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Newborn screening program

Newborn screening program

- It aims to save lives and raise awareness among parents about newborn screening.
- It is considered the first step in a child's life to get a healthy start.

Parents wish to have a healthy baby. as newborns are screened early to ensure there are no serious disorders which often cannot be seen at birth. These disorders can be found by taking a drop of the newborn’s blood, in addition to a hearing test and pulse oximetry. If a disorder is found early through one of these tests, early intervention can help diagnose the disease and give these children a new healthy start in life. Nevertheless, many parents are still unaware of the newborn screening system and its role in early detection of cases of hereditary and other diseases.

What is newborn screening?

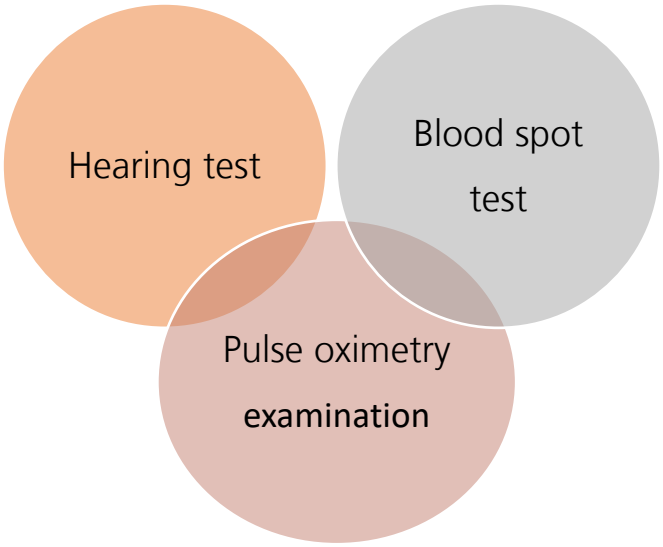
It is a screening process for newborn babies to detect serious health conditions associated with the newborn that can be treated if diagnosed early. A small number of babies who appear healthy at birth have a rare birth defect or disorder, and screening tests check to see if your newborn has one of these diseases, if it is found early, we can help prevent serious problems, such as intellectual or developmental disabilities or death. Furthermore, the blood of all children born in the Kingdom of Saudi Arabia is tested for a number of genetic disorders or rare medical conditions, which complications can be prevented if diagnosed early.

Why does my baby need a newborn screening?

Most babies with metabolic disorders appear “normal” at birth. Through newborn screening, metabolic disorders can be detected even before clinical signs and symptoms appear, and treatment can be given early to prevent complications of untreated conditions.

What does newborn screen include?

It includes three tests:



1. Blood spot test:

It now includes 20 rare diseases such as metabolic and endocrine and hematology diseases. Early detection can help prevent serious complications such as severe disability.

What disorders are included in the blood spot test?

The newborn screening panel currently includes: phenylketonuria, maple syrup urine disease, arginosuccinase deficiency, citrullinemia,HMG-CoA layase deficiency, isovaleric acidemia, methylmalonic acidemia, propionic acidemia, beta-ketothiolase deficiency, methylcrotonyl-CoA carboxylase deficiency, glutaric acidemia type-1,medium-chain acyl-CoA dehydrogenase deficiency, galactosemia, congenital hypothyroidism, congenital adrenal hyperplasia, , G6PD deficiency, biotinidase deficiency, very long chain acyl-CoA dehydrogenase deficiency, sickle cell disease.

2. Hearing test:

A hearing test uses a microphone or electrodes to determine whether the infant's ear and brain respond appropriately to sound. The test helps identify early hearing loss in children and maintain speech, language, and communication skills.

3. Pulse oximetry examination:

A sensor is placed on the skin to measure the level of oxygen in the blood through the skin. In case of low oxygen levels, children are referred to a cardiologist. This helps in early detection of a group of serious, life-threatening heart defects known as critical congenital heart disease. Note that congenital heart diseases can often be treated with surgery or other medical interventions.

How will my child be tested?

A few drops of blood are taken from the baby's heel, then it is swabbed on a special newborn screening card and sent to the National Newborn Screening Program.

Your nurse will complete a newborn screening card which includes the infant's name, sex, weight, date and time of birth, and date and time of blood collection.

This card consists of special absorbent paper used to collect a blood sample, after which a child's health care provider reports the results.

When is newborn screening done?

- Newborn screening is performed within the first hours (24 to 48) after birth.

What does the newborn screening result mean?

- A negative result means that the newborn screening result is normal. If the result is positive, the newborn must be returned to the hospital for further testing.

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

- Children with positive results should be immediately referred to the treating physician for confirmatory testing and action should be taken based on the results

Frequently asked questions

❖ If we do not have any hereditary diseases in the family, is there a risk?

most children diagnosed with metabolic diseases usually come from consanguine families , by early diagnosis we can prevent the serious complication by starting early intervention and treatment.

❖ How does the program help parents and children?

The program will notify your child's doctor if they test positive, and may contact you to confirm additional testing, final diagnosis and enrollment in the early intervention program as soon as possible and it will prevent complications.

❖ What if my child's test indicates a positive result?

These are screening tests only, not diagnostics. If your baby have positive results, additional tests may be required, leading to a diagnosis and subsequent monitoring and treatment.

❖ What if my child was diagnosed with any of these diseases? Will he will be like any healthy child?

Most children with one of these diseases can grow and develop normally if the condition is identified and treated early. Sometimes, lifelong treatment and monitoring is required. It is important to act quickly if your child needs further tests or treatment.

❖ If my child suffers from one of these disorders, can it be cured?

We cannot cure these disorders. But early treatment may prevent or control serious health problems. Doctors who specialize in these disorders are available to help diagnose and treat affected children early to prevent severe complications. Your doctor may refer you to one of these specialists.

Sources and references:

All illustrations from canva.com

Review and audit:

The content of this booklet was reviewed by genetic and metabolic disease consultants at King Fahd University Hospital.

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