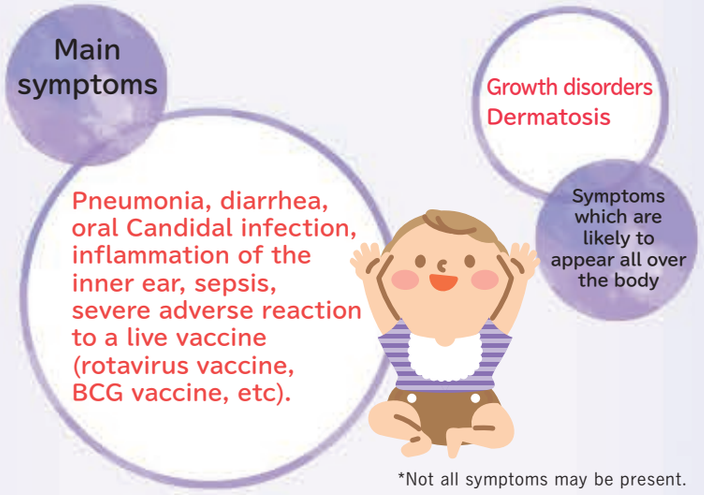
Severe combined immunodeficiency (SCID)

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.

Early detection makes treatment possible.
Taking a test for severe combined immunodeficiency is strongly recommended.

Supervisor: Hideki Muramatsu,
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Lysosomal diseases

What are lysosomal diseases?
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 diseases, 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 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.

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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.

Spinal muscular atrophy (SMA)

Spinal muscular atrophy (SMA) is a disease where the nerve cells, which control body movement and are located in the spinal cord within the backbone, degenerate. As a result, muscles atrophy and muscle strength progressively weaken. Decreased muscle strength and atrophy are observed in the torso, arms and legs. Disease type is classified according to the age at which symptoms appear and subsequent conditions as Type I, II, III, and IV. In Type I cases, symptoms begin to appear by around 6 months. The baby cannot sit up without support and has difficulty drinking milk, swallowing food, and breathing. In many cases, life-saving measures are needed, such as using an artificial respirator and providing nutrients through a tube directly to the stomach. Due to the development of new medicines, such as gene therapy, the prevention and reduction of symptoms are possible through early diagnosis and treatment.

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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, additional 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.