

Newborn Mass Screening for your baby's health



● What is “newborn mass screening”?

Newborn mass screening is essential for all newborns who are 4 – 7 days old.

Some congenital diseases may cause disability if they go undetected and ignored. Early identification and the appropriate treatment can help prevent babies from developing intellectual and developmental disabilities. Also, children who receive treatment will be able to minimize severe symptoms in their daily lives. Japan’s newborn screening system initially covered five disorders when it began in 1977. Since then, improved technology has enabled the screening to deal with a wider array of disorders.

● What kind of disorders does newborn mass screening cover?

Newborn mass screening covers two types of endocrine disorders (hormone abnormality) and 17 types of metabolic disorders (defective nutrient utilization); the screening may also sometimes detect 7 additional disorders.

Newborn mass screening is publicly funded, and carried out by the governments of Japanese prefectures and ordinance-designated cities. Obstetric hospitals/clinics accept screening applications.

Before the screening, a doctor or other medical staff will explain the importance of mass screening prior to taking a blood sample. The screening is practiced only with the family’s consent (The family expresses its willingness by filling out the application form). Today, 100% of the babies intended take the test in Hokkaido, though there are a couple of exceptional cases.

If the test diagnoses that a more detailed test will be necessary, the test result and the baby’s contact address will be reported to the medical institution, consultant doctor(s) and the public health center. The reported information is used by a nurse who belongs to the public health center or a municipal office in order to contact the baby’s legal guardian.

For babies who are born in Hokkaido, except in Sapporo, the Hokkaido Pharmaceutical Association Public Health Examination Center conducts newborn mass screening. For those born in Sapporo, the Sapporo Institute of Public Health performs this role.

For further information about the system, please contact:

**Child-raising Support Group, Bureau of Promotion for the Future of the
Children, Department of Health and Welfare
Hokkaido Government**

Tel: 011-231-4111 (ext. 25-770)

(For the babies born in Sapporo, please contact the City of Sapporo.)

FAQ: Newborn Mass Screening

What is the purpose of newborn screening?

A Babies who are apparently healthy may have congenital diseases/disorders. The screening aims to detect disorders which are treatable and preventable at the earliest time possible. Early detection and appropriate treatment may keep babies from developing intellectual disability or developmental disorders.

What types of disorders can be detected by the screening?

A The screening is designed to detect the following disorders. Newly-introduced tandem mass spectrometers have helped detect a wider array of disorders.

Endocrine Disorders

Congenital hypothyroidism (Cretinism)

This is a shortage of the thyroid hormone, which may cause growth retardation or intellectual disability.

Congenital adrenal hyperplasia

A shortage of corticosteroid can be life-threatening particularly when the growth is inadequate or if children become severely dehydrated.

Metabolic Disorders

Galactosemia

Inability to process galactose sugar, commonly contained in milk, may cause mental retardation or liver damage in babies.

Amino acid metabolism disorder (Aminoacidopathy)

Proteins from food normally break down to amino acids. However, a metabolism failure causes unprocessed, accumulated amino acids to develop into a physical disorder. There are many different disorders where the baby is unable to process amino acids properly: phenylketonuria, maple syrup urine disease, homocystinuria, citrullinemia I, argininosuccinic aciduria.

Organic acid metabolism disorders

This occurs when organic acids (the metabolism intermediate called *carboxylic acid*, which is created during the process of amino acid resolution) accumulates in the body caused by the failure of the intermediate metabolism. Individual disorders vary by the form of organic acid: methylmalonic academia, propionic academia, isovaleric academia, methylcrotonylglycinuria, hydroxymethylglutaryl (HMG) academia, multiple carboxylase deficiency and glutaric academia type I.

Fatty acid metabolism disorders

A shortage of carbon hydrate-derived energy makes fatty acid removed from neutral fat to act as an alternative energy to supplement the shortage. However, metabolic failure causes a chronic failure of energy production. Disorders, varying by the form of fatty acids, include MCAD deficiency, VLCAD deficiency, TFP/LCHAD deficiency, or CPT-1 deficiency.

Do all newborns have to take a screening test?

A The test is not mandatory, but all newborns are recommended to take one because inherited abnormalities may cause disabilities if the babies do not take a test and result in developing a disease. Newborn mass screening, which began in Japan in 1977, is a public-funded program for disability prevention. Today most newborns take the test.

**What is the screening test process?**

A A blood test is carried out 4 to 7 days following a baby's birth at a medical institution, whereby a drop of blood is drawn from the heel and sent to the examination center. Babies whose blood sample tested positive for a disorder are required to see a professional doctor in their area, and then receive the necessary treatment and daily life guidance.

Does it cost to take a screening test?

A The tests are free of charge, though you may be asked to pay for blood sampling. For details about the necessary cost, please ask your hospital/clinic.

How will I learn of the test result?

A If the result is negative, it will be sent to the hospital/clinic where the baby's blood was sampled. If positive, the result will be communicated immediately, through the hospital/clinic, to the baby's guardian. The examination center does not accept individual inquiries.

What is the difference between re-test (repeat specimen test) and medical examination?

A A re-test (repeat specimen 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. One in 100 babies is normally required to take a re-test. If the primary test and re-test show positive, the baby must take a thorough medical examination at a special pediatric hospital in order to correctly diagnose any disease/disorder. Please note that all babies under thorough examination will not always be deemed as patient; some babies are diagnosed negative at this stage.

What happens if a baby is diagnosed a possibility of disease/disorder?

A The result will be communicated through the hospital/clinic where the baby took the test. The

name of medical institutions where re-tests and treatments are available will also be informed so that guardians are able to discuss what to do next. Medical institutions for thorough examination, expert consultant doctors and local public health nurses are all cooperative and prepared to promptly provide babies with all the specialized treatments they need should a baby be diagnosed with a disease/disorder. Private information will be strictly protected and managed.



- **For families:**

With technological advancements, an increasing type of disease/disorder is diagnosed through newborn mass screening. Every disorder can be effectively treated, which means that the appropriate treatment will help newborns develop healthily.

Some diseases, symptoms or ill-health may appear prior to the test results; still, early detection leads to more prompt treatment and children's healthy development. For appropriate treatment, mass screening is expected to detect abnormalities at the earliest time possible.

Newborn mass screening may detect diseases/disorders which are not included in the intended 19 diseases (Present screening is capable of detecting 26 diseases altogether). If these diseases are diagnosed, a detailed explanation will be given at your medical institution when undergoing a thorough examination.

Filter papers used to take blood and the screening data are stored at the examination center for 3 years and 5 years, respectively, before their disposal. The screening data for thorough examinations is stored indefinitely for follow-up and treatment purposes.

The examination center counts the number of babies undergoing a thorough examination, for the purpose of assessing the accuracy of mass screenings, the prevalence of diseases/disorders and the effectiveness of treatments, as well as improving the mass screening system.

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