Attachment 1 (related to Article 6, Item 2)

Explanation of Inborn Errors of Metabolism Testing

Information on the Gifu Prefecture Inborn Errors of Metabolism Testing Program

Tests for inborn errors of metabolism are performed on infants aged 4 to 6 days old. Even though a newborn may appear healthy, it could have a congenital disease. Life-long disabilities may be prevented by detecting and treating these disorders early. Tests for inborn errors of metabolism are essential for detecting diseases before symptoms appear, allowing for effective treatment to be started immediately.

Applicable Diseases

(1)	Congenital hypothyroidism		Causes issues such as delayed intellectual development and growth.
(2)	Congenital adrenal hyperplasia		Causes issues such as dehydration and poor development.
(3)	Galactosemia		Causes issues such as delays in psychomotor development, cataracts.
(4)	Abnormal amino acid metabolism	Phenyl ketonuria	Causes issues such as delays in psychomotor development, convulsions, and pigment deficiencies.
(5)		Maple syrup urine disease	Causes issues such as loss of consciousness and convulsions.
(6)		Homocystinuria	Causes issues such as intellectual disability, psychological abnormalities, and skeletal abnormalities.
(7)		Citrullinemia type 1	Causes impairments ammonia processing when metabolizing amino acids. Leads to issues such as convulsions, vomiting, and brain damage. Treated with medication and diet.
(8)		Argininosuccinic aciduria	
(9)	Abnormal organic acid metabolism	Methylmalonic acidemia	Causes organic acids to accumulate in the body due to impaired metabolic processing of amino acids. May cause symptoms such as convulsions and vomiting and can also cause sudden death. Treated with medication and diet.
(10)		Propionic acidemia	
(11)		Isovaleric acidemia	
(12)		Methylcrotonylglycinuria	
(13)		Hydroxymethylglutaric aciduria (HMG)	
(14)		Multiple carboxylase deficiency	
(15)		Glutaric aciduria type 1	
(16)	Abnormal fatty acid metabolism	Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	Disorders impairing energy production processes. Can cause weakened muscle movement and sudden death through drops in blood sugar levels. Treated with medication and lifestyle guidance regarding energy intake.
(17)		Very-long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	
(18)		Trifunctional protein deficiency (TFP)	
(19)		Carnitine palmitoyltransferase type 1 deficiency (CPT1)	
(20)		Carnitine palmitoyltransferase type 2 deficiency (CPT2)	

Note: These tests may reveal secondary diseases (CACT deficiency, systemic carnitine deficiency, glutaric acidemia type 2) in addition to the above 20 diseases.

Things to know about this test

- The diseases tested for can cause encephalopathy and sudden infant death due to simple colds, etc., so early detection and treatment are vital.
- Mild cases that do not require treatment may be detected. Particularly mild cases of these diseases may not be detected in newborn blood.
- If the disease arises immediately after delivery, the results may not be available in time.
- Treatment may not be effective enough to prevent disability in particularly severe cases completely.
- Occasionally, mild abnormalities are detected shortly after birth, but test values eventually normalize after monitoring.

Application Form for Inborn Errors of Metabolism Testing

To the head of the medical institution and the attending physician

I have read the document explaining the inborn errors of metabolism test and have also received an explanation from the medical institution. I understand the test and how my personal information will be used and therefore:

Consent

Do not consent to my newborn infant undergoing these tests.

(Please circle one of the above)

Application Date	Year Month Day
Address	
Phone number	( ) - Note: Please make sure to enter a phone number you are contactable on.
Parent's signature	

Note: If you will be temporarily staying at another address after leaving the hospital, please fill in the contact information for this address.

Address	Care of:
Phone number	( ) -

Applying for the test

Please fill out the application form, affix your seal, and submit it to the medical institution.

Costs

If the test is conducted at a medical institution in Gifu Prefecture, the cost of the test itself will be covered by Gifu Prefecture. However, you will be responsible for costs related to blood collection and specimen delivery (if you live in Gifu Prefecture and give birth at a maternity hospital in another prefecture, you must take the test according to the system in that prefecture. For information on fees, etc., please directly contact the prefecture or ordinance-designated city with jurisdiction over the address of the medical institution where the baby will be born.)

Method

4 to 6 days after birth, a minimal amount of blood will be collected from the sole of your child's foot at the medical facility where they were born.

The blood will be sent from the medical institution to a specialized laboratory (Gifu Research Center for Public Health) for testing.

Receiving the test results

The test results will be explained to you by your doctor at your medical institution. Please also enter these results into your maternal and child health handbook. (The method and timing of the result explanation will vary from place to place, so please check with your institution).

Results: The difference between a repeat test and a confirmatory test (precise test)

What is a repeat test?

If there is uncertainty regarding the results of the first test, a repeat test will be performed. This re-test will be done at the medical institution where you gave birth.

What is a confirmatory test (precise test)

A more detailed test is performed if the initial or repeat test results suggest your child has a disease. If a confirmatory test is required, please follow the instructions from the attending physician at the institution where the test was performed and arrange to undergo a confirmatory test at a pediatric clinic. Only about one in three babies who undergo confirmatory testing are actually diagnosed with a disease. Occasionally, it may be found that one of the parents has an asymptomatic disease.

Support for confirmatory testing

If a confirmatory test is performed, the Gifu Research Center for Public Health will make a report to the Gifu Prefectural Government's Maternal and Child Health Section and a local public health center. A public health nurse will inquire about your child's physical condition, so please feel free to speak to them about any concerns.

Handling of personal information

The prefectural government collects and analyzes test results, the results of the confirmatory tests, and the status of medical care support. These actions enable the Gifu Prefecture Inborn Errors of Metabolism Testing Evaluation Committee to evaluate the effectiveness of the testing program in reliably detecting sick infants and providing them with appropriate treatment.

Personal information, such as the names of infants and their guardians, obtained in the course of testing and post-test support, will be strictly managed under Gifu Prefecture's Personal Information Protection Ordinance.

If the test reveals an illness...

Your infant will receive treatment from pediatricians who specialize in metabolic disorders and endocrine disorders. In addition, medical expenses may be subsidized for some disorders by the Medical Expense Subsidy System for Chronic Specified Diseases of Children.