

Family action sheet for biotinidase deficiency (BD)

A newborn blood screening result shows that your baby could have a serious condition called biotinidase deficiency. This result does not mean that your baby has this condition, but more tests are needed.

All babies born in California have a routine blood screen shortly after birth. The goal of this newborn screening is to find those at risk for serious medical conditions. Babies can look healthy at birth and still have one of these conditions. Babies with these conditions benefit from early diagnosis and treatment.

What is biotinidase deficiency or BD?

A baby with BD may have seizures, weak muscle tone, and hearing or vision loss. Symptoms can be mild or more severe, depending on how well an enzyme called biotinidase is working in the baby's body. Enzymes are a type of protein.

BD is inherited (passed down) in families. In BD, there is a change in a gene. Genes are in the body's cells. They carry information that sets the traits passed on to you from your parents. Both parents must have the gene change for the baby to get BD.

BD affects a gene that produces biotinidase. Biotinidase helps the body recycle a B vitamin called biotin. A baby with BD does not have enough biotin. Their body has trouble making energy and breaking down harmful substances.

Is there treatment for BD?

Yes, there are treatments for BD. Babies with this condition are treated with special biotin supplements. Children who get early and ongoing treatment for BD can have healthy growth and development.

Newborn screening takes blood from a newborn's heel to screen for medical conditions



Next steps

1. Your baby's medical provider will order more testing as soon as possible

These tests will let you know if your baby has BD.

2. Work closely with your baby's medical provider and follow recommendations

Your baby's medical provider may refer your baby to a doctor with more experience in BD. It is important that you keep all appointments and start treatment if instructed.

Where can I get more information?

Your infant's medical provider or metabolic specialist are the best people to teach you about BD. Find more information at these websites:

• Baby's First Test

(<https://www.babysfirsttest.org>). Type "biot" into the "Find a Condition" Box. Follow the link. For Spanish, click on the top right black box marked "en Español"

• Health Resources & Services Administration (HRSA)

(<https://newbornscreening.hrsa.gov/conditions/biotinidase-deficiency>)

