

فقيه.

أكاديمية فقيه الطبية
Fakeeh Care Academy

فقيه.

مجموعة فقيه للرعاية الصحية
Fakeeh Care Group



NEWBORN SCREENING TEST

NATIONAL PROGRAM FOR EARLY SCREENING OF NEWBORNS
TO PREVENT DISABILITY

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What are metabolic diseases?

Metabolic disorder or metabolic diseases are part of the genetic diseases that are transmitted from what is known as the recessive gene, that is, the mother and father carry the genetic characteristics of the disease and transmit it to their children by 25% in each pregnancy (the ratio of an infected newborn to three uninfected) and the known metabolic diseases are estimated at more than 400 metabolic diseases.

The nature of metabolic diseases is the lack of certain enzymes from birth, and these enzymes are important for converting a food substance into another in different parts of the body.

Purpose of the Newborn Screening Program:

Early screening of newborns to reduce morbidity, disability and mortality rates resulting from genetic diseases (metabolic and endocrine diseases included in the program).

My child seems healthy, is the screening necessary for him?

Yes, it is necessary, as most children whose diseases are discovered through the early screening do not show any symptoms immediately after birth, as they may appear later over time.

There is no sufferers in my family; does my child need this screening?

A family with healthy children does not expect any health problems at all, and this may be true, but the absence of a previous history in the family does not negate the possibility of these diseases to occur, so it is necessary to perform this screening for all births without exception.

Diseases covered by the screening program:

Amino acid diseases:

- Phenylketonuria.
- Maple urolithiasis.
- Homocystinuria.

Organic acid diseases:

- Propionic acidity.
- Acidity of methylmalonic.
- 3-MCC enzyme deficiency
- Biotinidase deficiency.
- Glutaric acidity – type I.
- acidity of isovaleric.
- Co-I-Lase deficiency

Urea cycle diseases:

- acidity of argininosuccin.
- Strollinemia.

Fat diseases:

- Lack of the enzyme M-CAD.
- Enzyme deficiency in VL CAD.

Diabetes:

- Glycemia.

Endocrine diseases:

- Hypothyroidism.
- Hypercloning of adrenal cells.

How to get my child screened?

The sample is taken 24–48 hours after the child's birth and before leaving the hospital. A few drops of your child's blood are taken from his/her heel by pricking it with a small medical needle, placed on a special paper card, and then sent to the laboratory after drying it.

How to get the result?

- You will not receive a call if the results of the screening are good.
- If there is an abnormal sign in the screening or the sample is insufficient, newborn screening coordinator or the pediatrician will call you and direct you to what you should do.
- You can also inquire about the results at your first visit to your child's doctor.

The early newborn screening detects if your baby is exposed to a high or low risk of these diseases, but it is not a positive or negative test, or what is known as a diagnostic test. If your child has any symptoms, his doctor must carry out the appropriate diagnostic tests. It is important to remember that newborn screening not checking all serious medical diseases.

Does the repetition of the screening mean that my child is infected?

The repetition of the screening does not necessarily mean that the child has a disease. The screening may sometimes be repeated if there is a problem in the drawing of the sample or while sending it to the laboratory. A positive result means that your child has a higher probability of developing a disease and it always requires repetition of the screening and doing diagnostic tests to see if the child has this disease or not.

How can I help the hospital to take care of my child?

- You must make sure that you have told the doctor about any hereditary disease in your family.
- You must make sure that the sample is taken from him before leaving the hospital.
- You must make sure that the hospital has your correct phone numbers to call you when needed, and update it if necessary.
- You must respond quickly if there is a need for repeating the screening or further screenings.

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