

Why Retest for Metabolic Diseases?



State law requires that all babies have the newborn screening (NBS) test before leaving the hospital. A few drops of blood were taken from your baby's heel and tested for certain diseases.

Why should my baby be retested?

Your baby's initial test showed a "positive" result for a metabolic disease. More specific testing is needed to find out if your baby actually has this disease. Not all babies with an initial "positive" result have a disease. In fact, metabolic diseases are rare, and *babies often have normal results after more testing*. However, babies can look healthy at birth and still have one of these diseases. If a disease is not identified and treated quickly, mental retardation and other serious health problems can occur.

What is metabolism?

Metabolism is the process of changing food into energy for the body. Food such as proteins, fats, and carbohydrates are changed into smaller parts. These smaller parts include *amino acids* from proteins, *fatty acids* from fats, and *organic acids* from proteins, fats and sugars. Special chemicals called enzymes are needed to complete this process.

What are metabolic diseases?

Metabolic diseases are a group of conditions in which the body is unable to break down certain parts of some foods. Most are caused by enzymes that are missing or not working correctly. Without these enzymes, metabolic products cannot be used properly, and build up in the body. Large amounts of these products can be harmful to the body and brain. To know if your baby has a metabolic disease, specific tests can be done to look at the levels of these products in your baby's blood.

Metabolic diseases are grouped into three main categories:

- Amino Acid Disorders
- Fatty Acid Oxidation Disorders
- Organic Acid Disorders

Are metabolic diseases common?

About one in 4500 babies in California (or about 120 babies per year) is born with one of these metabolic diseases.

What is the treatment?

If a disease is diagnosed, specially trained staff at a metabolic center will talk with you about a treatment plan for your baby. Treatment may include special diet, medication and regular exams.

What happens now?

The doctor will advise you as to what steps need to be taken. The NBS Program strongly recommends that newborns with positive screening results be referred to a California Children's Services (CCS) approved Metabolic Special Care Center (SCC) or CCS approved pediatric metabolic specialist for a diagnostic evaluation. More testing may be needed.



Who will pay for the diagnostic evaluation and treatment if needed?

All newborns referred to a CCS-approved SCC by the NBS Program are eligible for a diagnostic evaluation through the SCC regardless of income. You will be asked to complete an application form to determine eligibility for CCS payment. Most health insurance and HMOs provide at least some coverage for the diagnostic evaluation and any necessary treatment. If your baby has health insurance, the metabolic center will bill your health insurance company or HMO for the services. Infants who have Medi-Cal full scope, no share of cost, or Healthy Families subscribers will be authorized by CCS for diagnostic and treatment services and you will not need to pay anything for services including the laboratory tests. If you do not have health insurance or if your insurance only covers partial payment, your infant may be eligible for the CCS Program. If part of the diagnostic evaluation includes lab tests done through the state Metabolic Reference Lab at Quest Diagnostics, the lab will bill your insurance carrier, and you will not have to pay any out-of-pocket costs. To learn more about CCS, visit our website at: www.dhs.ca.gov

Is There Anything Else I Should Do?

Call your doctor **right away** if your baby:

- ◆ is not eating well
- ◆ is sleeping excessively
- ◆ has diarrhea or vomiting
- ◆ has abnormal odor or color of urine
- ◆ has a seizure or signs of coma

California
Department of
Health Services



Newborn Screening Program (NBS)
Genetic Disease Branch
www.dhs.ca.gov/gdb