

Newborn Screening

Basic Information about Newborn Screening

What are the disorders included in the Newborn Screening Package?

1. Congenital Hypothyroidism (CH)

CH results from lack or absence of thyroid hormone, which is essential to growth of the brain and the body. If the disorder is not detected and hormone replacement is not initiated within (4) weeks, the baby's physical growth will be stunted and she/he may suffer from mental retardation.

2. Congenital Adrenal Hyperplasia (CAH)

CAH is an endocrine disorder that causes severe salt lose, dehydration and abnormally high levels of male sex hormones in both boys and girls. If not detected and treated early, babies may die within 7-14 days.

3. Galactosemia (GAL)

GAL is a condition in which the body is unable to process galactose, the sugar present in milk. Accumulation of excessive galactose in the body can cause many problems, including liver damage, brain damage and cataracts.

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4. Phenylketonuria (PKU)

PKU is a metabolic disorder in which the body cannot properly use one of the building blocks of protein called phenylalanine. Excessive accumulation of phenylalanine in the body causes brain damage.

4. Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD Def)

G6PD deficiency is a condition where the body lacks the enzyme called G6PD. Babies with this deficiency may have hemolytic anemia resulting from exposure to certain drugs, foods and chemicals.

What is Newborn Screening?

Newborn Screening (NBS) is a simple procedure to find out if your baby has a congenital metabolic disorder that may lead to mental retardation and even death if left untreated.

Why is it important to have Newborn Screening?

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Most babies with metabolic disorders look normal at birth. One will never know that the baby has the disorder until the onset of signs and symptoms and more often ill effects are already irreversible.

When is Newborn Screening done?

Newborn screening is ideally done on the 48th hour or at least 24 hours from birth. Some disorders are not detected if the test is done earlier than 24 hours. The baby must be screened again after 2 weeks for more accurate results.

How is Newborn Screening done?

Newborn screening is a simple procedure. Using the heel prick method, a few drops are taken from the baby's heel and blotted on a special absorbent filter card. The blood is dried for 4 hours and sent to the Newborn Screening Laboratory. (NBS Lab).

Who will collect the sample for Newborn Screening?

A physician, a nurse, a midwife or medical technologist can do the newborn screening.

Where is Newborn Screening Available?

Newborn screening is available in practicing health institutions (hospitals, lying-ins, Rural Health Units and Health Centers). If babies are delivered at home, babies may be brought to the nearest institution offering newborn screening.

When is the Newborn Screening results available?

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Newborn screening results are available within three weeks after the NBS Lab receives and tests the samples sent by the institutions. Results are released by NBS Lab to the institutions and are released to your attending birth attendants or physicians. Parents may seek the results from the institutions where samples are collected.

A negative screen mean that the result of the test is normal and the baby is not suffering from any of the disorders being screened.

In case of a positive screen, the NBS nurse coordinator will immediately inform the coordinator of the institution where the sample was collected for recall of patients for confirmatory testing.

What should be done when a baby has a positive newborn screening result?

Babies with positive results should be referred at once to the nearest hospital or specialist for confirmatory test and further management. Should there be no specialist in the area, the NBS secretariat office will assist its attending physician.

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Disorder Screened	Effects if NOT SCREENED	Effects if SCREENED and treated
CH (Congenital Hypothyroidism)	Severe Mental Retardation	Normal
CAH (Congenital Adrenal Hyperplasia)	Death	Alive and Normal
GAL (Galactosemia)	Death or Cataracts	Alive and Normal
PKU (Phenylketonuria)	Severe Mental Retardation	Normal
G6PD Deficiency	Severe Anemia, Kernicterus	Normal