Overview of Glycogen Storage Disease

Glycogen storage disease (GSD) is an inherited disorder in which an abnormal amount or type of glycogen is stored in the liver. This abnormal storage results from the liver's inability to adequately regulate the metabolism of glycogen and glucose. "Glycogen storage disease occurs when an enzyme (proteins produced by the body) that regulates conversion of sugar (glucose) into its storage form (glycogen) or release of glucose from glycogen is missing" (Cincinnati Children's Hospital Medical Center [CCHMC], 2012).

"Many sugars (including glucose) are present in foods and are used by the body as a source of energy. After a meal, blood glucose levels rise. The body stores the extra glucose that is not needed right away as glycogen in the liver and muscles. Later, as the blood glucose levels in the body begin to decrease, the body uses this stored energy. These sugars, stored in the form of glycogen, need to be processed by enzymes in the body before they can carry out their functions. If the enzymes needed to process them are missing, the glycogen or one of its related starches can accumulate, causing problems" (CCHMC, 2012).

"There are at least 10 different types of GSDs, which are put into groups based on the enzyme that is missing. Approximately one in about 20,000 people are affected by glycogen storage diseases. The most common forms of GSD are types I, III and IV.

- GSD I (von Gierke disease) results from a deficiency of the enzyme Glucose-6-Phosphatase (CCHMC, 2012). It is the most common type of GSD and the effects are apparent very early in childhood. GSD I accounts for approximately 25 percent of all GSD cases" (American Liver Foundation, 2011).
- In GSD III (Cori disease) an enzyme called the debrancher is deficient, causing the body to form glycogen molecules that have an abnormal structure. This abnormal structure also prevents the glucogen from being broken down into glucose.
- In GSD IV (amylopectinosis) glycogen that accumulates in the tissues has very long outer branches. This is due to a genetic deficiency of the branching enzyme. This abnormal glycogen is thought to stimulate the immune system. The result is tremendous scarring (cirrhosis) of the liver as well as other organs, such as muscle and heart (CCHMC, 2012).

Causes of Glycogen Storage Disease

Glycogen Storage Disease (GSD) occurs when there is an absence or deficiency of one of the enzymes responsible for making or breaking down glycogen in the body. This is known as an enzyme deficiency (Association for Glycogen Storage Disease, 2012).

Symptoms of Glycogen Storage Disease

"Symptoms of GSD vary based on the enzyme that is missing. They usually result from the buildup of glycogen or from an inability to produce glucose when needed. Because GSD occurs mainly in muscles and the liver, those areas show the most obvious symptoms.
Symptoms of GSD may include:

- growth failure;
- muscle cramps;
- low blood sugar;
- enlarged liver;
- swollen belly; and
- abnormal blood test (CCHMC, 2012).

The age when symptoms begin and how severe they are depends on the type of GSD. Children with GSD I rarely develop cirrhosis (liver disease), but they are at an increased risk for developing liver tumors. In some ways, GSD III is a milder version of GSD I. It also is a very rare cause of liver failure, but it may cause fibrosis (early scarring of the liver, which may be caused by a healing response to injury, infection or inflammation).

GSD II is a muscle disease and does not affect the liver. Glycogen storage disease IV causes cirrhosis; it may also cause heart or muscle dysfunction. Often, infants born with GSD IV are diagnosed with enlarged livers and failure to thrive within their first year of life; they develop cirrhosis of the liver by age three to five" (CCHMC, 2012).

Treatment of Glycogen Storage Disease

Treatment of GSD depends on the type of GSD. Some GSD types cannot be treated; others can be treated by controlling the presenting symptoms. For the types of GSD that can be treated, patients must carefully follow a special diet as per the licensed care provider's recommendations.

- **Frequent high carbohydrate meals during the day.** For some children, eating several small meals rich in sugars and starches every day helps prevent blood sugar levels from dropping.
- **Cornstarch.** For some young children over the age of 2, giving uncooked cornstarch every four to six hours – including during overnight hours – can also relieve the problem.
- **Continuous tube feeding.** In order to maintain appropriate blood glucose levels, gastrointestinal tube feedings with solutions containing high concentration of glucose may need to be administered. Younger children may have to use this treatment method during the night until they get older. In the daytime the feeding tube is sometimes removed, but the patient must eat foods rich in sugars and starches about every three hours. This treatment can be successful in reversing most symptoms.
- **Drug treatment.** GSD tends to cause uric acid (a waste product) to accumulate, which can cause gout (painful inflammation of the joints) and kidney stones.
- Medication is often necessary (CCHMC, 2012).

Taken from "Guidelines for Managing Life-threatening Food Allergies in Connecticut Schools- CT. State Dept. of Education