MEDICOVER
WOMAN & CHILD
HOSPITALS

NEWBORN SCREENING FOR BABIES

Newborn Screening for babies

1. Newborn screening is the first and most important test to be done for the baby.
2. It is a simple procedure that checks the baby for hidden medical conditions that cannot be identified by the physician just by looking at the baby.
3. These conditions if left undetected can lead to serious illness or brain damage.
4. Newborn screening enables early detection and treatment that can help the baby lead a normal life.

The Newborn Screening Test screens the baby for many conditions under:

1. Haemoglobinopathies - Genetic disorders resulting in faulty hemoglobin, includes sickle cell disease, Thalassemia and so on.
2. Endocrine Disorders - Conditions causing hormonal excesses or deficiencies, include Hypothyroidism.
3. Carbohydrate Metabolism Disorders - Conditions affecting the metabolism of carbohydrates, include Lactose Intolerance.
4. Protein Metabolism Disorders - Classical Hyperphenylalaninemia.
5. Inborn Errors of Metabolism - Genetic conditions affecting metabolism.
6. In India, only less than 1% of infants are screened and **Prevalence of inborn errors of metabolism in India is nearly 1 in 2500**

Why Newborn Screening & when to do it?

1. A newborn may appear normal at birth but may have a hidden medical condition.
2. As the child grows, these conditions could later manifest into serious illnesses and affect the growth and life of the child.
3. Newborn screening test is to be done between 48 & 72 hours from the time of the baby's birth.
4. It is important that parents get this test done for their babies before leaving the hospital.
5. However it is also recommended that older babies who did not undergo screening can avail test irrespective of their current age.

Why is newborn screening essential?

1. Newborn screening checks for rare, serious but treatable conditions at birth before they cause irreversible damage to the baby.
2. Sometimes a baby could have a condition even if there is no family history.
3. Early detection allows early treatment which usually involves a change in diet or medication that can help the baby live a normal life.

Below are the Neonatal Screening - 11 conditions, Causes and Effects:

S.NO	CONDITIONS	EFFECTS IF UNTREATED
1	Biotinidase deficiency (Causes due to enzymatic defect in late-onset multiple carboxylase deficiency)	Scaly perioral & Facial rash, Mental retardation
2	Congenital Adrenal Hyperplasia (Causes due to the deficiency of 21-Hydroxylase)	Weight loss, Dehydration, Genital problems, Infertility
3	Cystic Fibrosis (Disease caused by defects in the CFTR gene)	Delayed growth, Infertility, Serious lung problems
4	Congenital Hypothyroidism (Causes due to an anatomic defect in the gland, an inborn error of thyroid metabolism or iodine deficiency)	Mental retardation, Abnormal facial features
5	G6PD deficiency (It is a genetic condition caused by lack of the enzyme G6PD)	Chronic anemia
6	Galactosemia (It is a genetic disorder where galactose (a byproduct of lactose digestion) fails to convert to glucose)	Liver Dysfunction, Cataracts
7	Sickle Cell Anemia (Hb SS) (It is caused by an inherited abnormal hemoglobin (the oxygen-carrying protein within the red blood cells)	Hemolytic anemia, Vasoocclusive complications
8	Sickle Cell Disease (Hb S/C) (It is a abnormal hemoglobin, called hemoglobin S, which causes sickle cell disease (SCD))	Infection, Sepsis, Growth delay, Hemolytic anemia, Vaso-occlusive complications
9	Beta thalassemia (It is a disorder characterized by a genetic deficiency in the synthesis of beta-globin chains of Haemoglobin)	Severe anemia, Bone deformity, Growth failure
10	Variant hemoglobinopathies including Hb E (Var Hb) (Genetic defect that results in abnormal structure of one of the globin chains of the hemoglobin molecule)	Hemolytic anemia, Splenomegaly
11	Hyperphenylalaninemia (HPHE) (Inherited disorder that increases levels of the amino acid phenylalanine in the blood)	Brain damage