Glucose-Galactose Malabsorption

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Glucose-galactose malabsorption is a condition in which the body cannot take in (absorb) the sugars glucose and galactose, which primarily results in severe diarrhea.

Keywords: genetic conditions

1. Introduction

Beginning in infancy, severe diarrhea results in weight loss and dehydration that can be life-threatening. Small amounts of the simple sugar glucose in the urine (mild glucosuria) may occur in this disorder. Rarely, affected infants develop kidney stones due to deposits of calcium in the kidneys (nephrocalcinosis).

The signs and symptoms of glucose-galactose malabsorption appear early in life when affected infants are fed breast milk or regular infant formulas. These foods contain glucose, galactose, and another sugar called lactose that gets broken down into these two sugars. When these sugar-containing foods are ingested by affected individuals, it leads to diarrhea and other health problems. If foods that contain glucose, galactose, and lactose are removed from the diet, the diarrhea stops.

2. Frequency

Glucose-galactose malabsorption is a rare disorder; only a few hundred cases have been identified worldwide. However, as many as 10 percent of the population may have a somewhat reduced capacity for glucose absorption without associated health problems.

3. Causes

Mutations in the *SLC5A1* gene cause glucose-galactose malabsorption. The *SLC5A1* gene provides instructions for producing a protein called sodium/glucose cotransporter protein 1 (SGLT1). This protein is found mainly in the intestinal tract and the kidneys. It spans the membrane of cells in these body systems and moves (transports) glucose and galactose from outside the cell to inside the cell. Sodium and water are transported across the cell membrane along with the sugars in this process. Glucose and galactose are simple sugars; they are present in many foods, or they can be obtained from the breakdown of lactose or other sugars and carbohydrates in the diet during digestion.

In the intestinal tract, the SGLT1 protein helps the body absorb glucose and galactose from the diet so the body can use them. During the digestion of food, the protein transports the sugars into the cells that line the wall of the intestine (intestinal epithelial cells) as food passes through.

The SGLT1 protein in kidney cells plays a role in maintaining normal blood glucose levels. The kidneys filter waste products from the blood and eliminate them in urine. They also reabsorb needed nutrients and release them back into the blood. The SGLT1 protein transports glucose into specialized kidney cells, ensuring that the sugar goes back into the bloodstream and is not released into the urine.

SLC5A1 gene mutations impair or eliminate the function of the SGLT1 protein. As a result, glucose and galactose are not absorbed by intestinal epithelial cells but instead accumulate in the intestinal tract. In addition, water that normally would have been transported with the sugars remains in the intestinal tract, resulting in dehydration of the body's tissues and severe diarrhea. The SGLT1 protein in kidney cells cannot transport glucose; however, other proteins in the kidneys are able to absorb enough glucose into the bloodstream, so that glucosuria is mild, if present at all, in people with glucose-galactose malabsorption.

3.1. The gene associated with Glucose-galactose malabsorption

- SLC5A1

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- carbohydrate intolerance
- complex carbohydrate intolerance
- congenital glucose-galactose intolerance
- congenital glucose-galactose malabsorption
- GGM
- monosaccharide malabsorption

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