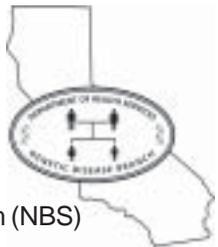


# Why Retest for Metabolic Diseases?



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.

California  
Department of  
Health Services



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

## Why should my baby be retested?

Your baby's initial test showed a "positive" result for a metabolic disease. However, 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*. 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 may have 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 are the symptoms?

Signs of metabolic diseases may include poor feeding and slow growth, vomiting, diarrhea, excessive sleeping, abnormal odor or color of urine, seizures and coma. Tell your baby's doctor immediately if you notice any of these signs in your baby. If your baby is ill and the doctor cannot be reached, take your baby to the emergency room for evaluation.

## 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 to expect after getting a positive result

- ◆ Your baby's doctor will be notified about the positive test result by a NBS Coordinator from one of the Newborn Screening Area Service Centers.
- ◆ Your doctor or a NBS Coordinator will contact you to discuss and answer questions about the follow-up process.
- ◆ You will be able to talk about the result with your baby's doctor. You may be advised to put your child on a special diet and/or medications prior to the retest.
- ◆ A metabolic doctor in your area may work with your baby's doctor to find out if your baby truly has a metabolic disease. The doctor(s) may order more tests and/or schedule a physical exam for your baby.
- ◆ Your baby's doctors will review and discuss the results of the tests and/or exams with you.
- ◆ It is helpful to make a list of questions to ask your doctor before each visit.

## For more information, check our website.

[www.dhs.ca.gov/gdb](http://www.dhs.ca.gov/gdb), then click on Newborn Screening.

## The MS/MS Screen

A few drops of blood were taken from your baby's heel shortly after birth and tested through the mandatory California Newborn Screening Program. You consented to having the same blood sample tested through the Tandem Mass Spectrometry (MS/MS) Research Project at no extra cost. After your baby's blood specimen was screened for the mandatory tests for PKU, galactosemia, primary congenital hypothyroidism, and sickle cell disease and other hemoglobinopathies, the specimen was screened for additional *metabolic disorders* with MS/MS technology. MS/MS testing can screen for many conditions using just a few drops of blood.

### MS/MS "Unusual Result"

Your baby's specimen showed an "*unusual*" MS/MS screening result. Further testing will be needed to find out if your baby actually has a metabolic disorder. Not all babies with an "unusual" initial screening result have a disorder. In fact, metabolic disorders are rare and *babies often have normal results when retested*. However, it is important that all babies with an "unusual" result get tested. Babies can look healthy at birth and still have one of these disorders.

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