

Why Does My Baby Need More Testing for Biotinidase Deficiency?



State law requires that all babies have the newborn screening test before leaving the hospital. A few drops of blood were taken from your baby's heel and tested for certain diseases. Your baby now needs more testing as soon as possible. Not all babies with an initial "positive" result will have biotinidase deficiency (BD).

California
Department of
Public Health



Newborn Screening Branch
Genetic Disease Screening Program
www.cdph.ca.gov/nbs

Why Does My Baby Need More Testing?

Your baby's test showed a "positive" result for biotinidase deficiency (BD). More testing is needed to find out if your baby has this disease. Not all babies with an initial "positive" result will have this disease.

Babies can look healthy at birth and still have BD. If untreated, this disease can cause severe health problems.

What Is BD?

Biotinidase is an enzyme which the body needs to use and recycle biotin, a B vitamin found in food. Deficiency means there isn't enough of the enzyme. There are 2 types of BD, profound and partial. Babies with profound BD need to begin, and stay on, treatment to help prevent the health problems it can cause. Without treatment, babies with BD can have seizures, mental retardation, vision problems, hearing loss, and/or other health problems. Babies with partial BD usually don't have symptoms, and usually don't need treatment. More testing is needed to know if your baby has a type of BD.

How is BD treated?

Babies with profound BD must take a special biotin supplement every day. Treatment is life-long.

How Does a Baby Get This Disease?

A baby with BD inherited a gene from each parent. Both parents must carry the gene for a baby to inherit this disease.

Is BD Common?

California's rate is higher than the worldwide rate. In California, about 8 babies will be born with profound deficiency each year.

Other babies can have the milder form of BD. They may not need treatment. Newborn Screening only finds some of the babies with the milder form.

How Will I Know My Baby's Result?

The baby's doctor or the metabolic specialist will let you know the results.

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) for a diagnostic evaluation which includes more testing.

What Should I Do Right Now?

- Your baby's doctor or a metabolic specialist will advise you where to go to have your baby tested again.
- Continue breast milk or milk-based formula feedings as directed by the doctor while waiting for the test result.

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 with full benefits, 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. The diagnostic evaluation includes lab tests done through the state Biotinidase Deficiency Reference Lab at Stanford Hospital. You or your insurance carrier will not be billed for testing. You or your insurance carrier may be billed a small fee for specimen collection. To learn more about CCS, visit the website at: www.dhcs.ca.gov/ccs.

Where Can I Get More Information?

Call the doctor or the Newborn Screening staff listed below for any questions or concerns. For a complete list of diseases screened for by the California Newborn Screening Program visit our website at www.cdph.ca.gov/nbs

