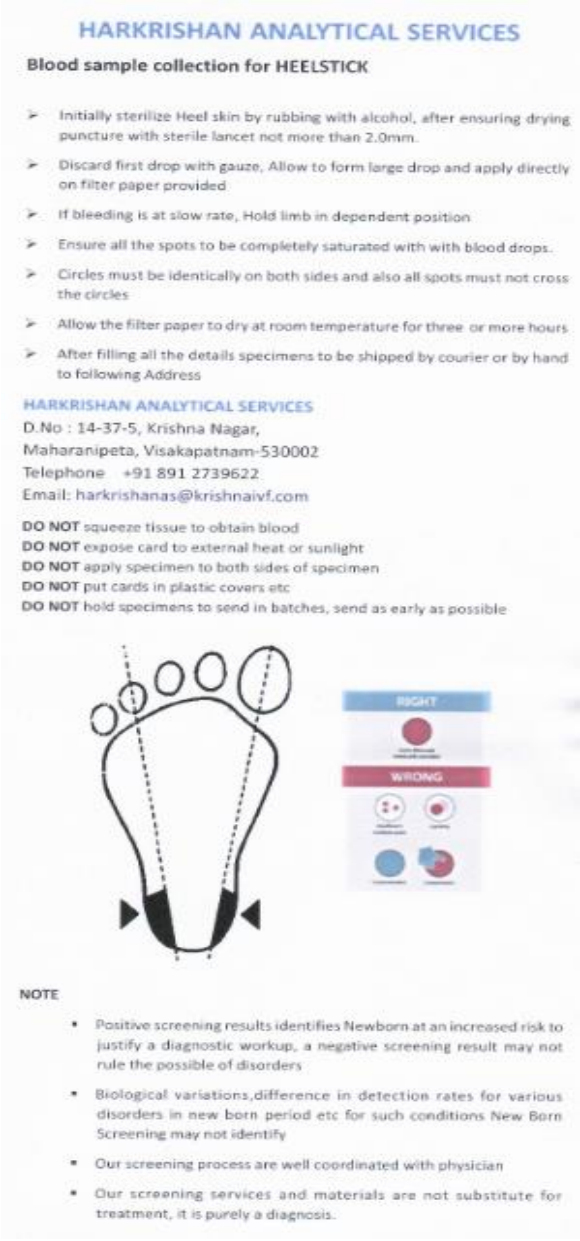
HARKRISHAN ANALYTICAL SERVICES

About Newborn Screening

Newborn screening is the practice of testing every newborn for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth.

With screening, doctors often can tell whether newborns have certain conditions that eventually could cause problems. Although these conditions are rare and most babies are given a clean bill of health, early diagnosis and proper treatment sometimes can make the difference between lifelong impairment and healthy development.

BLOOD SAMPLE collection procedure:



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Blood sample collection for HEELSTICK

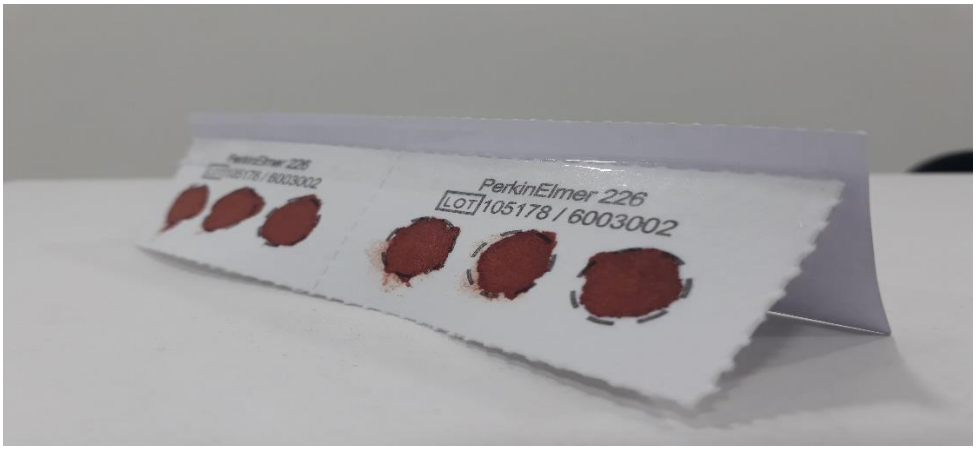
- Initially sterilize Heel skin by rubbing with alcohol, after ensuring drying puncture with sterile lancet not more than 2.0mm.
- Discard first drop with gauze, Allow to form large drop and apply directly on filter paper provided
- If bleeding is at slow rate, Hold limb in dependent position
- Ensure all the spots to be completely saturated with with blood drops.
- Circles must be identically on both sides and also all spots must not cross the circles
- Allow the filter paper to dry at room temperature for three or more hours
- After filling all the details specimens to be shipped by courier or by hand to following Address

HARKRISHAN ANALYTICAL SERVICES
D.No : 14-37-5, Krishna Nagar,
Maharanipeta, Visakapatnam-530002
Telephone +91 891 2739622
Email: harkrishanas@krishnaivf.com

DO NOT squeeze tissue to obtain blood
DO NOT expose card to external heat or sunlight
DO NOT apply specimen to both sides of specimen
DO NOT put cards in plastic covers etc
DO NOT hold specimens to send in batches, send as early as possible

NOTE

- Positive screening results identifies Newborn at an increased risk to justify a diagnostic workup, a negative screening result may not rule the possible of disorders
- Biological variations,difference in detection rates for various disorders in new born period etc for such conditions New Born Screening may not identify
- Our screening process are well coordinated with physician
- Our screening services and materials are not substitute for treatment, it is purely a diagnosis.



TESTS OFFERED:

Tests carried out through FLUORESCENCE AND LUMINESCENCE READER

The disorders listed here are those usually included in newborn screening programs:

- ✓ Phenylketonuria (PKU)
- ✓ Congenital hypothyroidism
- ✓ Galactosemia
- ✓ Biotinidase deficiency
- ✓ Cystic fibrosis (CF)

PHENYLKETONURIA (PKU)

- ✓ Phenylketonuria (PKU) is a metabolic disorder
- ✓ Babies with PKU are missing an enzyme that's needed to process phenylalanine.
- ✓ Phenylalanine is an amino acid necessary for normal growth in kids and for normal protein use.
- ✓ Amino acids are the "building blocks" for the proteins that make up our muscles and other important tissues.
- ✓ If too much phenylalanine builds up, it damages brain tissue and can cause developmental delay.

- ❖ When PKU is detected early, feeding an infant a special formula low in phenylalanine can prevent intellectual disability.
- ❖ A low-phenylalanine diet is needed throughout childhood and adolescence and sometimes into adult life.

CONGENITAL HYPOTHYROIDISM

- ✓ The thyroid, a gland at the front of the neck, releases special chemicals called **hormones** that control metabolism and growth.
- ✓ These hormones control how fast the body uses up energy and are also key factors in helping kids grow.
- ✓ Babies with congenital hypothryoidism don't have enough thyroid hormone.
- ✓ The condition can cause slow growth and brain development.

- ❖ If congenital hypothryoidism is detected early, a baby can be treated with oral doses of thyroid hormone.

GALACTOSEMIA

- ✓ Babies with galactosemia lack the enzyme that converts galactose (one of two sugars found in lactose) into glucose, the major source of energy for the body's cells.
- ✓ For a baby with galactosemia, milk (including breast milk) and other dairy products must be removed from the diet.
- ✓ Otherwise, galactose can build up in the body and damage cells and organs, leading to blindness, severe intellectual disability, growth deficiency, and even death.
- ✓ Many less severe forms of galactosemia may not require any treatment.

BIOTINIDASE DEFICIENCY

- ✓ Babies with biotinidase deficiency don't have enough biotinidase, an enzyme that recycles biotin (B vitamin) in the body.
- ✓ Biotinidase deficiency may cause seizures, poor muscle control, problems with the immune system, hearing loss, intellectual disability, coma, and even death.

- ❖ If biotinidase deficiency is detected quickly, problems can be prevented by giving the baby extra biotin.

CYSTIC FIBROSIS (CF)

- ✓ It is a genetic disorder that mainly affects the lungs and Digestive system and makes kids who have it more vulnerable to lung infections.
- ❖ There is no known cure for cystic fibrosis — treatment involves trying to prevent serious lung infections (sometimes with antibiotics) and providing adequate nutrition.
- ❖ Early detection may help doctors reduce the problems associated with cystic fibrosis.

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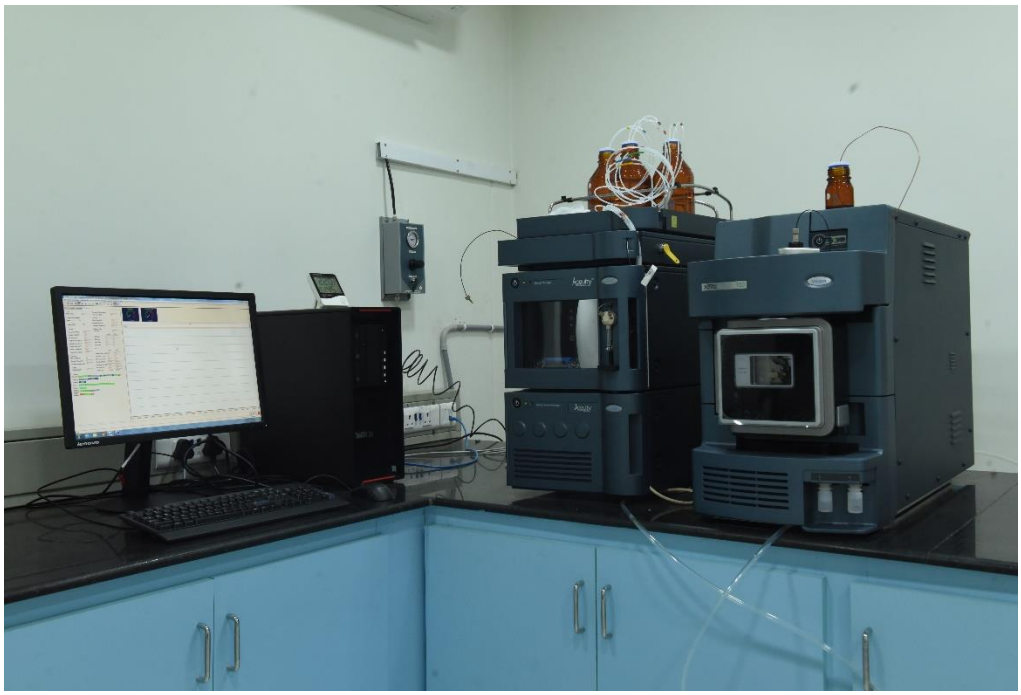
TESTS CARRIED OUT BY TANDEM MASS SPECTROSCOPY:



NEW BORN INBORN METABOLIC DISORDERS:

- AMINO ACID DISORDERS
- ORGANIC ACID DISORDERS
- FATTY ACID DISORDERS

Amino Acid Disorders

- Argininemia (ARG1 Deficiency)
- Argininosuccinic Aciduria (ASL Deficiency)
- 5-Oxoprolinuria1
- Carbamoylphosphate Synthetase Deficiency 1 (CPS1 Deficiency)
- Citrullinemia I (ASS Deficiency)
- Citrullinemia II
- Homocystinuria
- Hypermethioninemia
- Hyperammonemia, Hyperornithinemia, Homocitrullinemia Syndrome1
- Hyperornithinemia with Gyral Atrophy 1
- Maple Syrup Urine Disease
- N-acetyl Glutamate Synthetase Deficiency (NAGS Deficiency)
- Phenylketonuria
- Classical/Hyperphenylalaninemia
- Defects of Biopterin Cofactor Biosyntheis
- Defects of Biopterin cofactor regeneration
- Tyrosinemia (detected by SUAC)
  - Transient Neonatal Tyrosinemia
  - Tyrosinemia Type I
  - Tyrosinemia Type II
  - Tyrosimenia Type III
- Ornitine transcarbamoylase deficiency (OTC)

Organic Acid Disorders

- 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
- Glutaric Acidemia Type I
- Isobutyryl-CoA Dehydrogenase Deficiency
- Isovaleric Acidemia
- 2-Methylbutryl-CoA Dehydrogenase Deficiency
- 3-Methylcrotonyl-CoA Carboxylase Deficiency
- 3-Methylglutaconyl-CoA Hydratase Deficiency
- Methylmalonic Acidemias
- Methylmalonyl-CoA Mutase Deficiency
- Some Adenosylcobalamin Synthesis Defects
- Maternal Vitamin B12 Deficiency
- Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
- Propionic Acidemia
- Multiple-CoA Carboxylase Deficiency
- Malonic Aciduria

Fatty Acid Oxidation Disorders

- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
- Medium/Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency
- 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency (LCHAD)
- Short Chain Acyl-CoA Dehydrogenase Deficiency
- Medium Chain Ketoacyl-CoA Thiolase deficiency
- Carnitine uptake deficiency
- Carnitine/Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type II
- Carnitine Palmitoyl Transferase Ia deficiency
- Carnitine Palmitoyl Transferase Ib deficiency
- 2,4-Dienoyl-CoA Reductase Deficiency1
- Glutaric Acidemia type II
- Trifunctional Protein Deficiency