Galactosemia
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with Galactosemia will have all of these features.

Galactosemia is a metabolic disorder, caused by a missing enzyme that converts galactose into glucose for energy. Galactose is a simple sugar that is found in many foods, particularly dairy products. Galactose may be found alone in foods but is usually found as part of another sugar, lactose. Galactosemia is typically diagnosed within the first few weeks of life through newborn screening. There are varying types of galactosemia, classical galactosemia and galactosemia variants**. The information on these pages applies only to classical galactosemia. There are mild forms that may be called galactosemia variants that are not included in this section.**

In classical galactosemia, the child must be put on a strict dairy-free and galactose-free diet immediately following birth, in order to help prevent severe complications such as cataracts, sepsis, multi-organ failure, brain damage or death. Maintaining this strict dairy-free/galactose-free diet is the only available treatment at this time.

Research suggests that despite strict dietary adherence, development, such as speech and motor, can be affected as early as pre-school age and continue into adulthood. Depending on how they are affected, children may have difficulties with communication, comprehension of new concepts, and sensory-motor integration skills.

An individual with galactosemia may or may not experience any of the following potential complications.

- Cataracts
- Learning difficulties
    - Speech/language difficulties
    - Difficulties with math or reading in school.
- Motor difficulties

- Neurological
    - Ataxia
    - Fine motor tremors
- Primary Ovarian Insufficiency: (POI)
- Growth delayed

Dietary

People with galactosemia need to completely avoid foods with ***galactose*** and/or ***lactose (glucose and galactose)***. This includes all dairy products from animals and foods with dairy products. If galactosemia is left untreated, galactose will accumulate in the blood and body tissues and will cause damage. Children with galactosemia may need a 504 plan to accommodate dietary needs

The following diet changes are necessary:

- No milk from any mammal is allowed (human milk, cow, goat, camel, ewe, yak etc.)…
- Besides dairy, also can not have anything with lactose or galactose, including dates, papayas, bell peppers, persimmons, tomatoes, and watermelon.
- Avoid breast milk, proprietary infant formulas containing lactose, cow's milk, dairy products, and casein or whey-containing foods
- Medications with lactose and galactose.

Foods and ingredients that contain galactose

Any food containing one or more of these ingredients is considered unacceptable for those with galactosemia:

- Butter
- Buttermilk
- Buttermilk solids
- Casein
- Cheese - including cottage cheese, cream cheese and other cheese-based products
- Cream
- Curds
- Dry milk
- Dry milk protein
- Garbanzo beans (chickpeas)

- Ghee
- Hydrolyzed protein (when made from casein or whey)
- Ice cream
- Lactalbumin (can also be called milk albuminate)
- Lactoglobulin
- Lactose
- Margarine (a few diet margarines or Kosher margarines do not contain)
- Milk products and are acceptable.
    - If margarine is listed as an ingredient, check with the manufacturer to make sure it is milk free.
- Milk
- Milk chocolate
- Milk solids
- Nonfat dry milk
- Nonfat dry milk solids
- Nonfat milk
- Organ meats (these include liver, heart, kidney, brains, sweetbreads, and Pancreas. These are often listed as "meat byproducts" on labels.)
- Sherbet (contains nonfat dry milk. This is different from *sorbet*)
- Sour cream
- Whey and whey solids
- Yogurt

Recommended Routine Surveillance

- Routine growth monitoring
- Routine monitoring for the accumulation of toxic analytes
- Monitor for cataracts
- Monitor for premature ovarian insufficiency
- Monitor for osteoporosis
    - Individuals may require calcium, vitamin D and vitamin K to prevent decreased bone mineralization
- Developmental evaluation:
    - Speech therapy
    - Physical therapy
    - Occupational therapy

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Individuals should have an emergency letter for healthy care providers who may not be familiar with Galactosemia. This letter should detail a detailed explanation of dietary restrictions

Emergencies should be handled as with any child.

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Endocrinologist
    - Delayed puberty
    - Primary and/or secondary amenorrhea
    - Hormone replacement

- Geneticist /Genetic Counselor:
    - Diagnosis
    - Coordination of care
    - Genetic risk for family
    - Clinical trials

- Metabolic Multispecialty Clinic
    - Clinical follow-up as needed with a metabolic specialist
    - Dietician familiar with Galactosemia
    - Social worker
    - Nurse specialist

- Neurologist
    - Ataxia

- Ophthalmologist
    - Cataract surveillance

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed with Classic Galactosemia. This type of galactosemia is characterized by a lack of an enzyme. Medical complications

with Galactosemia in an untreated individual could lead to life-threatening complications including feeding problems, failure to thrive, hepatocellular damage, bleeding, and E. coli sepsis. If a lactose-restricted diet is provided during the first ten days of life, the neonatal signs usually quickly resolve and the complications of liver failure, sepsis, and neonatal death are prevented. Individuals with classic galactosemia remain at increased risk for developmental delays, speech problems and abnormalities of motor function. A. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

"Galactosemia at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

GeneReviews- Galactosemia
http://www.ncbi.nlm.nih.gov/books/NBK1518

New England Consortium of Metabolic Programs
http://newenglandconsortium.org/for-families/galactosemia/

Understanding Galactosemia - A Diet Guide (PDF)
http://galactosemia.org/PDFs/UnderstandingGalactosemiaDietGuide3.pdf

International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up
https://pubmed.ncbi.nlm.nih.gov/27858262/

PDF version
https://onlinelibrary.wiley.com/doi/epdf/10.1007/s10545-016-9990-5