VII, Pompe’s disease, Fabry disease (boys only), Gaucher disease, Niemann-Pick disease type A or B, and Krabbe disease
- **Adrenoleukodystrophy (boys only)**
- **Spinal muscular atrophy**
- **Severe combined immunodeficiency**
- **Adenosine deaminase deficiency**

※ Spinal muscular atrophy and severe combined immunodeficiency may not be included in the optional screening depending on medical institutions, and they may be tested using public funds.

Test Method

In principle, the same method as that for an inherited metabolism disorder test (test of diseases included in newborn mass screening) will be used so that a baby should feel almost no stress.

①	**Blood collection**	Blood will be drawn in filter paper from the baby’s heel 4 to 6 days after birth.
②	**Test**	The results will be available in 1 week to 10 days after blood collection.
③	**Results**	When normal You will receive the results at the time of the 1-month health checkup through the medical institution where your baby was born. When any abnormality is suspected You will be immediately contacted. The medical institution where your baby was born will call you to inform you of the results and introduce you to a medical institution that is able to make a diagnosis and
④	**Results**	If any abnormality is suspected in Item ③, whether or not it is a disease will be examined in detail at the institution that will make a diagnosis and give treatment.

Lysosomal storage diseases (LSDs)

What kind of disease is it?

"Lysosome" is an organ in a cell that controls cell metabolism and decomposes unnecessary metabolites. Because an "enzyme" in lysosomes does not work well in patients with LSDs, lipids and glucose are accumulated in cells, resulting in various symptoms.
Approximately 60 types of LSDs are known. This test will examine 10 types: Mucopolysaccharidosis I, II, IVA, VI, and VII, Pompe's disease, Fabry disease, Gaucher disease, Niemann-Pick disease type A or B, and Krabbe disease.
Finding the disease as early as possible after birth and starting treatment is expected to prevent symptoms from progressing.

Treatment Methods

The main treatment method is "enzyme replacement therapy." It may be initiated early or started as a follow-up of the course. In addition, taking a drug to increase enzyme activity or hematopoietic stem cell transplantation may be effective for some types of LSD.

*For Gaucher disease and Niemann-Pick disease type A, the currently available therapies cannot prevent the progression of neurological disorders such as intellectual impairment.
*For Krabbe disease, enzyme replacement therapy is not available at present. Tests will periodically be carried out, and hematopoietic stem cell transplantation soon after onset is considered effective.

Mucopolysaccharidosis I, II, IVA, VI, and VII

Symptoms gradually appear shortly after birth. Bone joint deformities and contractures become prominent at the age of 1 to 3 years.

Main symptoms

- Decreased visual acuity
- Intellectual impairment
- Abnormal gait
- Pigmentation
- Decreased hearing
- Cardiac valvulopathy

Pompe's disease

An infantile-onset type occurring in infants with severe symptoms and a late-onset type occurring after infancy with mild symptoms are known.

Main symptoms

- Muscular weakness
- Breathing disorder
- Heart failure
- Motor developmental delay

Fabry disease (boys only)

The onset period and severity of symptoms vary among individuals. Because it mainly occurs during childhood for males, this test will be applicable to boys only.

Main symptoms

- Pain in hands and feet
- Intolerance to hot temperatures
- Difficulty sweating
- Abdominal pain/Diarrhea
- [Adulthood] Renal disorder
- [Adulthood] Cardiac disorder
- [Adulthood] Cerebrovascular disorder

Gaucher disease

The age of onset varies widely from infants to adults. Many Japanese people are diagnosed with it at the age of 5 years or younger.

Main symptoms

- Enlarged liver and/or spleen
- Bone pain
- Bleeds easily
- Developmental delay
- Anemia
- Convulsion
- Strabismus

Niemann-Pick disease type A or B

Symptoms appear in a wide range of age groups from childhood to adulthood.

Main symptoms

- Muscle weakness
- Developmental delay (type A)
- Convulsion
- Tightness of hands and legs
- Enlarged liver and/or spleen
- Anemia
- Bleeds easily

Krabbe disease

Symptoms appear in a wide range of age groups from early infancy to adulthood.

Main symptoms

- Irritability
- Poor suckling
- Developmental delay
- Paralysis
- Blindness
- Hearing loss

Adrenoleukodystrophy (ALD)

(Boys only)

What kind of disease is it?

It is a disease that causes abnormalities in the adrenal gland, brain, and spinal cord. ALD is a progressive intractable disease with an unpredictable onset period. It is an X-linked genetic disease and is severe in male patients so this test will be performed only in boys.

Treatment Methods

Tests will periodically be carried out. As soon as an abnormality is found, treatment to prevent development or progression will be given, such as adrenal corticosteroids and hematopoietic stem cell transplantation.

Main symptoms

- Decreased visual acuity
- Decreased academic performance
- Abnormal gait
- Pigmentation
- Hearing loss
- Character change
- Convulsion

Spinal muscular atrophy

What kind of disease is it?

Cells in the spinal cord that work to move muscles (motoneurons) are altered, resulting in impairment of muscles such as those in the hands and legs and motor function of the whole body. This disease is caused by an abnormality of a gene called *SMN1*.

Treatment Methods

In recent years, revolutionary therapeutic drugs have been developed: nucleic acid medicine to modify the gene function, gene therapy to replace deficient gene function, small-molecule agents to increase SMN protein that is necessary for moving muscles.

Main symptoms

- Poor suckling
- Muscle weakness
- Joint contractures
- Gait disturbance
- Respiratory failure
- Difficulty in swallowing

Severe combined immunodeficiency

What kind of disease is it?

The most severe type of disease with abnormal immune function (immune deficiency disease) in which immune cells (T-lymphocytes) that are important for protecting the body cannot be produced from birth, thus causing severe infections to repeat or become worse. Administration of live vaccines such as a BCG and rotavirus vaccine may induce a critical condition.

Treatment Methods

Treatment with antimicrobial agents and immunoglobulins (antibodies collected from the blood of people with normal immune function) is helpful for the prevention of infection but does not cure this disease. Radical therapies include hematopoietic stem cell transplantation using bone-marrow blood and cord blood.

Main symptoms

- Pneumonia
- Diarrhea
- Otitis media
- Susceptibility to infections

Adenosine deaminase deficiency

What kind of disease is it?

It is an immune deficiency disease caused by the impairment of lymphocytes that are important for immune function, and it results from the accumulation of metabolites in cells due to a function deficiency of an enzyme called adenosine deaminase. Adenosine deaminase deficiency is considered to account for 15% of all severe combined immunodeficiencies.

Treatment Methods

As with other severe combined immunodeficiencies, a common radical treatment is hematopoietic stem cell transplantation. Even if hematopoietic stem cell transplantation is not available, enzyme replacement therapy to inject the enzyme adenosine deaminase may be effective.

Main symptoms

- Pneumonia
- Diarrhea
- Poor weight gain
- Susceptibility to infections