

Classic Galactosemia (GALT)

(metabolic condition: other)

Information for Health Professionals

Also known as:

- galactosemia type 1
- galactose-1-phosphate uridylyltransferase (GALT) deficiency

What is GALT?

Classic galactosemia (GALT) is a disorder of sugar metabolism. Infants with this metabolic condition are not able to metabolize galactose, a sugar found in all milk products including breast milk. In these infants, galactose and its toxic byproducts build up in large amounts in blood and other tissues.

What causes GALT?

GALT is caused by a deficiency of galactose-1-phosphate uridylyltransferase, an enzyme involved in converting galactose to glucose.

How common is GALT?

GALT is a rare condition with an estimated incidence of between 1 in 30 000 and 1 in 60 000 infants born in Canada.

What are the clinical features of GALT?

Infants with GALT appear normal at birth but they can develop serious health problems if they are not treated. Early signs of GALT include hepatomegaly, jaundice, hepatitis/liver failure, feeding problems, failure to thrive, lethargy, and irritability. Serious complications of GALT can include cataracts, seizures and sepsis.

What is the screening test for GALT?

Screening for GALT is performed by measuring GALT enzyme activity on the newborn bloodspot and if indicated, the amount of total galactose. Newborn screening will not detect all infants with GALT. Infants with symptoms or signs consistent with GALT need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of GALT is confirmed by measuring GALT enzyme activity in a blood sample. Molecular genetic analysis may be performed. The Clinical and Metabolic Genetics Program will arrange diagnostic testing.

How is GALT treated?

GALT is treated with a lactose-free diet including lactose-free formula in lieu of breast milk. Individuals with GALT are treated and monitored by a healthcare team including a metabolic specialist and a dietician. Treatment is lifelong. Early treatment reduces cataracts and acute liver complications. Even with appropriate treatment, some children with GALT show delays in learning and development, and females are at risk of ovarian insufficiency.

Is GALT inherited?

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

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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