Early Assurance:

The Vitality of Newborn Screening

Fernandez

Built for Birthing

A Drop of Blood Can Prevent Disabilities and Save Lives!

Newborn screening

The birth of a newborn baby brings unbound happiness to the new parents. Their joy doubles when the baby looks normal and healthy. But those joyous moments can be short-lived if the baby is affected by any serious/life-threatening disorders that are not apparent at birth. Newborn screening tests give you an opportunity to identify those rare disorders in babies soon after their birth.

When and how babies are screened

A SIMPLE BLOOD TEST

A trained staff will take a few drops of blood from the baby's heel. The blood sample is sent to a newborn screening lab for testing.

- The test is performed when the baby is 48-72 hours old, by a simple prick on the baby's heel and collecting a few drops of blood on a special filter paper card.
- The heel prick does not cause any harm to your baby. A fresh, disposable, specialised needle called a 'Lancet' is used for every baby, hence there is no chance of your baby getting any infections.

Should all babies be screened?

All babies should be screened, even if they look healthy, because some medical conditions cannot be seen by just looking at the baby. Finding these conditions soon after birth can help prevent some serious problems, such as brain damage, organ damage, and even death.

Because these disorders are present at birth and are often life-threatening, it becomes even more important to identify babies with these disorders as early as possible and when asymptomatic. Otherwise, the damage done may be permanent and irreversible.

Conditions Tested

More than 40 conditions are tested in newborn screening.

These conditions are mainly related to:

- Metabolic conditions that affects how the body processes food.
- Endocrine conditions which affects the levels of important hormones.
- Pulmonary conditions that affects growth and lungs.

SCREENING RESULTS

If the result is "**normal**" (within range result) it means that the baby's test results did not show signs of any of the conditions included in the screening.

If the result is "**abnormal**" ("high" or out-of-range result) it means that the baby's test results showed signs of one or more of the conditions included in the newborn screening. This does not always mean that the baby has the condition. It may just mean that more testing is needed.

The child's doctor might recommend that the child get screened again or have more specific tests to diagnose a condition.

What are the risks of not screening? Are there any alternatives to screening?

- Unfortunately affected babies who are not identified through screening will at some stage of life become sick and start to show symptoms of their illness.
- While they may be offered treatment at that stage but unfortunately their growth and development may already have been permanently impaired.
- Babies who receive early and continuous treatment can grow up to enjoy long, healthy and productive life.

Is there any treatment to these disorders?

- The good news is - YES, most of the conditions can be treated if diagnosed early.
- The symptoms and effects can be decreased if they are detected and treated from birth, leading to a reasonably normal and healthy life.

Condition Categories

AMINO ACID DISORDERS

- Phenylketonuria (PKU) or Hyperphenylalaninemia
- Maple Syrup Urine Disease (MSUD)
- Homocystinuria (cystathionine synthase deficiency) or Hypermethioninemia
- Tyrosinemia type 1 and possibly type II or III
- 5-oxoprolinuria (glutathione synthetase deficiency)

FATTY ACID OXIDATION DISORDERS

- Short Chain acyl-CoA dehydrogenase Deficiency (SCAD)
- Isobutyryl-CoA dehydrogenase deficiency (IBCD)
- Glutaric aciduria, type 2 (GAII) or Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Long chain 3 hydroxyacyl-CoA dehydrogenase def. (LCHAD)
- Trifunctional protein deficiency (TFPD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Carnitine palmitoyl transferase deficiency, type 2 (CPTII)
- Carnitine palmitoyl transferase deficiency, type 1A (CPT1A)
- Carnitine/acylcarnitine translocase deficiency (CACT)
- Carnitine uptake defect (CUD)

ENDOCRINE DISORDERS

- Thyroid-stimulating hormone (TSH)
- 17-hydroxyprogesterone (17-OHP)

UREA CYCLE DISORDER

- Citrullinemia
- Argininosuccinic aciduria (ASA)
- Argininemia

PULMONARY CONDITIONS

- Cystic Fibrosis

ORGANIC ACID DISORDERS

- Propionic academia (PA)
- Methylmalonic academia (MMA)
- Malonic aciduria (MA)
- Multiple carboxylase deficiency (MCD)
- 3-hydroxy 3methylglutaryl-CoA lyase deficiency (3HMG)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-methylglutaconic aciduria (3MGA)
- 2-methylbutyryl-CoA dehydrogenase deficiency (2MBD)
- Isovaleric academia, type 1(GAI)
- Beta-ketothiolase deficiency (BKT)

CARBOHYDRATE DISORDERS

- Glucose 6-Phosphate Dehydrogenase Deficiency (G6PD)
- Galactosemia

OTHER METABOLIC DISORDERS

- Biotinidase Deficiency (BIOT)

Disclaimer

These tests should be considered screening tests only and the results of screening tests do not establish a diagnosis but rather serves to identify an infant that may be at risk for a disorder included in the testing panel. The possibility of a disorder should never be ruled out solely based on the screening results.

We trust you found this information useful. If you have any more questions about newborn screening tests, please consult your Neonatologist.

Call our toll-free helpline at **1800 419 1397**

F/PIL/SSC/326/Ver6/Nov23