Information on Additional test for primary immunodeficiency and lysosomal diseases

We advise you to have your baby tested <u>at the same time as the Newborn Mass Screening test.</u>

The Newborn Mass Screening test is conducted by local governments across Japan. Even if a baby appears to be healthy, early detection and treatment of congenital diseases can minimize the onset of diseases and developmental disorders. In Hokkaido, this test is conducted for 26 different diseases.
In addition to these diseases, the Hokkaido Early Diagnosis Network for Rare Diseases has started an additional screening test for early detection of primary immunodeficiency diseases and lysosomal diseases (5 types).
These diseases are quite rare and have been difficult to diagnose and treat, but with the advancement of medicine, it has become possible to suppress the progression of symptoms by detecting and starting treatment early in life.
Primary immunodeficiency diseases cause repeated infections and become severe due to the inability to produce T-lymphocytes and B-lymphocytes, which are responsible for immunity, due to inherent immunological abnormality. The two most common types are severe combined immunodeficiency (SCID), in which there are no T lymphocytes, and B cell deficiency, in which there are no B lymphocytes. If found early and treated with bone marrow transplantation, etc., the chances of living a healthy life are increased. SCID, in particular, can be a fatal disease without immediate appropriate treatment, if it is detected late and an unknowing carrier has a routine vaccination with a live vaccine such as a rotavirus vaccine, a BCG vaccine, etc.
Lysosomal diseases cause a variety of symptoms due to an innate abnormality in the enzymes that break down unwanted fat and carbohydrates in the cells. This test is expected to detect the five types of lysosomal diseases (mucopolysaccharidosis type I and II, Pompe disease, Fabry disease, and Gaucher disease) at an early stage, enabling enzyme replacement therapy and hematopoietic stem cell transplantation to be performed earlier, and reducing the progression of symptoms.
This test is already widely used in the U.S., Taiwan, etc., and many lives have been saved, but in Japan, it is only available in some areas.

The cost of the test
The test is optional, so you will have to pay for it. Regarding the actual cost, please contact the medical institution where you will give birth.

How to take the test and receive the results
This test uses the same blood that is drawn for the Newborn Mass Screening test, so **<u>there is no need for a new blood sample from the baby</u>**. Please apply to your maternity medical institution, by filling out the additional test application form and consent form enclosed in the test set.
The results of the test should be explained by your attending physician at the medical institution along with the results of the Newborn Mass Screening test. In case of no abnormality, the results will be mailed to the medical institution within two to three weeks after the blood is drawn. If a detailed examination is required, you will be immediately referred to another institution for the examination, through the medical institution where you gave birth, so please visit it immediately.

Note
- For any of these diseases, the results of the test may show that your baby is normal or very mildly ill and does not require treatment. If he/she is found to be severely ill, symptoms may progress even

after early treatment.

- Normal screening test results do not completely rule out all the diseases concerned. Especially in the case of Fabry's disease, girls may develop the disease from puberty to adulthood, even if test results at birth had found no abnormality. The test may also lead to the detection of disease in adult patients in the family.
- The personal information obtained at the time of application for the test will be used only for the purpose of this test. In addition, in conducting the test and follow-up surveys, the information will be strictly managed in compliance with laws and regulations and in accordance with the personal information protection policy of this center.
- When filling out the application form, parents/guardians are requested to consent to the use of their child's specimens after the completion of the test. The specimens will be stored for five years. Even once you have given your consent, you may withdraw your consent at any time of your own free will. Please sign the "Withdrawal Form" below, cut out the relevant part, and mail it to the contact address below.

Hokkaido Early Diagnosis Network for Rare Diseases

Testing organization:	Hokkaido Pharmaceutical Association Public Health Examination Center
Diagnosis and Treatment Organization:	Department of Pediatrics, Hokkaido University Graduate School of Medicine
	National Hospital Organization Hokkaido Medical Center
	Department of Pediatrics, Asahikawa Medical University

Contact Information for inquiries:

Hokkaido Pharmaceutical Association Public Health Examination Center
Address: 8-6-6, Hiragishi 1, Toyohira-ku, Sapporo, 062-0931, Japan
Phone: 011-824-1348
Fax: 011-824-1627

If you would like to know more about this test, please visit the following website.

https://www.douyakken.or.jp/HEDNet-RD/

---------- Cut here ----------

Consent Withdrawal Form

Date: _____

To Hokkaido Early Diagnosis Network for Rare Diseases

I hereby withdraw my consent to the use of specimens from the additional test, previously granted on _____(Date) for research purposes.

Name of Child: _____ Date of Birth: _____

Signature of Parent/Guardian: _____ Relationship to Child: _____

Address: _____