Neuberg DIAGNOSTICS

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Neu INSIGHTS

New Born Screening

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What is Newborn Screening?

All newborns should have a blood test shortly after birth to screen for metabolic and other inherited disorders. The newborn screening helps to identify babies who may have one of these disorders, and can alert the baby's doctor to the need for further testing and special care.

Why should my baby be screened?

Most babies born are healthy, but there are some babies who may seem fine at birth that have a serious unseen disorder. Early diagnosis and treatment of these conditions has been shown in many cases to reduce morbidity, premature death, mental retardation and other developmental disabilities.

How and when will baby be screened?

A sample of blood is obtained by pricking the baby's heel. This sample is then placed on a special paper which is sent to the designated laboratory for testing. The lab uses this one sample of blood to test for all the required disorders. The sample is usually obtained after 24 hours to 48 hours. Sample should not be taken within first 24 hours of birth.

Conditions where baby needs to be re-tested:

There are 3 main reasons why a repeat screening test may be needed:

(1)There was a problem with the sample,

(2)The test was done prior to 24 hours of age, or

(3)The test result was abnormal.

Generally, if the results of the repeat screening test are also abnormal, there is the need for further treatment or testing. If baby needs to be retested, get it done as soon as possible.

Service Code	Service Name	Methodology	TAT DD:HH:MM	Price
NBS5	New Born Screening 5	ELISA/FEIA	01:10:00	1400.0
NBS5HB	New Born Screening 5 + Hemoglobinopathy	ELISA, FEIA, LC-MS/MS	02:08:00	2600.0
NBS7	New Born Screening 7	ELISA/FEIA	01:10:00	1800.0
NBS7HB	New Born Screening 7 + Hemoglobinopathy	ELISA, FEIA, LC-MS/MS	02:08:00	3000.0
NBS45	New Born Screening 45 + Comprehensive	LC-MS/MS & ELISA/FEIA	02:08:00	4500.0
NBS-AA	New Born Screening - Aminoacids	LC-MS/MS	02:08:00	2500.0
NBS-AC	New Born Screening - Acylcarnitines	LC-MS/MS	02:08:00	2500.0
NBSG6PD	New Born Screening - G6PD	ELISA	01:10:00	280.0
NBSgal	New Born Screening - Total Galactose	FEIA	01:10:00	500.0
NBSTSH	New Born Screening - TSH	ELISA	01:10:00	280.0
NBS17	New Born Screening - 17 - OHP	ELISA	01:10:00	280.0
NBSbio	New Born Screening - Biotinidase	FEIA	01:10:00	500.0
NBSphe	New Born Screening - Phenylalanine	FEIA	01:10:00	500.0
NBSIRT	New Born Screening - Immuno reactive trypsinogen (IRT)	ELISA	01:10:00	280.0
NBSHB	New Born Screening-Hemoglobinopathy	LC-MS/MS	02:08:00	1000.0
T2549	Congenital Adrenal Hyperplasia (Seq+ MLPA)	Sequencing + MLPA	24:08:20	12000.0
T4595	ORION Focus (Clinical Exome/ Mendeliome/Panel testing)	Next Generation Sequencing	36:02:00	16000.0

New Born Screening 5 (NBS5)

G6PD, Total Galactose, Thyroid stimulating hormone (TSH), 17- hydroxyprogesterone (17-OHP), Biotinidase

New Born Screening 7 (NBS7)

G6PD, Total Galactose, Thyroid stimulating hormone (TSH), 17-hydroxyprogesterone (17-OHP), Immuno reactive trypsinogen (IRT), Phenylalanine, Biotinidase

New Born Screening 45 + Comprehensive (NBS45)

Acylcarnitines, Aminoacids, New Born Screening 7, Hemoglobinopathy

Newborn screening panel currently includes the following disorders:

01 Congenital Hypothyroidism (CH)

CH results from lack or absence of thyroid hormone, which is essential for growth of the brain and the body. If the disorder is not detected and hormone replacement is not initiated within(4) weeks, the baby's physical growth will be stunted and she/he may suffer from mental retardation.

02 Congenital Adrenal Hyperplasia (CAH)

CAH is an endocrine disorder that causes severe salt loss, dehydration and abnormally high levels of male sex hormones in both boys and girls. If not detected and treated early, babies may die within 7-14 days.

03 Galactosemia (GAL)

GAL is a condition in which the body is unable to process galactose, the sugar present in milk. Accumulation of excessive galactose in the body can cause many problems, including liver damage, brain damage and cataracts.

04 Phenylketonuria (PKU)

PKU is a metabolic disorder in which the body cannot properly use one of the building blocks of protein called phenylalanine. Excessive accumulation of phenylalanine in the body causes brain damage.

05 Glucose-6- Phosphate Dehydrogenase Deficiency (G6PD Def)

G6PD deficiency is a condition where the body lacks the enzyme called G6PD. Babies with this deficiency may have hemolytic anemia resulting from exposure to certain drugs, foods and chemicals.

06 Biotin deficiency

Biotinidase deficiency is a genetic disorder that is found in a few babies born each year. When a baby has biotinidase deficiency, he or she cannot use biotin, a vitamin that is found in foods, including breast milk and infant formula. Without biotin, the growth and development of the baby is affected.

07 Cystic fibrosis-Immunoreactive Trypsinogen (IRT)

IRT is a protein made by the pancreas. IRT can be elevated for a number of reasons, including cystic fibrosis (CF). If the IRT was elevated, the baby's blood is tested for 39 most common variants (changes) to the gene causing CF (CFTR gene).

08 Amino Acid/Urea Cycle Disorders

These disorders are inherited as autosomal recessive defects of amino acid metabolism. Each amino acid disorder is associated with a specific enzyme defect. Affected infants cannot properly metabolize certain amino acids, resulting in elevated levels of the amino acid or metabolites in body fluids. Clinical findings may include poor feeding, vomiting, lethargy or irritability, seizures, coma, respiratory distress and liver damage.

09 Fatty acid oxidation-Acylcarnitines

Fatty acid oxidation (FAO) disorders are autosomal recessive inherited metabolic conditions. Each FAO disorder is associated with a specific enzyme defect in the fatty acid metabolic pathway, and affects utilization of dietary and stored fats. Clinical findings may include lethargy, hypotonia, failure to thrive, persistent vomiting and hepatomegaly, rhabdomyolysis, and Reye syndrome-like episodes.Significant disability may result from prolonged episodes of hypoglycemia.

10 Organic Acid Disorders-Acylcarnitines

Organic acid disorders are autosomal recessive inherited metabolic conditions. Each organic acid disorder is associated with a specific enzyme deficiency that causes the accumulation of organic acids in blood and urine. Usually infants with organic acid disorders appear normal at birth, but may develop vomiting, poor feeding, hypoglycemia, seizures, hypotonia and lethargy progressing to coma.There is a significant risk of death in infancy due to organic acid disorders; early diagnosis and treatment can greatly improve disease outcome.

Following is a list of the FAO disorders that may be detected:

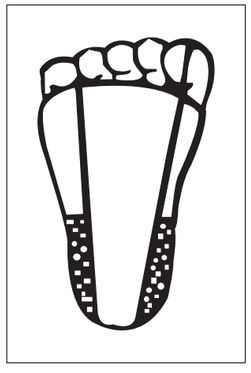
- Short chain acyl-CoA dehydrogenase deficiency (SCADD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Carnitine palmitoyl transferase deficiency type 2 (CPT 2)
- Carnitine/acylcarnitine translocase deficiency (CACT)
- Carnitine palmitoyl transferase deficiency type 1 (CPT 1 or CPT 1A)
- Glutaric aciduria type 2 (GA 2)/Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Carnitine uptake defect (CUD)
- Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)
- Isobutyryl-CoA dehydrogenase deficiency (IBCD)

Following is a list of the organic acid disorders that may be detected:

- Propionic acidemia (PA)
- Methylmalonic acidemia (MMA)
- Isovaleric acidemia (IVA)
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (3HMG)
- 3-methylglutaconic aciduria (3MGA)
- 3-methylcrotonyl CoA carboxylase deficiency (3-MCC)
- Glutaric acidemia, type 1 (GA 1)
- 2-methylbutyryl-CoA dehydrogenase deficiency (2MBCD)
- Malonic aciduria (MA)
- Betaketothiolase deficiency (BKT)
- Multiple carboxylase deficiency (MCD)

Instructions

- ✓ Please fill out the form completely. Use ball point pen.
- ✓ Store specimen card in a cool dry place.
- ✓ Do not handle filter paper portion. Skin oils prevent saturation.



Right — **ACCEPTABLE**

Circle filled and evenly saturated

Wrong — **UNACCEPTABLE**

Layering

Insufficient, Multiple applications

Serum ring present

Collect Sample From Shaded Area

1. Sterilize and dry the skin. Puncture heel with a sterile lancet
2. Allow large blood droplet to form (without excessive squeezing)
3. Touch filter paper to blood; allow blood to soak through completely in each circle. Total saturation of the circles must be evident when the paper is viewed on both sides, but do not apply blood to both sides.
4. Allow blood spots to air-dry throughly for 3-4 hours at room temperature. Keep away from direct sunlight and heat. Never superimpose one wet filter paper on another before thorough drying.
5. Submit specimen to the laboratory within 24 hours of collection.

Hemoglobin disorders- Hemoglobinopathies

1

This group of disorders is characterized by abnormal hemoglobin production

2

Depending on the type, the severity may vary.

3

Infants with diseases like sickle cell disease, sickle hemoglobin C disease, sickle beta thalassemia and beta thalassemia major are very susceptible to anemia, life-treatening infections and other complications.

4

Infants diagnosed with alpha or beta thalassemia may experience severe anemia requiring blood transfusions

5

Hemoglobin traits and other less serious disorders produce adequate amounts of functional hemoglobin and do not usually require treatment.

Knock out medical emergencies, before it's too late!

HEALTH ko aasani se na lo, TEST aasani se karo

- Blood Tests
- Health Packages
- ICMR Approved COVID Tests

" Get your blood test done Anywhere, Anytime... Neuberg hain na "

Notes :

PARTNERS IN HEALTH

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