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Congenital Glucose and Galactose Malabsorption

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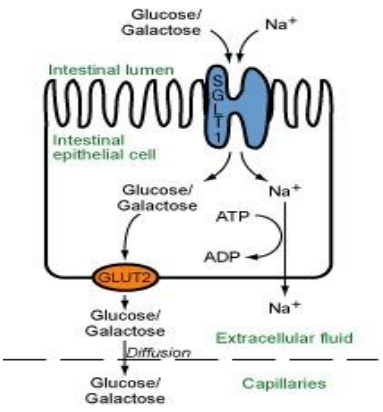
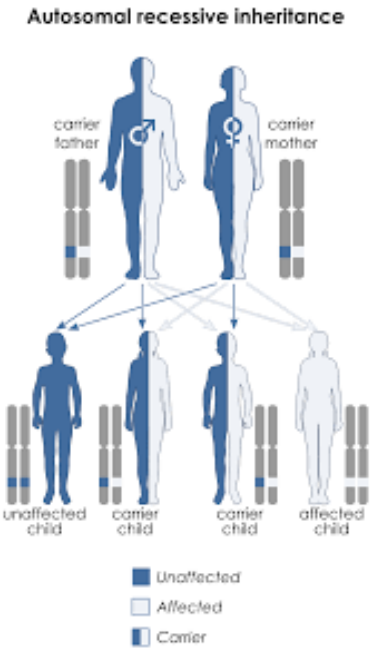
Abstract: ***Congenital Glucose and Galactose Malabsorption is a rare autosomal recessive disorder. The condition arises due to an impairment in the transport of glucose and galactose across the intestinal lining, which is normally sodium - coupled. From the very first day of life, Congenital Glucose and Galactose Malabsorption manifests as profound diarrhea and dehydration. If lactose (milk sugar), sucrose (table sugar), glucose, and galactose are not eliminated from the diet, CCGM poses a significant risk of causing fatality. In the space enclosed by the small intestine, referred to as the lumen, lactose is typically broken down into glucose and galactose by an enzyme known as lactase. An enzyme known as sucrase breaks down sucrose into glucose and fructose. Subsequently, the protein produced by SGLT1 facilitates the transfer of glucose and galactose from the small intestine's lumen into the cells of the intestine. Usually, the mutations carried by individuals with Congenital Glucose and Galactose Malabsorption result in nonfunctional truncated SGLT1 proteins or in the improper placement of the proteins, preventing them from transporting glucose and galactose out of the intestinal lumen. The untransported glucose and galactose draw water out of the body into the intestinal lumen, resulting in diarrhea.***



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.



Half of 200 sever Glucose and Galactose Malabsorption cases found world wide result from familial intermarriage. It is an metabolic disorder in which affected individuals inherit two defective copied of the SGLT1 gene, located on chromosome 22.

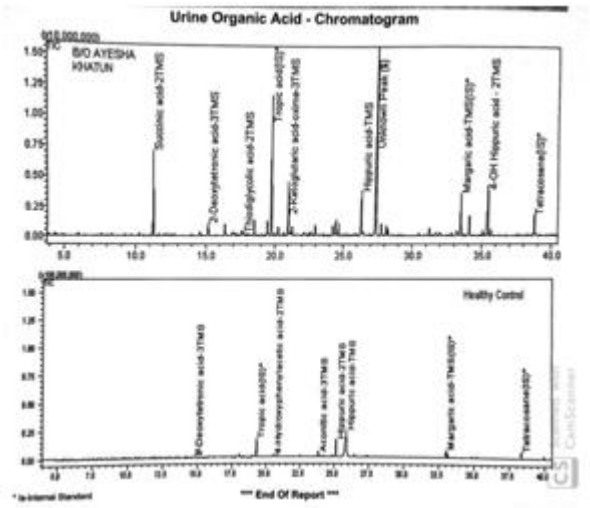
1. Case Study

A two - month - old child presented with a history of 7 to 8 loose stools since past 2 months prior to admission. For the above complaints child was investigated and treated at a nearby hospital. In view of the persistence of symptoms, he was referred to Children's Hospital for further management.

Birth history:
Born to a consanguineous parent, with a birth weight of 2.8 kg. The child was fed direct breast milk from day 1 of life and had loose stools from the 16th day of life. Treated in a local hospital. With the advice of a local pediatrician, the child was given chicken soup and continued DBM.

Hospital stay:
The child was admitted and treated withlactose - free formula, initially started with cyclical feeds. As per tolerance child was advised to start oral feeds.

In the ward stay child had recurrent hypoglycaemic attacks, which were treated accordingly and the child had regular blood sugar monitoring which remained normal. In view of this child was seen by the Consultant Genetist, who advised to do stool for reducing substance (is a stool sample test used to diagnose lactose intolerance and some rare metabolic abnormalities.) which was negative, Urine for reducing substance was positive (1.0%). Total Galactose levels (is a quantitative fluorometric test for the total galactose from the dried blood spot samples. The test is based on the combined determination of free and galactose liberated from the cleavage of the galactose – 1 – phosphate) 3.0 mg/dl (cut off value: <10mg/dl), and Immune Trypsin levels 2.1 ng/ml (cut off value: < 70 ng/ml), TMS was normal, Urinary organic acidogram shows mild elevation of Thiodiglycolic acid and 4 - hydroxy phenyl lactic acid below figure.



2. Discussion

Glucose - galactose malabsorption is a rare genetically determined disorder believed to be inherited as an autosomal recessive trait (Meeuwisse & Dahlqvist 1968, Elsas et al.1970). The absorptive defect is confined to the two structurally similar monosaccharides: glucose and galactose. Severe diarrhea is present shortly after birth, and a precise diagnosis with the elimination of dietary glucose and galactose may be life - saving.

The strict diagnostic criteria for congenital glucose - galactose malabsorption are:

- Watery diarrhea shortly after birth
- Clinical improvement upon withdrawal of dietary glucose and galactose
- Normal mucosal disaccharidase activities
- Absorptive defect confined to glucose and galactose.

Although no cure exists for CGGM, patients can manage their symptoms by removing lactose, sucrose, and glucose from their diets. Infants who receive a prenatal diagnosis of GGM will thrive on a fructose - based replacement formula.

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References

[1] Brown GK. Glucose transporters: structure, function and consequences of deficiency. J Inherit Metab Dis.2000 May; 23 (3): 237 - 46. Review.
[2] El - Naggar W, Balfe JW, Barbar M, Taha D. Nephrocalcinosis in glucose - galactose malabsorption, association with renal tubular acidosis. Pediatr Nephrol.2005 Sep; 20 (9): 1336 - 9. Epub 2005 Jul 12.
[3] Lam JT, Martín MG, Turk E, Hirayama BA, Bosshard NU, Steinmann B, Wright EM. Missense mutations in SGLT1 cause glucose - galactose malabsorption by trafficking defects. Biochim Biophys Acta.1999 Feb 24; 1453 (2): 297 - 303.
[4] Wright EM, Hirayama BA, Loo DF. Active sugar transport in health and disease. J Intern Med.2007 Jan; 261 (1): 32 - 43. Review.
[5] Wright EM, Martín MG, Turk E. Intestinal absorption in health and disease - - sugars. Best Pract Res Clin Gastroenterol.2003 Dec; 17 (6): 943 - 56.
[6] Wright EM. I. Glucose galactose malabsorption. Am J Physiol.1998 Nov; 275 (5 Pt 1): G879 - 82. Review.
[7] Kay Tanaka et. al. Clin. Chem., (13), 1839 - 1846 (1980).
[8] Nenad Blau, Marinus Duran, Milan E. Blaskovics, Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases.