Neonatal Screening

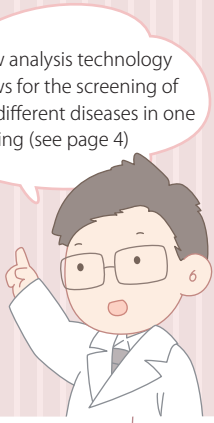
Pamphlet
Application
Form

Neonatal Screening

Neonatal screening is a test that can identify over 20 different disorders in newborns with only a few drops of their blood. The test aims to support the healthy development of your baby through early disease detection.

The screening is offered free of charge* to all newborns delivered in Sapporo. Neonatal screening is conducted by local governments throughout the country.

*Blood sampling and other hospital fees related to the screening are separately charged.

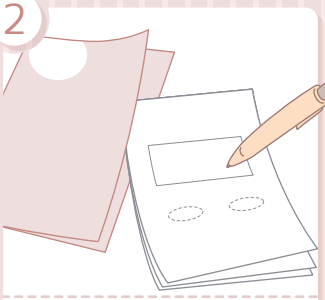


How to apply for neonatal screening

1

Read this pamphlet thoroughly.

2



Fill out the application form included in this pamphlet.

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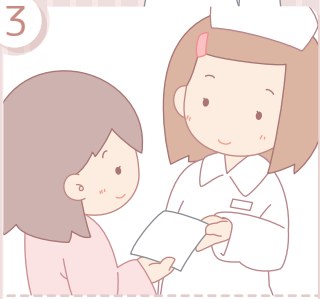

Submit the application form to your maternity hospital.

Contact:

Institute of Public Health　Health Science Section (Maternal and Child Screening)
Kikusui 9-jo 1-chome, Shiroishi-ku, Sapporo 003-8505
Tel.: 011-841-7672

sapporo　screening

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The neonatal screening is a public program implemented in partnership with the city of Sapporo (Institute of Public Health, Public Health Office, and ward health centers), Sapporo Medical Association (Sapporo Association of Obstetricians and Gynecologists), maternity hospitals and medical institutions conducting clinical evaluation.

Neonatal Screening Q & A

How can I apply for the screening?

Please fill out the application form and submit it to your maternity hospital. Blood samples are collected from your child at the hospital and they are sent to the Sapporo Institute of Public Health for screening. You will receive the results of the screening at your baby's one-month health check-up. If any diseases are detected, however, you will be informed of the results immediately.

How strenuous is the screening on my baby?

Several drops of blood are obtained by pricking your baby's heel. The sampling collects only small amounts of blood and it places very little stress on newborns.

Is neonatal screening a mandatory test for all newborns? How much is the screening fee?

Neonatal screening is not mandatory, however it is recommended for all newborns to take because some of the diseases that can be detected by the screening may cause severe impairment if left untreated. Neonatal screening is provided by all municipal governments throughout the country free of charge*. Apply for the screening to ensure the healthy development of your child.

*Blood sampling and other hospital fees relating to the screening are separately charged.

If my child has diseases, would not my maternity doctor find them immediately?

Some newborns who look healthy may have conditions which do not readily show symptoms. There are diseases which may cause sudden death as the result of catching a cold, or those which may lead to developmental disabilities. Such diseases can be prevented or properly treated if they are detected early even before the appearance of any symptoms.

The screening results indicate that my child needs a second screening. Does this mean that my child may have a disease?

The call for a second screening does not necessarily mean that your child has a disease. A second screening is conducted to confirm the results from the primary screening when some conditions are not able to be correctly determined due to various factors other than diseases, such as a child' s physical condition.

If a detailed examination is advised does it mean that my child has developed a disease?

The call for a detailed examination does not always mean that your child has a disease. The neonatal screening alone cannot determine for certain whether or not your child has a disease. It is necessary to see specialists and undergo a detailed examination for a proper diagnosis.

What are the percentages of actual disease cases found in second screenings and detailed examinations?

Diseases are found roughly in one in 20 children subjected to a second screening and one in four children subjected to a detailed examination.

What should I do if my child has a disease?

Many diseases detected by the neonatal screening can be prevented or treated if they are discovered early. One of the purposes of the neonatal screening is to provide the diagnosed newborn with treatment support (e.g., doctor referral). Treatment costs are partially covered by a subsidy program by the city of Sapporo.

Diseases Screened for by the Neonatal Screening

Endocrine disorders

Endocrine disorders are caused by an abnormal secretion of hormones. They are treated by the oral administration of hormone supplement medication.

Congenital hypothyroidism (cretinism)	A disease caused by a lack of hormones secreted by the thyroid gland, causing conditions such as poor development and mental retardation
Congenital adrenal hyperplasia	A disease caused by the failure to produce adrenal hormones, causing conditions such as dehydration and poor development

Metabolic disorders

Metabolic disorders are diseases caused by defects in the metabolism of amino acids or sugars, or failure in the energy production process. Treatments include the prescription of a special formula milk and modified diet as well as medication, and lifestyle guidance.

23 diseases to be screened for by tandem mass spectrometry (a new analysis method)

Tandem mass spectrometry allows for the screening of many different diseases in one sitting. In Sapporo, the following 23 diseases are screened for by tandem mass spectrometry.

Amino acid metabolism disorders (6 diseases) *Diseases screened for by the conventional neonatal screening method are also analyzed by the new tandem mass spectrometry.	Phenylketonuria*	Amino acids are compounds which build proteins. These disorders occur when certain types of amino acids fail to be utilized or decompose properly and accumulated in the body, causing mental retardation or severe health conditions.
	Maple syrup urine disease*	
	Homocystinuria*	
	Citrullinemia Type I	
	Argininosuccinic aciduria	
	Citrin deficiency	
Organic acid metabolism disorders (9 diseases)	Methylmalonic acidemia	Organic acids are compounds converted from amino acids within the body. These disorders occur when certain types of organic acids fail to be metabolized and accumulate in the body, causing poor suckling, nausea and convulsions.
	Propionic acidemia	
	Isovaleric acidemia	
	Methylcrotonylglycinuria	
	3-hydroxy-methylglutaric aciduria (HMG)	
	Multiple carboxylase deficiency	
	Glutaric aciduria type I	
	β-ketothiolase deficiency	
	Methylglutaconic aciduria	
Fatty acid metabolism disorders (8 diseases)	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	Fats are decomposed to produce energy when the body does not produce enough energy through food consumption (e.g., when the stomach is empty, during exercise, etc.). When the process of energy production is not functioning well, the lack of energy causes severe health conditions.
	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	
	Long-chain 3-hydroxy acyl-CoA dehydrogenase (LCHAD) deficiency	
	Carnitine palmitoyltransferase I (CPT I) deficiency	
	Carnitine palmitoyltransferase II (CPT II) deficiency	
	Carnitine-acylcarnitine translocase deficiency	
	Carnitine transporter deficiency	
	Glutaric aciduria type II	

Galactosemia	When galactose (a sugar found in foods) is not properly decomposed and builds up in the body, mental and physical retardation, cataracts, or other conditions may develop.