Alberta Health Services

Newborn Metabolic Screening

Information for Health Professionals

Propionic Acidemia (PA)

(metabolic condition: organic acid disorder)

Also known as:

- propionic aciduria
- ketotic glycinemia
- ketotic hyperglycinemia

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 PA?

Propionyl-CoA carboxylase is an enzyme involved in the breakdown of several amino acids which are components of all proteins. When this enzyme is missing, toxic metabolites accumulate.

What causes PA?

PA is caused by mutations in the gene for propionyl-CoA carboxylase resulting in absent or decreased enzyme activity.

How common is PA?

The incidence of PA is about 1 in every 50,000 to 100,000 infants born worldwide.

What are the clinical features of PA?

PA typically presents in the first few days of life with an acute metabolic crisis with lethargy, vomiting, enlarged liver, acidosis and increased ammonia level, which may progress to seizures, coma and death. Some patients with a less severe form present later with symptoms precipitated by a period of fasting or during a minor illness.

What is the screening test for PA?

A specific pattern of organic acid metabolites 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 PA is confirmed by detecting specific metabolites in urine or on blood acylcarnitine analysis, together with amino acid profile in blood. Further testing may include enzyme analysis and/or mutation analysis of the propionyl-CoA carboxylase gene. Specialists at the clinics listed below will arrange diagnostic testing.

How is PA treated?

PA is treated by avoidance of fasting and dietary supplementation with carnitine. A low protein diet and a special formula low in some amino acids are also recommended. Parent education is an important component of treatment, including advising parents on actions to be taken when their child develops a minor illness. Prompt treatment of a metabolic crisis with intravenous fluids of glucose and lipids is necessary. The treatment is lifelong.

What is the outcome of treatment for PA?

Prevention and prompt treatment of metabolic crises improve the outcome of PA. However, some patients may have developmental delay and other neurological problems even with treatment.

Is PA inherited?

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

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.