NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Biotinidase Deficiency (BTN)

What is newborn blood spot screening?

This is a simple blood test that is done shortly after a baby is born to test for conditions that could affect your child's health. A baby is tested at birth because it is important to start treatment early for best health outcomes.

There may be no signs of these conditions at birth. It is important to remember that many treatable conditions can not be seen by looking at your baby.

What is Biotinidase deficiency?

Biotinidase deficiency is an inherited (genetic) condition affecting the enzyme that is responsible for recycling a vitamin called Biotin. Biotin is an important helper for a number of processes in the body that make certain fats and carbohydrates and breakdown proteins. If the body is unable to recycle biotin, all of the enzymes that it helps are unable to do their jobs which causes serious health problems. Some people may still have a small amount of biotinidase enzyme and then will have a milder form of the disease.

Biotinidase deficiency is not caused by anything that happened during pregnancy.

What are the clinical features of the disease?

Babies with Biotinidase deficiency look normal at birth but they can develop serious problems without treatment. Problems can include seizures, low muscle tone, developmental delays, hearing loss and skin and hair problems.

If treatment for Biotinidase deficiency begins before symptoms start, babies can grow and develop normally.

Why does my baby need more tests for Biotinidase deficiency (BTN)?

It is normal that babies may need to have more tests, but it doesn't mean that your baby has Biotinidase deficiency (BTN). More testing is needed to find out if they do or do not have BTN. You will be referred to a specialist who will arrange more tests.

What is the treatment of the disease?

Babies with BTN are treated with biotin supplementation. The treatment is life long and is very effective at preventing the problems that go along with BTN.

Treatment is coordinated by specialists at BC Children's Hospital.

What is the outcome of treatment?

If biotin supplementation is continued lifelong, babies with BTN will grow and develop normally.

Can a family have more than one child with Biotinidase Deficiency?

Biotinidase deficiency is inherited as an autosomal recessive disease. Parents of a child with this condition are assumed to be carriers for the gene and have a 1 in 4 (25%) chance, in each pregnancy, of having another child with Biotinidase deficiency. Carriers are healthy and do not have symptoms.

What if I have more questions?

Be careful when searching for information on the internet as many websites are outdated and unreliable. We recommend speaking to your doctor or visiting:

www.perinatalservicesbc.ca/our-services/screening-programs/newborn-screening-program

www.babysfirsttest.org

Revised Sept 2022

Provincial Health Services Authority

Newborn Screening BC is a collaboration of Provincial Lab Medicine Services, BC Children's Hospital and BC Women's Hospital and Health Centre, and Perinatal Services BC, all part of the Provincial Health Services Authority.