Alberta Health Services
Alberta Newborn Screening

Talking with Parents > When talking to a parent whose baby has received an abnormal screen result, let them know that an abnormal screen result doesn't mean that the infant has the condition. It means that they have a higher chance of having the condition, and they need more tests to find out for sure. A printable sheet for parents with more condition specific information can be used to support your conversation, visit ahs.ca/nms and under Quick Reference click on *What conditions are screened for?*

Biotinidase (BIOT) Deficiency

(metabolic condition: organic acid disorder)

Information for Health Professionals

Also known as:

- BTD deficiency
- late-onset biotin-responsive multiple carboxylase deficiency
- late-onset multiple carboxylase deficiency

What are organic acid disorders?

Organic acid disorders (sometimes called organic acidemias) are a group of inherited metabolic conditions in which certain components of proteins, for example amino acids, cannot be broken down. This leads to an accumulation of toxic metabolites in the body which can cause serious health problems.

What is BIOT deficiency?

Biotinidase is an enzyme required for recycling biotin, one of the B group vitamins, in the body. Biotin is required for the normal function of carboxylase enzymes, key enzymes in the metabolism of proteins, fats and carbohydrates. In the absence of biotinidase, individuals may present with clinical features of BIOT deficiency.

What causes BIOT deficiency?

BIOT deficiency is caused by pathogenic variants in the biotinidase gene which results in decreased or absent activity. Some pathogenic variants may cause partial deficiency of biotinidase activity.

How common is BIOT deficiency?

The incidence of BIOT deficiency is about 1 in every 80,000 infants born in Canada.

What are the clinical features of BIOT deficiency?

Infants with BIOT deficiency appear normal at birth. Clinical features are variable depending on the dietary intake of biotin and the degree of residual biotinidase activity. Symptoms may develop in the first few weeks or months of life. While a minority of infants present with a life-threatening metabolic crisis, most infants present in the first few months of life with skin rash, hair loss, lethargy, seizures, hearing and visual problems. Individuals with partial biotinidase deficiency may be asymptomatic but may develop clinical features when stressed or with minor illness.

What is the screening test for BIOT deficiency?

Absence or a marked decrease in biotinidase activity is detected on the newborn blood spot screen. Newborn blood spot screening will not detect all infants with BIOT deficiency. Infants with clinical symptoms of BIOT deficiency need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of BIOT deficiency is confirmed by biochemical diagnostic blood and urine testing. Molecular genetic analysis may also be performed. The Clinical and Metabolic Genetics Program will arrange diagnostic testing.

How is BIOT deficiency treated?

BIOT deficiency is treated with oral supplementation of biotin. Individuals with BIOT deficiency are monitored by a team of specialists at the clinic listed below. Early treatment of BIOT deficiency before symptoms develop is associated with good outcome. Patients with BIOT deficiency require regular monitoring and lifelong treatment.

Is BIOT deficiency inherited?

BIOT deficiency is inherited as an autosomal recessive disorder. Parents of a child with BIOT deficiency are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. BIOT deficiency carriers are healthy. Genetic counselling is available to families with BIOT deficiency.

Additional resources are available through:

Clinical & Metabolic Genetics Program (Edmonton)

8-53 Medical Sciences Building
8440 – 112 St. NW
Edmonton, AB T6G 2H7
Phone: 780-407-7333
Fax: 780-407-6845

Emergency consultations:
Phone 780-407-8822 and ask for the specialist on call for metabolic diseases.

Clinical & Metabolic Genetics Program (Calgary)

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7587
Fax: 403-955-3091

Emergency Consultations:
Phone 403-955-7211 and ask for the specialist on call for metabolic diseases.

Early screening and follow-up care – every baby, every time

For more information about the Alberta Newborn Screening Program, visit www.ahs.ca/newbornscreening

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