SYNLAB

Routine Newborn Screening

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Testing Infants and Elder Children for Metabolic Defects

Metabolic birth defects can cause physical problems, mental retardation and, in some cases, death. It is best for the baby and the family if these conditions are detected and treated early. Synlab offers an expanded metabolic screening and can confirm or exclude disorders of the newborn infants (32 disorders) and in elder children (27 disorders).

The development of a new screening technique known as **tandem mass spectrometry** (often abbreviated as MS/MS) can detect the blood components that are elevated in certain disorders, and is capable of screening for inherited metabolic disorders with a single test such as sickle cell anaemia, G6PD, cystic fibrosis, hypothyroidism, biotinidase deficiency, galactosaemia, adreno genital syndrome (AGS), phenylketonuria (PKU), maple syrup urine disease (MSUD), homocystinuria, tyrosinaemia type I, MCAD deficiency, propionic acidaemia, glutaric aciduria type I, isovalerianic acidaemia and methylmalonic acidaemia and other 15 more rare disorders. Furthermore, the used method also detects aberrant amino acid concentrations (including Tyrosinemia Type I).

How is the screening done?

For the screening, capillary blood drops are placed on a special Filter Card, which will be available from any Synlab office. The time of withdrawal is 3rd to 6th day of life.

The newborn screen is most accurate if the infant's blood is taken after the first 36 hours of their life. If it was done before 36 hours of age, a second sample needs to be taken if the infant is 1 to 2 weeks old.

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Order: Newborn screening

Precautions: Do not touch the marked circular area(filter card).
Do not use capillary or venous blood collection (no EDTA or other additives!). Disinfection of skin (heel).

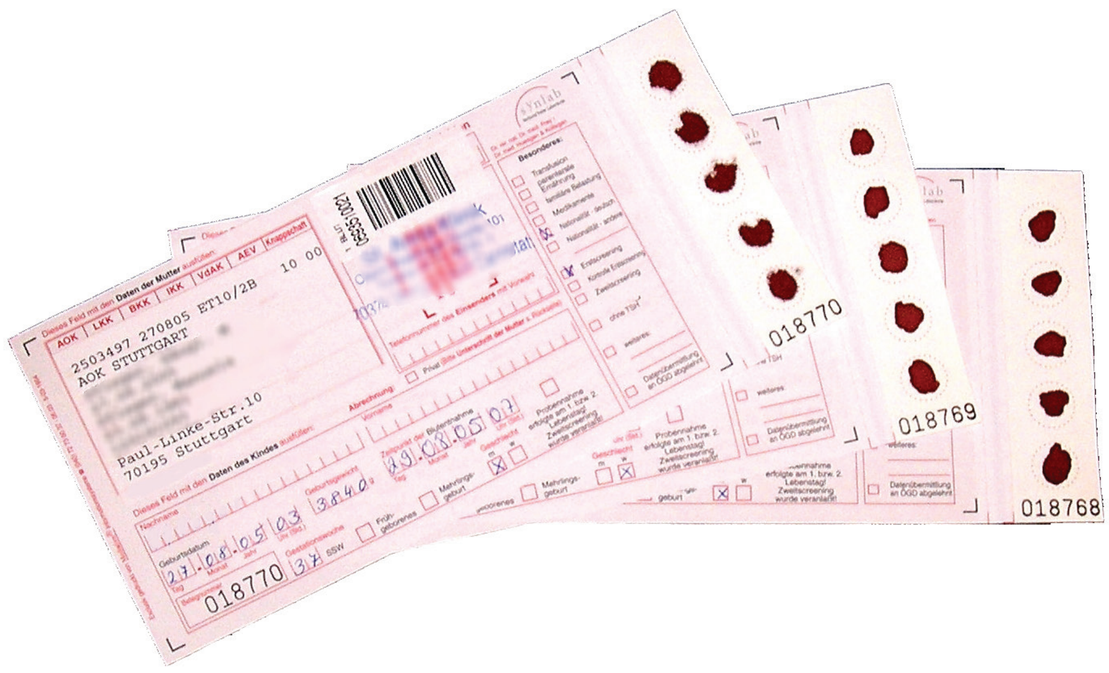
Blood collection: Prick the heel with a blood lancet.
Fill each of the marked circular areas completely, let it dry for 2-4 hours and keep it in a cool light-protected place.
Complete the filter card with all required patient data.

Preanalytic: Send the filter card on the same day.
Keep it separate and do not stick it to other filters or leave it in putative blood contaminated areas, such as labora-tory desks.

TAT: will be 10-15 working days from day sample is received in the lab

Result: unregular, positive or suspicious single parameters the lab will immediately forward to the customer
Note: Exclusions which are not recognized by the screening are lysosomal storage disorders, mito-chondrial disorders as well as organic acidurias.

Price: on request



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What the screening can exclude ?

Sickle Cell Anaemia G6PD deficiency

Hypothyreoidism TSH	1:4000
Adrenogentital Syndrome	1:11000
Galactosemia	1:60000
Biotinidase-Deficiency	1:82000
SCID (Severe Combined Immunodeficiency)	1:100000
PBGS (Tyrosinemia Type 1)	<1:200000

Disorders of Amino Acid Metabolism and Urea Cycle

Phenylketonuria and Hyperphenylalaninemia	1:5.500
Hypertyrosinemia Type I and II	1:137000
Maple Sirup Urine Disease (MSUD)	1:164000
Hypermethioninaemia	1:205000
Ornithine Transferase Deficiency	
Arginase Deficiency	
Argininosuccinate Synthase Deficiency	
Argininosuccinate Lyase Deficiency	

Organic Acidurias

Glutaric aciduria Type I	1:102000
Methylmalonic aciduria	1:117000
Propionic acidaemia	1:117000
Isovalerianic acidaemia	1:91000
Isolated 3-Methylcrotonyl-CoA Carboxyl.--Deficien.	
3-Methylglutaconyl-CoA Hydratase Deficiency	
3-Hydroxy-3-Methyl-Glutaric-CoA Lyase Deficiency	
Beta Ketothiolase Deficiency	
Pyroglutamate Acidaemia	

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Beta Oxidation

SCAD Deficiency	
MCAD Defiency	1:9000
LCAD Deficiency	
(VLCAD Deficiency, exceptions)	1:137000
Long Chain 3-Hydroxy-Fatty Acids (LCHAD-Defi¬ciency)	1:273000
Glutaric Aciduria Type II	
2.4-Dienoyl-CoA Deficiency	

Transport of Fatty Acids (Carnitin Cycle)

Carnitin Transporter Deficiency

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Description of some diseases & conditions:

Sickle Cell Anaemia:

This inherited blood disease causes bouts of pain; damage to vital organs such as the lungs, kidneys and brain; and sometimes serious infections and death in childhood.

PKU (phenylketonuria):

Babies with this disorder cannot pro¬cess a substance called phenylalanine that is found in almost all food. Without treatment, phenylalanine builds up in the blood-stream and causes brain damage and mental retardation. When PKU is detected early, mental retardation can be prevented by feeding the child a special diet.

Hypothyroidism:

Babies with this disorder have a hormone deficiency that slows growth and brain development. If it is detected in time, a baby can be treated with oral doses of the hormone to permit normal development.

Glucose-6-Phosphate Dehydrogenase (G6PD):

Babies with G6PD deficiency appear normal at birth. They may experience neonatal jaundice and hemolysis that can be so serious as to cause neurologic damage or even death.
Exposure to certain antimalarial drugs and sulfonamides, infection stress environmental agents (e.g., moth balls), and eating certain foods (e.g., fava beans), impact the baby's ability to handle oxidative reactions, leading to acute hemolytic anemia.

Galactosemia:

Babies with this disorder cannot convert galactose, a sugar present in milk, into glucose, a sugar that the body uses as an energy source. Galactosemia can cause death in infancy, or blindness and mental retardation.

Congenital adrenal hyperplasia (CAH):

Babies who have this group of disorders are deficient in certain hormones. CAH affects genital development and, in severe cases, can disturb kidney function and cause death.

Biotinidase deficiency:

Babies with this condition don't have enough biotinidase,

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an enzyme that recycles biotin (one of the B vitamins) in the body. The deficiency may cause seizures, poor muscle control, immune system impairment, hearing loss, mental retardation, coma, and even death.

Homocystinuria:
This metabolic disorder results from a deficiency of one of several enzymes for normal development. If untreated, it can lead to dislocated lenses of the eyes, mental retardation, skeletal abnormalities, and abnormal blood clotting.

Tyrosinemia:
Babies with this disorder have trouble processing the amino acid tyrosine. If it accumulates in the body, it can cause mild retardation, language skill difficulties, liver problems, and even death from liver failure.

Severe Combined Immuno Deficiency (SCID):
Severe combined immunodeficiency (SCID) is a group of rare disorders caused by mutations in different genes involved in the development and function of infection-fighting immune cells. Infants with SCID appear healthy at birth but are highly susceptible to severe infections. The condition is fatal, usually within the first year or two of life, unless infants receive immune-restoring treatments, or enzyme therapy. More than 80 percent of SCID infants do not have a family history of the condition. However, development of a newborn screening test has made it possible to detect SCID before symptoms appear, helping ensure that affected infants receive life-saving treatments.

Organic Acidemia Disorders (GA-1, PA, MMA, IVA, 3-MCC, MAD, HMG):
Organic Acidemias (OA) are a class of inherited metabolic disorders that lead to accumulation of organic acids in biological fluids (blood and urine). Clinical symptoms of OA disorders may include vomiting, metabolic acidosis, ketosis, dehydration or coma, hyperammonemia, lactic acidosis, hypoglycemia, failure to thrive, hypotonia, global developmental delay, sepsis, and hematological disorders.

Maple syrup urine disease (MSUD):
Babies with MSUD are missing an enzyme needed to process three amino acids that are essential for the body's normal growth. These babies usually have little appetite and are extremely irritable. If not detected and treated early, MSUD can cause mental retardation, physical disability, and even death.

Fatty Acid Oxidation Disorders (MCAD, VLCAD, LCHAD, CPT1, CPT2, CAT):
Fatty Acid Oxidation Disorders (FOD) are a class of inborn errors of metabolism where there is an enzyme defect in the fatty acid metabolic pathway (use of dietary and stored fat). Clinical symptoms of FOD disorders include hypotonia, lethargy, and vomiting; the hypoglycemia can lead to coma, encephalopathy, hepatic failure, or death.

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