



Expanded Newborn Screening Explanation



英語

Congenital Metabolic Disorder Screening is a series of blood tests used to determine whether a newborn baby has a serious congenital disease. In Okayama Prefecture, Congenital Metabolic Disorder Screening for 25 diseases is provided through public funding, allowing for early detection and treatment.

In addition to this, Expanded Newborn Screening is also available on request for early detection of three more diseases: **Severe Combined Immunodeficiency (SCID)**, **B-cell Deficiency (BCD)**, and **Spinal Muscular Atrophy (SMA)**.

★ Screening Fee:

Since screening is optional, an additional fee is typically required, however if you agree to participate in research conducted by the national government (Children and Families Agency) by submitting the Expanded Newborn Screening Application & Consent Form, the screening will be provided at no expense.

*1 Depending on the medical facility, an additional fee may be charged for blood sample collection, etc.

*2 Only available for babies born after 1 April 2025.

*3 Data collected from the SCID and SMA tests (data that cannot be used to identify the individual, such as the number of individuals tested, number of positive results, etc.) will be provided to the national government and a government research group (Children and Families Scientific Research Tajima Group) in order to determine if Expanded Newborn Screening including these two tests should be provided to newborns around the country.

Screening fees cannot be refunded after applying for the tests.

★ Screening Application:

Those who would like the screening must fill out the screening application form (3 sheets) and submit it to the medical facility where the baby was delivered.

★ Screening Method:

A very small amount of blood is collected from the sole of the baby's foot at around 5 days of age and soaked into a filter paper for testing. The same blood sample that is currently used to screen for 25 diseases can be used, so there is no need for an additional sample.

★ Screening Results:

Results will be delivered as positive (possibility of disease), retest, or negative (low possibility of disease).

In the case of positive results, Okayama University Hospital will contact you by phone (the number will begin with 086-235-6 or 086-235-7).

In the case of negative results or a retest, you will be informed by the medical facility where the baby was delivered.

★ Regarding Targeted Diseases:

Please see the reverse side.

★ Communication of Results:

If nothing is detected during screening, you will be informed during the one-month checkup at the medical facility where the baby was delivered. If an irregularity is detected during screening, the medical facility where the baby was delivered or Okayama University Hospital will contact you immediately by phone. Please follow their instructions and promptly undergo retesting or further tests.

★ Regarding Personal Information:

Personal information obtained during the application will only be used for the screening and for providing research data to the government as consented to. Furthermore, this information will be strictly managed.

★ Additional Notices:

This screening is entirely optional. The decision to undergo screening or not is left entirely to the discretion of the individual and cannot be forced upon them.

Due to the nature of the tests, even if nothing is detected, the absolute absence of the tested diseases cannot be guaranteed. Conversely, even if an irregularity is detected, there are cases where the patient is actually in normal condition, or where their condition is minor enough not to require treatment.

Additionally, in severe cases, conditions may get worse even with treatment at an early stage.

Traditionally, due to the difficulty of diagnosis, treatment of these diseases could only begin after symptoms appeared. However, thanks to advancements in medical science, this screening can be used to detect and begin treatment of these diseases at an early stage.

Okayama Expanded Newborn Screening Promotion Association

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