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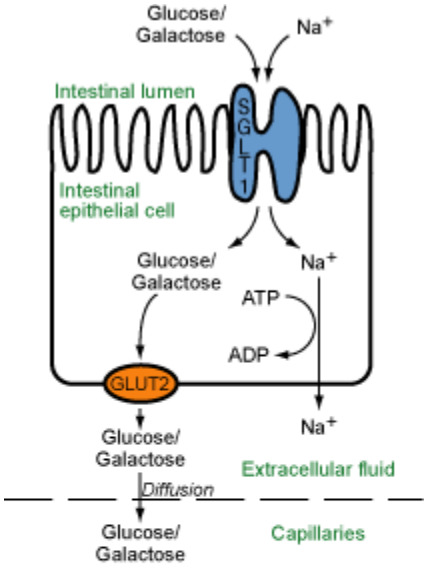
Glucose galactose malabsorption



Co-transport of sodium and glucose or galactose by SGLT1. For every two sodium ions SGLT1 moves inside the cell down the sodium concentration gradient, one glucose or galactose molecule moves with it. The glucose/galactose is then transported into the extracellular fluid by GLUT2, and diffuses into the capillaries. Sodium is actively transported out of the cell into the intercellular space so as to maintain the intracellular sodium concentration gradient.

Glucose Galactose Malabsorption (GGM) is a rare metabolic disorder caused by a defect in glucose and galactose transport across the intestinal lining. GGM is characterized by severe diarrhea and dehydration as early as the first day of life and can result in rapid death if lactose (milk sugar), sucrose (table sugar), glucose, and galactose are not removed from the diet. Half of the 200 severe GGM cases found worldwide result from familial intermarriage. At least 10% of the general population has glucose intolerance, however, and it is possible that these people may have milder forms of the disease.

GGM is an autosomal recessive disorder in which affected individuals inherit two defective copies of the *SGLT1* gene, located on chromosome 22. Normally within the space enclosed by the small intestine (called the lumen), lactose is broken down into glucose and galactose by an enzyme called lactase, while sucrose is broken down into glucose and fructose by an enzyme called sucrase. The protein product of *SGLT1* then moves the glucose and the galactose from the lumen of the small intestine into intestinal cells. Usually the mutations carried by GGM

individuals result in nonfunctional truncated SGLT1 proteins or in the improper placement of the proteins such that they can not transport glucose and galactose out of the intestinal lumen. The glucose and galactose, if left untransported, draw water out of the body into the intestinal lumen, resulting in diarrhea.

Although no cure exists for GGM, patients can control their symptoms (diarrhea) by removing lactose, sucrose, and glucose from their diets. Infants showing a prenatal diagnosis of GGM will thrive on a fructose-based replacement formula and will later continue their "normal" physical development on a fructose-based solid diet. Older children and adults with severe GGM can also manage their symptoms on a fructose-based diet and may show improved glucose tolerance and even clinical remission as they age.

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