Alberta Health Services

Newborn Metabolic Screening

Information for Health Professionals

Glutaric Acidemia Type 1 (GA1)

(metabolic condition: organic acid disorder)

Also known as:

- glutaryl-CoA dehydrogenase deficiency
- glutaric aciduria type 1
- dicarboxylic aminoaciduria

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 GA1?

Glutaryl-CoA dehydrogenase is an enzyme involved in the breakdown of the amino acids lysine, hydroxylysine and tryptophan, components of all proteins. When this enzyme is missing, toxic metabolites accumulate and can cause neurological problems.

What causes GA1?

GA1 is caused by mutations in the glutaryl-CoA dehydrogenase gene resulting in absent or decreased enzyme activity.

How common is GA1?

The incidence of GA1 is about 1 in every 40,000 infants born worldwide.

What are the clinical features of GA1?

While most newborns with GA1 may appear normal at birth some may have an increased head size (macrocephaly). Some children develop normally during the first year of life, while others may be jittery, irritable and hypotonic. Subdural hematomas not associated with trauma may also occur. Without treatment, infants may have an acute metabolic crisis, often precipitated by a minor illness or period of fasting. Clinical features include acidosis, seizures and encephalopathy. Patients often have persistent neurological problems as a consequence with abnormal movements, poor coordination and muscle weakness.

What is the screening test for GA1?

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 GA1 is confirmed by detecting specific metabolites in urine or on blood acylcarnitine analysis. Further testing may include enzyme analysis and/or mutation analysis. The glutaric acid levels are elevated and typically 3-OH-glutaric acid is found. Specialists at the clinics listed below will arrange diagnostic testing.

How is GA1 treated?

GA1 is treated by avoidance of fasting and dietary supplementation with carnitine. A low protein diet and supplementation with a special formula is required. Prompt treatment of any infection is also recommended to minimize risk of neurological problems. 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 and glucose and carnitine is essential to prevent neurological damage. The treatment is lifelong.

What is the outcome of treatment for GA1?

The outcome of GA1 is good if a metabolic crisis can be prevented. However, there are mild and severe variants of GA1 and the outcome may be variable. Even with treatment some patients may develop neurological problems.

Is GA1 inherited?

GA1 is inherited as an autosomal recessive trait. Parents of a child with GA1 are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. GA1 carriers are healthy. Genetic counselling and prenatal testing is available to all families with GA1.

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 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.