Alberta Health Services
Newborn Metabolic Screening

Information for Health Professionals

Isovaleric Acidemia (IVA)
(metabolic condition: organic acid disorder)

Also known as:

- isovaleric acid-CoA dehydrogenase deficiency
- IVD deficiency
- isovaleryl-CoA dehydrogenase 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 IVA?

Isovaleryl-CoA dehydrogenase is an enzyme involved in the breakdown of the amino acid leucine, a component of all proteins. When this enzyme is missing, toxic metabolites accumulate.

What causes IVA?

IVA is caused by mutations in the gene for isovaleryl-CoA dehydrogenase resulting in absent or decreased enzyme activity.

How common is IVA?

IVA is a rare condition with an incidence of about 1 in every 100,000 to 200,000 infants born in Canada.

What are the clinical features of IVA?

Most infants with IVA present in the first few days of life with an acute metabolic crisis with lethargy, vomiting, acidosis and increased ammonia level, which may progress to seizures, coma and death. Infants may have a characteristic smell of sweaty feet. 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 IVA?

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 IVA is confirmed by detecting specific metabolites in urine or on blood acylcarnitine analysis. Further testing may include enzyme analysis and/or mutation analysis. Specialists at the clinics listed below will arrange diagnostic testing.

How is IVA treated?

IVA is treated by avoidance of fasting and dietary supplementation with carnitine and glycine. A low protein diet and a special formula low in leucine 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 IVA?

Prevention and prompt treatment of metabolic crises improves the outcome of IVA. However, some patients may develop neurological problems even with treatment.

Is IVA inherited?

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

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.