Alberta Health Services

Newborn Metabolic Screening

Information for Health Professionals

Biotinidase Deficiency (BIOT)

(metabolic condition: organic acid disorder)

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?

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, patients may present with clinical features of BIOT.

What causes BIOT?

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

How common is BIOT?

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

What are the clinical features of BIOT?

Newborns with BIOT 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?

Absence or a marked decrease in biotinidase activity is detected on the newborn blood spot screen. Newborn blood spot screening is not 100% perfect. Infants with clinical symptoms need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of BIOT is confirmed by applicable biochemical diagnostic blood and urine testing. Confirmation by mutation analysis can also be performed. Specialists at the clinics listed below will arrange diagnostic testing.

How is BIOT treated?

BIOT is treated with oral supplementation of biotin. Parent education is an important component of treatment, including advising parents on actions to be taken when their child develops a minor illness. The treatment is lifelong.

What is the outcome of treatment for BIOT?

Early treatment of BIOT before symptoms develop is associated with good outcome. Patients with BIOT require regular monitoring and lifelong treatment.

Is BIOT inherited?

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

For additional resources, please call:

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.