

# Re-test after the Test for Inborn Error of Metabolism, etc. and Detailed Medical Examination

## Babies' Guardians Need to Understand the Following Information

### Introduction

The test for inborn error of metabolism, etc. is conducted to detect various diseases before their onset. Generally, development of these diseases or associated disorders can be prevented as long as they are detected at an early stage and appropriate treatment is started. In order to provide appropriate treatment for babies requiring a re-test/detailed medical examination and to assure their sound growth, Hiroshima Prefecture has established a system that provides necessary support for them through the cooperation of laboratories, medical institutions and local governments. This brochure covers all the information that babies' guardians should know, focusing on the examinations and treatments that the babies will undergo and their daily lives in the future. We hope their guardians find this brochure useful.

### Examinations and treatments that babies' guardians should understand

#### Test for inborn error of metabolism

##### Q1. How is the re-test carried out?

The re-test is carried out if the primary test fails to produce a conclusive result as to whether or not a baby is negative for any disorders.

Occasionally, the baby's metabolic functions are partially underdeveloped, which may result in producing abnormal laboratory values.

For this reason, to obtain the correct results of some test items, examinations will be repeated twice or three times.

##### Q2. Will you confirm the safety and accuracy of the examination?

The re-test is the same as the initial examination. A small amount of blood is collected from the baby's heel for this simple examination. The stress on the baby is minimized.

As a result of recent advances in testing equipment, the laboratory data show a remarkably high accuracy.



### Physicians answer your concerns and questions



- Is there any advice that the baby's guardian should follow for the period from the detailed medical examination until notification of a definitive diagnosis?



- Will you give us a list of hospitals specializing in treatment of these diseases?



- Are there any support centers that babies' guardians can consult about diseases, treatments and childcare?



- If the physician in charge gives you any advice about drug administration, special therapeutic milk and feeding intervals, please follow the advice.

If your baby shows any symptoms including vomiting, convulsion or ill-health, please inform the hospital where your baby underwent the detailed medical examination immediately.



- Besides the institutions for detailed medical examination including Hiroshima University Hospital and National Hospital Organization Fukuyama Medical Center, affiliated hospitals and cooperative hospitals in Hiroshima Prefecture can provide appropriate treatment.

The babies referred through a specialized medical institution can receive treatment there.



- Babies' guardians can consult with medical institutions, laboratories, local public health centers or municipal health nurses. Contact information for local public health centers and municipal health nurses is listed on another page. They provide consultation activities to respond to your concerns and questions.



- I am concerned about the possibility of detection of disease in the re-test that may affect future development.



- With the advancement of technology, we can examine for many diseases in newborn mass screening. Treatment has been established for each disease, and, by receiving appropriate treatment, babies can grow up healthily.



## Detailed Medical Examination after Examination for Inborn Error of Metabolism, etc., Disease and Treatment

### Q1. Can you provide more information on endocrine disorders and inborn error of metabolism?

The living body has various "endocrine organs" that produce chemical substances called "hormones" and release them into the blood. Hormones are carried to the target sites through the bloodstream and exert various physiological effects.

As a result of an excessive or insufficient effect of hormones, certain symptoms appear and these conditions are called "endocrine disorders." The newborn mass screening is conducted to examine newborns for diseases resulting from lack of thyroid hormones or adrenal hormones.

The living body constantly carries out "metabolism" to synthesize or degrade various substances including carbohydrates, lipids and protein. Metabolism needs the effects of specific "enzymes" for each substance.

The function of a specific enzyme can decrease and the necessary substances may become deficient or harmful substances accumulate. In this manner, a large variety of symptoms appear and this condition is called "inborn error of metabolism."

### Q2. What is the detailed medical examination process? Is there any physical stress on a baby?

In the examination for inborn error of metabolism, excess or deficiency of metabolites characterizing each disease is checked, while in the examination for endocrine disorders, whether thyroid hormones or adrenal hormones are actually deficient is checked.

Moreover, whether any effect of disease has already appeared is examined. To examine these conditions, blood and urine samples should be collected from a baby. Ultrasonic examination is also used to detect some diseases.

### Q3. Will you explain how a definitive diagnosis is made? Is there any physical stress on a baby?

The extent of examination to make a definitive diagnosis of "disease" or to form the conclusion that a baby is "negative for any disease" differs widely from disease to disease.

In principle, if no abnormality is detected in the medical examination explained in Q2 above, the result of mass screening is regarded as a "false positive." If typical abnormal findings accumulate, the necessity for additional tests should be considered. These tests include measurement of "enzyme activity" and examination of a change in the sequence of genes that form the blueprint of enzymes.

Generally, examinations of enzyme activity and genes are conducted by using blood samples.

### Q4. Will you explain the possible treatments in the future? Is there any physical stress on a baby?

The main treatments for inborn error of metabolism detected in mass screening include dietary therapy and oral administration of a vitamin preparation. A baby with endocrine disorder needs to take drugs to supply missing hormones. These treatments themselves do not have any serious side effects, but depending on disease type or severity, the kinds or amount of food that the baby can consume may be limited.

### Q5. Will you explain the influence of disease on growth or development if a definitive diagnosis of any disease is made?

The diseases to be searched for in newborn mass screening are a group of selected diseases that can be detected and treated immediately after birth. The babies with these diseases can grow healthily as long as appropriate treatment is provided as soon as possible.

The basic treatments for these diseases are daily dietary therapy and oral drug therapy. Several types of inborn error of metabolism and diseases resulting from adrenal hormone deficiency may suddenly cause severe symptoms and leave damage in the event of fever or gastroenteritis associated with infection. Until recovery of the baby's physical condition, glucose may be given intravenously to supply additional energy or the dose of adrenal hormone may be increased. In this manner, acute exacerbation should be controlled.

### Q6. Will you explain the rate of detection of patients after introduction of newborn mass screening by tandem mass spectrometry?

In Hiroshima Prefecture, as the first project in Japan, experimental research has been conducted since 1999, a long time before formal adoption of tandem mass spectrometry. Tandem mass spectrometry was introduced into the local government project in February 2012.

According to the overall patient detection rate, "one in every 4,924 newborns" were affected by some disease, while according to the rate of detection of patients other than those with mild diseases requiring minimum treatment, "one in every 16,412 newborns" were affected by moderate or severe diseases. These patients undergo appropriate treatment and show satisfactory progress.



### Messages from the health nurse



• What consultation service can we receive in the health guidance?

#### • Health guidance

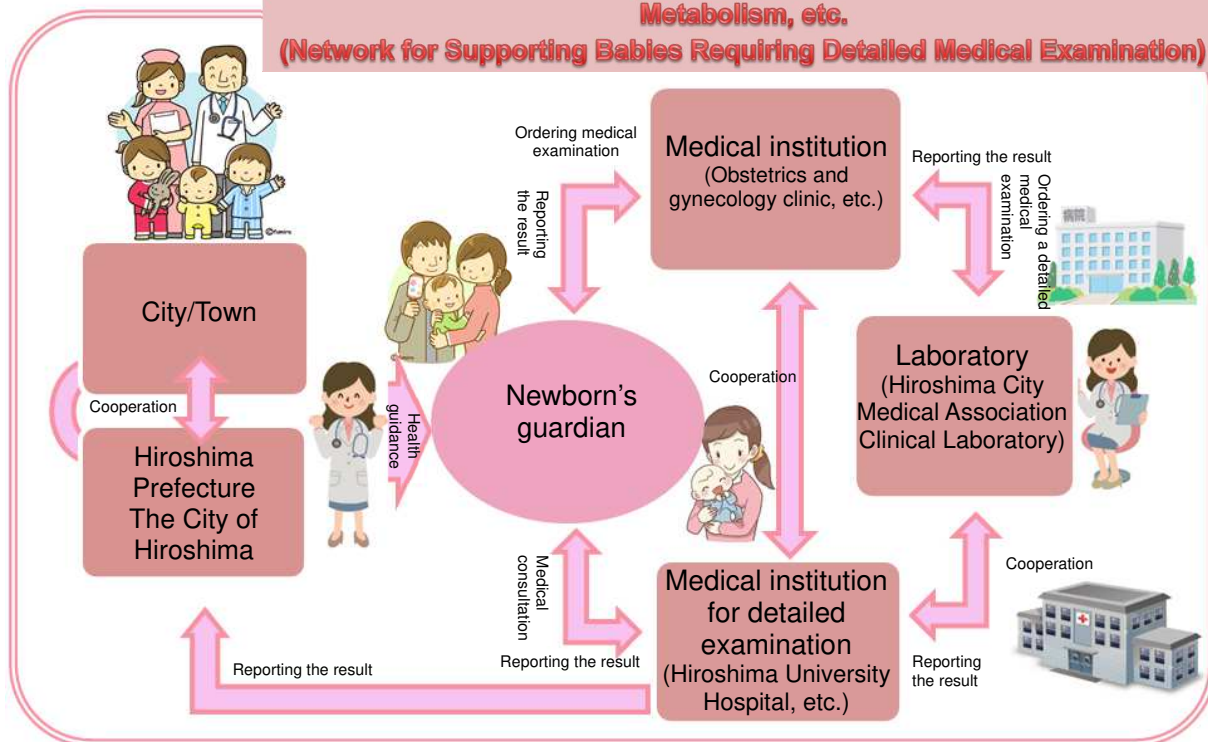
You may receive the notification from the medical institution where your baby underwent blood sampling. The health nurse in charge will call or visit you, provide the relevant information and answer your questions so that your baby can safely undergo detailed medical examination and appropriate treatment.

In cooperation with the medical institutions that provide detailed examination, physicians working for medical institutions and community health nurses, we have established the support network to protect your precious baby so that the baby can receive special treatment immediately after detection of disease by some chance. The personal data are used only when we contact the baby's guardian and thus they are strictly controlled.

• If you have any concerns about the conditions after delivery, diseases, development and your baby's future or any mother-baby problems, you can consult the health nurses in the city/town where you live.

### Hiroshima Prefectural System for Examination for Inborn Error of Metabolism, etc.

(Network for Supporting Babies Requiring Detailed Medical Examination)



### Support for medical treatment expenses, etc.

#### Medical Expenses Subsidy System for Children with Designated Chronic Diseases

Under the "Medical Expenses Subsidy System for Children with Designated Chronic Diseases," part of the medical expenses for treatment of designated chronic pediatric diseases is covered by public funds. Your baby may meet the requirements of this system.

For details, please contact a public health center (branch) in Hiroshima Prefecture or the relevant division of the City of Hiroshima, Kure City or Fukuyama City.

#### Medical Expenses Subsidy System for Children

Under the "Medical Expenses Subsidy System for Children," part of the patient's copayment of expenses to be paid for health insurance treatment for babies/infants at medical institutions is supported by local governments. For details, please contact the division in charge of the city/town where you live.

### Helpful information/related links

• The Pediatric Intractable Disease Consultation Center, Hiroshima University Hospital, provides counseling programs and holds lecture meetings/get-togethers for patients with pediatric intractable disease and their families.



• Incorporated Non-profit Organization Tandem Mass Screening Promotion Association



• Mass-screening Laboratory, National Center for Child Health and Development



• Japanese Society for Mass Screening



## Consultation Offices Including Public Health Centers, etc. in Hiroshima Prefecture

Akitakata City	Hiroshima Branch, Seibu Health Center	Norinchosha 1F, 10-52 Motomachi, Naka-ku, Hiroshima City	082-513-5526
Fuchu-cho			
Kaita-cho			
Kumano-cho			
Saka-cho			
Akiota-cho			
Kitahiroshima-cho			
Etajima City	Kure Branch, Seibu Health Center	3-25, Nishichuo 1-chome, Kure City	0823-22-5400
Takehara City	Seibu Higashi Health Center	13-10 Saijoshowa-machi, Higashihiroshima City	082-422-6911
Higashihiroshima City			
Osakikamijima-cho			
Mihara City	Tobu Health Center	26-12 Kohama-cho, Onomichi City	0848-25-4641
Onomichi City			
Sera City			
Fuchu City	Fukuyama Branch, Tobu Health Center	1-1 Miyoshi-cho 1-chome, Fukuyama City	084-921-1311
Jinsekikogen-cho			
Miyoshi City	Hokubu Health Center	6-1 Tookaichi Higashi 4-chome, Miyoshi City	0824-63-5186
Shobara City			

## Local Community Consultation Offices

Residence (city/town)	Local community consultation offices	Division in charge	Phone number
Hiroshima City	Children/Home Support Department	Mother/Child Healthcare Division	0 8 2 - 5 0 4 - 2 6 2 3
	Naka-ku Naka Healthcare Center	Health Guidance Division	082-504-2109・2528
	Higashi-ku Higashi Healthcare Center	Health Guidance Division	082-568-7735・7729
	Minami-ku Minami Healthcare Center	Health Guidance Division	082-250-4133・4108
	Nishi-ku Nishi Healthcare Center	Health Guidance Division	082-294-6384・6235
	Asaminami-ku Asaminami Healthcare Center	Health Guidance Division	082-831-4944・4942
	Asakita-ku Asakita Healthcare Center	Health Guidance Division	082-819-0616・0586
	Aki-ku Aki Healthcare Center	Health Guidance Division	082-821-2820・2809
	Saeki-ku Saeki Healthcare Center	Health Guidance Division	082-943-9733・9731
Kure City	Higashi Healthcare Center	Group 1	0 8 2 3 - 7 1 - 9 1 7 6
	Nishi Healthcare Center	Group 2	0 8 2 3 - 2 5 - 3 5 4 2
Fukuyama City	Health Center Health Promotion Department	1st Health Promotion Division	0 8 4 - 9 2 8 - 1 2 5 2
Takehara City	Health and Welfare Department	Health Promotion Strategy Division	0 8 4 6 - 2 2 - 7 1 5 7
Mihara City	Healthcare and Welfare Department	Healthcare Promotion Division	0 8 4 8 - 6 7 - 6 0 6 1
Onomichi City	Health Promotion Department	Health Management Division	0 8 4 8 - 2 4 - 1 9 6 0
Fuchu City	Health and Medical Care Department	Healthcare Guidance Division	0 8 4 7 - 4 4 - 6 6 8 8
Miyoshi City	Health Promotion Department	Health Promotion Division	0 8 2 4 - 6 2 - 6 2 5 7
Shobara City	Healthcare and Medical Care Department	Health Promotion Division	0 8 2 4 - 7 3 - 1 2 1 4
Otake City	Healthcare and Medical Care Department	Healthcare/Disease Prevention Division	0 8 2 7 - 5 9 - 2 1 4 0
Higashihiroshima City	Children/Home Service Department	Mother/Child Healthcare Division	0 8 2 - 4 2 0 - 0 4 0 7
Hatsukaichi City	Childcare Support Office	Healthcare Group	0 8 2 9 - 3 0 - 9 1 8 8
Akitakata City	Health and Longevity Department	Health Promotion Division	0 8 2 6 - 4 2 - 5 6 3 3
Etajima City	Healthcare and Medical Care Department	Health Promotion Division	0 8 2 3 - 4 2 - 2 8 5 2
Fuchu-cho	Health Promotion Department	Health Enhancement Division	0 8 2 - 2 8 6 - 3 2 5 8
Kaita-cho	Healthcare Center	Health Enhancement Division	0 8 2 - 8 2 3 - 4 4 1 8
Kumano-cho	Childcare/Health Promotion Department	Health Enhancement Group	0 8 2 - 8 2 0 - 5 6 3 7
Saka-cho	Insurance/Healthcare Department	Insurance/Healthcare Division	0 8 2 - 8 2 0 - 1 5 0 4
Akiota-cho	Healthcare Guidance Department		0 8 2 6 - 2 2 - 0 1 9 6
Kitahiroshima-cho	Health Management Department	Health Enhancement Division	0 5 0 - 5 8 1 2 - 1 8 5 3
Osakikamijima-cho	Health/Hygiene Department	Healthcare Guidance Division	0 8 4 6 - 6 2 - 0 3 3 0
Sera-cho	Childcare Support Department	Childcare Support Division	0 8 4 7 - 2 5 - 0 2 9 5
Jinsekikogen-cho	Health Management Department	Childcare Support Division	0 8 4 7 - 8 9 - 3 3 6 8

**Publisher : Children's Future Division, Hiroshima Prefecture**

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Date of publication: June 2017, February 2023

Website



Iku-chan  
(mascot character)