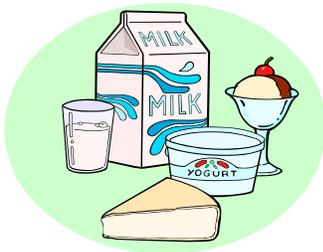


UNDERSTANDING GALACTOSEMIA

What is galactosemia?

Galactosemia is a genetic disorder that affects how certain sugars are broken down in the body. About 1 in 60 000 babies are born with galactosemia each year in Canada.

People with galactosemia are missing the liver enzyme that changes galactose into glucose for energy in the body. **Galactose** is a sugar that may be found on its own in food, but it is usually found as part of another sugar called **lactose**. Lactose, the sugar found in milk, is a combination of glucose and galactose.



You may be thinking, "What's the difference between galactosemia and lactose intolerance?" That's a good question. There is a difference. Galactosemia is life threatening, lactose intolerance is not. Untreated galactosemia causes brain damage, speech problems and reproductive problems; untreated lactose intolerance causes diarrhea, bloating and intestinal cramping.

How do people get galactosemia?

Galactosemia is an inborn error of metabolism. It is genetic. This means that the person has the disorder from the time they are conceived. At conception, the baby receives two sets of genetic material, one from the mother, and one from the father. This genetic material, called DNA, acts as a recipe for the baby's development.



DNA includes information about the baby's eye and hair colour, sex and even whether the baby will be right or left-handed. DNA comes in units called genes. Each pair of genes gives directions to a certain part of the body.

Galactosemia is caused by getting two copies of the faulty gene, one from the mother and one from the father. If the child inherits only one copy of the gene, they are a carrier for the disorder but are not affected. There is a 1 in 4, or 25% chance that 2 carriers of the gene will have a baby with galactosemia. Boys and girls are equally affected.

How is galactosemia diagnosed?

Since 1965, every baby born in British Columbia is tested for galactosemia at birth by a blood test. Babies born with galactosemia before 1965 were not diagnosed at birth. Because they were not on the special diet right away, they did not grow and develop normally. They are mentally challenged with severe speech problems. This is due to the effect of high galactose levels on the developing brain.

How is galactosemia treated?

1. DIET



Galactosemia is treated by **limiting** galactose from the diet. All sources of galactose and lactose must be **limited**. This is done with the help of a registered dietitian.

Examples of foods that must be eliminated from the diet:

- Milk from any animal (cow, goat)
- Cheese (**some aged cheeses are allowed—check with your dietitian**)
- Yogurt
- Cottage cheese
- Cream cheese
- Ice cream
- Sherbet
- Pudding
- Creamed soups and sauces
- Milk chocolate

Caution must be taken to **read labels** carefully for "hidden" sources of lactose in bread, crackers, cookies, chips, cereal, pancake mixes, hot dogs, bologna, breaded meat and fish, gravy, processed foods, artificial sweeteners, non-dairy creamers and drugs. Many foods and most drugs have lactose added during processing.



2. MEDICATIONS

- People with galactosemia still need **calcium and vitamin D** to maintain healthy bones. Since most dietary sources of calcium are found in foods containing lactose, calcium supplements are necessary. These supplements must be lactose-free. TUMS® are a good example. Both calcium and vitamin D supplements are sold at drug stores and pharmacies.
- Young women with galactosemia are at risk for **early menopause**. Hormone levels need to be closely monitored starting at age 9 or 10 to determine if hormone replacement therapy is needed. Young men with galactosemia appear to have normal hormonal development. Hormone replacement therapy can consist of pills, liquids, patches and/or shots to replace the female sex hormones. This ensures that the young woman with galactosemia goes through normal development.

What are the long-term complications of galactosemia?

If galactosemia is not diagnosed and treated at birth, it can cause liver, brain, eye and kidney damage. Fortunately, all babies born in British Columbia are tested for galactosemia at birth and started on the special diet. Despite this early diagnosis and good diet control, some people with galactosemia still grow up to have learning problems, speech difficulties and cataracts. Women often experience ovarian failure and are infertile. This is the frustrating part; the treatment for galactosemia is incomplete and no one has figured out exactly what is missing. What is known, however, is that the special lactose-free diet **greatly improves** the chances for normal development. There are many people with galactosemia who have closely followed the diet and have no adverse effects.

The balancing act

It is important to closely monitor any changes to the body that may result from galactosemia. The person with galactosemia should be followed by a metabolic physician and have regular bone density scans. These scans check bone strength and can identify osteoporosis.

Regular visits to the metabolic clinic are needed to consult with the team. Referrals can be made to other specialists (e.g. eye specialists) to closely monitor the effects of galactosemia on the body.

Blood tests are needed to check the galactose levels in the blood and liver function.

What is a "Normal" level?

What is tested is the **red blood cell galactose-1-phosphate** or "**gal-1-P**". This indicates the level of galactose in the blood. A person with galactosemia should have a blood gal-1-P level below **4.5 mg/100 mL**.



How can friends and family help?

Family and friends play a special role in supporting the person with galactosemia. It may be very difficult for the person to remain motivated and committed to treatment regimes, especially the lactose-free diet. Family and friends can provide support through understanding and providing a listening ear to frustrations.

Learn to prepare some favourite special dishes. If you are having a person with galactosemia over for a meal, ask them what they would like to eat and offer snack options such as:

- All nuts except hazelnuts
- Popcorn
- Plain potato chips
- Fresh fruits (oranges, plums, mangoes, grapefruits, apples, bananas)
- Angel food cake
- Beverages (tea, coffee, pop)

Helpful resources

Galactosemia Foundation

<http://galactosemia.org/>

Genetics Home reference: Galactosemia

<http://ghr.nlm.nih.gov/condition=galactosemia>

