



Answers to Frequently Asked Questions

Q. What is the Newborn Screening (NBS) Test?

A. It tests babies for serious disorders and is usually performed when your baby is 24 - 72 hours old. Ideally, the specimen should be sent to the laboratory by the fastest way possible to prevent delay.

Q. Why is the test done?

A. The test is done to find out if your baby has a disease or condition for which early treatment can prevent death, mental retardation, or physical disability.

Q. How is the test performed?

A. The test is performed by pricking a baby's heel and putting a few drops of blood on a special filter paper. The paper is allowed to dry and then sent to the newborn screening laboratory where several different tests will be performed.

The heel prick feels no worse than being stuck by a pin. Problems from the prick, such as infection of the heel, are very rare.

Q. But we have no family history of these disorders . . .

A. Parents who have no family history of problems and/or who have already had healthy children can still have children with these disorders. In fact, most children with these disorders come from families with no previous history of the condition.

Genes for these diseases can be passed along through generations of healthy people without anyone ever knowing about them. These "carriers" are healthy because the normal gene in the pair (genes come in pairs—one from each parent) is working, making up for the flawed gene. But when two people who coincidentally carry the same flawed gene get together, their risk of having an affected child is 25% for each pregnancy. This pattern, called recessive inheritance,

explains how most metabolic diseases appear unexpectedly.

Q. But my baby looks healthy . . .

A. Most babies with disorders look and act normal and seem perfectly healthy. The newborn screening test helps your doctor catch a problem with your baby before it makes him or her sick. Most babies that are diagnosed and treated early do well. The earlier the disorder is detected, the higher the chance of having a good prognosis.

Q. What is a Retest?

A. If the result of your child's test is abnormal, a repeat test or a "retest" is usually required. A request for a retest does not necessarily mean your child has a disorder, but it is possible. If you are asked for a retest, it is important that you take your baby for the retest as soon as possible.

Q. How will I know the results of my baby's test?

A. Generally, parents and doctors are notified of the test results. However, it is a good idea to call your doctor or NeoGen Labs and request the results if you have not received it within 2 weeks of the test. This is important to ensure that your child's test results have not been lost or misplaced.

If your child's test shows an abnormal result, you will be notified immediately and given directions about what to do next. Follow the directions of your doctor very carefully. If your child's test is abnormal, additional tests are usually necessary to verify if your child has the disorder. It is important that you advise your doctor if you move or change phone numbers soon after your baby is born in case there is a problem with your baby's test.

Q. What does a positive result indicate?

A. Parents should not be alarmed by



positive results as the screening gives only preliminary information, albeit with a high degree of accuracy. It should be followed by a precise confirmatory test.

Q. What exactly are Inborn Errors of Metabolism (IEM)?

A. These are disorders caused by the accumulation of chemicals produced naturally in the body to abnormal levels. The symptoms manifest themselves in a variety of ways; slow physical development or mental retardation. In some cases, they could result in death. Unfortunately, most infants with these disorders show no signs of these conditions. If these conditions are detected at birth by NBS, the child can lead a normal, healthy life.

Q. How can these disorders be cured?

A. These conditions have no cures and are inherited. The symptoms and effects can be mitigated if they are detected and treated early, leading to a normal and healthy life.

Q. Can Newborn Screening be performed on older children or adults?

A. Yes. Older children or adults also can be screened.

Q. Will Newborn Screening by NeoGen Labs detect all conditions?

A. The analyses conducted produce results that can be used by qualified physicians in the diagnosis of IEMs. Evidence of these conditions will be detected in the vast majority of affected individuals; however, due to genetic variability, age of the patient at the time of specimen collection, quality of the specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.

Q. When will NeoGen Labs start offering Newborn Screening?

A. NeoGen Labs expects to start screening commercially from July 1, 2007. From April 15, 2007 until June 30, 2007, NeoGen Labs will be conducting validation testing.

Q. What disorders can Newborn Screening identify?

NeoGen Labs tests babies for over 42 metabolic disorders from a dried blood spot. Some of these are described below. These disorders are present at birth, rare, and often serious. Some are passed on from parents while others are caused by a chemical imbalance. Some are life-threatening while others may slow down physical development or cause mental retardation or other problems. These disorders can affect a child early in life, often within the first few days or weeks of life. This is why it is important to identify babies with these disorders as early as possible.

- **Maple Syrup Urine Disease (MSUD)** results when the baby's body does not break down parts of a food protein causing the urine to smell like maple syrup. Treatment with a special diet can prevent mental retardation and other complications.
- **Galactosemia (ga-LAK-toe-see-me-a)** results when milk sugar (galactose) is not broken down due to the lack of a chemical in the body. A diet low in galactose can prevent irreversible damage and other complications.
- **Phenylketonuria (FEN-nil-KEE-tone u-ree-ah)** is also called PKU and results when a part of a food protein (phenylalanine) is not broken down by the baby's body. Brain damage that would normally result can be prevented by a special diet low in phenylalanine.
- **Tyrosinemia I (TY-ro-SIN-e-me-ah)** results when another part of a food protein (tyrosine) cannot be broken down and used properly in the baby's body. Some forms of this disorder can result in liver and brain damage, and may be life-threatening.
- **Homocystinuria (HO-mo-SIS-tin-u-ree-ah)** results from the absence of a

chemical in the liver. A special diet can help prevent mental retardation, body changes and life-threatening complications.

- **Hypothyroidism (HI pO-THI-royd-ism)** results when the baby's body does not produce enough of a hormone (substance) called thyroxin. Treatment with thyroxin tablets helps prevent mental and growth retardation.

- **Congenital Adrenal Hyperplasia (CAH) (con-GEN-I-tle ah-DRE-nal HY-per-PLA-se-a)** results when the baby's body does not produce enough of a substance (hormone) called cortisol. Treatment with hormone medications can prevent low blood sugar, salt loss, poor growth and abnormal body changes.

- **Fatty Acid Oxidation Disorders**

The body usually gets energy from sugars and fats. The sugar is used first but when the sugar is all used up, the body must use fats. In this group of disorders, the body cannot use fats because of the lack of one of several enzymes. The disorders in this group do not have common names. They are usually described by the length of the fatty acid that cannot be used. The most common of these is,

- **Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD).** Because they cannot use stored fat for energy, babies with these disorders may develop seizures, coma and life threatening complications when fasting (no food eaten for longer than 4 hours). Treatment includes making sure the baby eats regularly and avoiding fasting. A special diet and medications may also be used.

Other fatty oxidation disorders screened for are,

- **Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD)**
- **Trifunctional Protein Deficiency (TFP Deficiency)**
- **Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)**
- **Carnitine Uptake Defect (CUD)**

- **Organic Acidemias**

The body cannot use the branched chain amino acids from the protein in food properly because of the lack of one of several enzymes. The breakdown products of these amino acids are organic acids. The organic acids build up to dangerous levels in the blood damaging the nervous system. Babies with some of these disorders can become very sick very fast. Symptoms can sometimes be lessened with special diets (low in protein) and medications.

Most of these disorders do not have common names and are described by the name of the organic acid found in the urine. The organic acidemias screened for include,

- **Isovaleric Acidemia (IVA)**
- **Glutaric Acidemia Type I (GA-1)**
- **3-Hydroxy-3Methylglutaryl-CoA Lyase Deficiency (HMG)**
- **Multiple CoA Carboxylase Deficiency (MCD)**
- **Methylmalonyl-CoA Mutase Deficiency (MUT)**
- **Methylmalonyl Adenosyl-Cobalamine Synthesis Defects (Cbl A & B)**
- **3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)**
- **Propionic Acidemia (PROP)**
- **Beta-Ketothiolase Deficiency (BKT)**

- **Urea Cycle (Amino Acid) Disorders**

The body cannot dispose of nitrogen properly. The body usually disposes of nitrogen by changing it into a substance



called urea, which leaves the body in the urine. Because of the lack of one of several enzymes, the body cannot make urea effectively and the nitrogen builds up as ammonia in the blood. This makes the baby very sick very fast. These disorders cause seizures, poor muscle tone, breathing problems, and coma. Death will result if the baby is not treated. Symptoms can be lessened with special diet and medications that

help the baby to get rid of ammonia in other ways. Screening for this group of disorders is very new. The urea cycle disorders screened by NeoGen Labs include,

- Argininosuccinic Aciduria (ASA)
- Citrullinemia (CIT)

The FAQ was created from information from multiple sources that include **Pediatrix Screening, Inc.** (www.pediatrix.com), **Save Babies Through Screening Foundation** (www.savebabies.org) and **Virginia Dept. of Health** (www.vahealth.org).