Ministry of Health

The National Newborn Screening Program

Early Detection of Metabolic and Endocrine Disorders

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What is the National Newborn Screening Program?

It is a preventive national program for newborns which includes a package of screening tests.

What is the National Newborn Screening Program for Early Detection of Metabolic and Endocrine Disorders in newborn?

It is one of the preventive screenings recently added to the current packages of screening in the National Newborn Screening program which include clinical examination, hearing tests, and screening for congenital hypothyroidism. This expansion aims to detect a range of metabolic disorders and some endocrine function disorders that may affect the child's health and development.

What disorders are included in early newborn screening program?

Metabolic disorders, which affect the body's ability to process certain metabolic substances or eliminate toxins associated with metabolic reactions. Examples include phenylketonuria and urea cycle disorders.

Endocrine disorders, which affect the levels of hormones necessary for the body's functions. Examples include congenital hypothyroidism and adrenal gland disorders.

The National Program for Early Detection of Metabolic and Endocrine Disorders covers 22 biochemical markers that allow for the detection of 26 types of such diseases.

What are inborn metabolic and endocrine disorders?

Inborn metabolic disorders are genetic conditions that result in the inefficiency of one or more metabolic reactions, leading to a variety of health issues associated with the accumulation of metabolic intermediates or toxins, or a deficiency in vital metabolic products. These disorders may occur due to a deficiency in one or more enzymes or a failure in the co-factors that aid these reactions. Endocrine disorders result from congenital or acquired deficiencies in the secretion or stimulation of one of the endocrine hormones.

Why will the tests be conducted even if no symptoms are present?

The preventive importance of this program lies in enabling medical professionals to diagnose affected newborns before symptoms appear, especially before irreversible complications occur. Many of these diseases are rare and not commonly considered in routine evaluations, and some may present only after permanent damage occurs or even result in unexpected death. Early diagnosis allows for timely intervention, thereby preventing complications.

Do I have the right to refuse the testing for my child?

The preventive screening for metabolic and endocrine disorders is a vital national program that the Ministry of Health in Oman provides for all newborns. According to the Child Law (article 21) , parents or guardians are required to carry out the necessary examinations and tests for their child. If a parent or guardian refuses the test, legal actions will be taken according to the Child Law.

CHILD LAW

When will the blood sample be taken for the screening?

> The test will be carried out within 24 -48 hours after birth, and before the newborn is discharged from the healthcare facility.

How will the sample be taken?

> A few drops of blood will be collected from the baby's heel, as this is considered as the best area for sample collection for this test.

How will I know the test results?

If the result is normal «negative», the family usually will not be contacted. However, contact will be made if:

- The sample was taken before 24 hours after birth.
- The initial result was inconclusive.
- The initial blood sample was insufficient for analysis.
- The initial result was positive.

If you have been contacted by hospital team, please visit the health facility as soon as possible and follow the doctor's instructions.

Remember

The call for re-testing does not mean that your child is affected.

What does it mean if I am contacted with a positive preliminary result?

The preliminary test is a screening, and not a definitive diagnosis. If the result is positive, the medical team will contact the family to discuss the child's health and schedule an appointment for further clinical evaluation, either at the newborn screening clinic or at the nearest specified hospital in the governorate. During this visit, the child will be examined by a specialist or trained nurse to complete the necessary tests for confirming or ruling out the disease.

Therefore, it is crucial to provide up-to-date and correct contact information in the child›s electronic health record.

Do not worry.

The treating team will counsel you regarding your child condition, answering your queries and will develop a plan for management and follow up.

Is there treatment available for the disorders included in this early screening?

Yes, treatment plans are available for all disorders covered in the comprehensive newborn screening program. Treatments may include dietary adjustments, medications, or a combination of both.

Does a negative test result mean my child will not develop any inborn metabolic or endocrine disorders later?

The program only detects 26 disorders, which are expected to appear during the first few days, weeks, or months of a child's life. Milder forms, which may present later, may not be detected by this program. Additionally, the program does not detect disorders outside the scope of the screening criteria set by specific biochemical markers.

Early detection of metabolic and endocrine disorders in newborns contributes to reducing the complications of these disorders.