

Primary Immunodeficiency disease

Primary Immunodeficiency disease is the disease that causes recurrence or worsening infections due to a congenital disorder in immune system fighting infection. Representative examples include severe combined immunodeficiency and B-cell deficiencies. It is crucial to diagnose and initiate treatment before the onset of infections (live vaccination without treatment may cause a dangerous condition).

Severe Combined Immunodeficiency (SCID)

Main Symptoms:

- Persistent diarrhea and cough
- Failure to gain or lose of weight
- Aggravation of viral infections
- Oral Candida infection

B-cell Deficiency (BCD)

Main Symptoms:

- Recurrent otitis media and/or sinusitis
- Pneumonia, meningitis, sepsis
- Bronchiectasis

Treatment Method

It is crucial to diagnose the above diseases and start treatment promptly with infection preventing measures. The treatment is to restore immune function by transplantation or to supplement antibodies by immunoglobulin preparations.

Spinal Muscular Atrophy (SMA)

SMA is the disease that causes the progressive muscle weakness throughout the body and that may cause infant death without treatment. The majority of cases manifest in infancy. Its ratio is estimated as one with SMA among 20,000 babies.

Main Symptoms:

- Feeding difficulties
- Weak cry, shallow breathing
- Muscle weakness
(inability to support the head/sit up)

Treatment Method

It is possible to undergo effective treatment using nucleic acid therapeutic drugs and gene therapy. Early detection and treatment is crucial because the early treatment has a significant effect.

In the event that the illness is detected, the following institutions will collaborate to provide prompt support for receiving appropriate treatment.

Okayama Expanded Newborn Screening
Promotion Association

Okayama University Hospital
(Diagnosis and Treatment Facility)
Okayama Prefecture Association of Obstetricians
and gynecologists
Okayama Prefecture / Okayama City
Okayama Health Foundation (Secretariat)

Inquiry : Okayama Health Foundation

[Tel:086-246-6260](tel:086-246-6260)

Points to Note

Due to the nature of the test, no disorder detected does not guarantee the complete absence of illness.

Conversely, even if disorder is identified in the test, there are healthy people and mild patients who do not require any treatment. Additionally, in severe cases, the condition may get worse despite treatment from the very early stage.

Until today, treatment for the disease begins after symptoms appeared due to the difficulty of diagnosis. However, advancements in medical science now allow for the early detection of diseases through this test, and enable to take appropriate measures.

If you would like more detailed information about this examination, please watch the video here.



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