Glucose-galactose malabsorption: a case report

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Abstract

Glucose-Galactose Malabsorption (GGM) is one of the rare autosomal recessive disorders of intestinal transport of glucose and galactose/Na+ cotransport system (SGLT1) that leads to osmotic watery diarrhea, dehydration, failure to thrive, or early death. It is caused by mutations in the gene coding for the intestinal brush border of sodium-glucose co-transporter. More than 30 different mutations in this gene have been found to cause abnormalities in the transporter. Because of the wide number of mutations known today, genetic testing for defects is difficult, leaving clinicians to rely on clinical testing, including the glucose or galactose hydrogen breath test as a diagnostic test of choice. Treatment includes the elimination of glucose and galactose from diet. We report a male infant with suspected GGM with acidosis and diarrhea.

Keywords: Glucose-galactose malabsorption, Symptoms, Treatment

Case Report

A 56-day-old Iranian boy was admitted to our ward with diarrhea, poor feeding, fever, and dehydration. He was born at 38 gestational weeks with normal and good prenatal care and with a birth weight of 2000 g, but after that his weight was 2500 g. He was the first child of parents. He was fed with breast milk after the birth. He was admitted several times for his diarrhea. His symptom was watery diarrhea (10-18 times/day and approximately 100-140 ml/kg, measured by weighing the napkin). It was developed within four weeks after birth. Fever, diarrhea, and dehydration were severe from two days before the admission. In our ward he was ill with the signs of dehydration such as lethargy, confusion, tachypnea, and hypotension. In physical examination, he had dehydration, sunken eye, heart and lungs were normal, abdominal examination was normal but no moro reflex. Laboratory examination was Hb 10 g/dL, BS 60 mg/dL, NA 155 mmol/L, K 4.1 mmol/L, CL 110 mmol/L, BUN 42 mg/dL, creatinine 1.1 mg/dL, calcium 7.9 mg/dL, Mg 1.9 mg/dL, phosphate 4.6 mg/dL. The arterial blood gas analysis was a pH of 7.20, serum bicarbonate (HCO3) 10.1 mmol/L, base excess (BE) -17 mmol/L, pCO2 30.1 mmHg and pO2 100 mmHg. Urine analysis was normal. CMV, HIV, TORCH study was done in a normal range. Stool pH was 5.3, and stool sugar testing by Clinitest was positive 3+. No fat droplet was detected in stool examination. Stool osmolarity was compatible with osmotic diarrhea (osmotic gap: 129 mOsm/kg). S/E and S/C were normal (Table 1). So, at first we started 3×20 cc/kg NS, then acidosis and dehydration were improved. Watery diarrhea and dehydration restarted after the infant was re-fed with breast milk, so we commenced for the patient the lactose free formula (Soya and Amino-acid based formula sequentially) but diarrhea and acidosis did not improve. Thyroid function tests, LFT, and sweat tests were normal. Echocardiography and abdominal ultrasonography were normal as well. Ophthalmologic examination for ruling out the cataract (galactosemia) was normal. Metabolic tests and Ig flow cytometry were normal. Glucose loading test with 0.5 g/kg oral glucose revealed a flat serum glucose curve with fasting. Blood glucose was measured at 0, 30, 60, 90 and 120 minutes after the loading of glucose, and blood glucose levels were at 80, 87, 111, 99, and 100 mg/dL. Hydrogen breath test was above 20 pp. Therefore, after the lab tests were completed, we started fructose-based formula (galactomin B-19). In this case, after 72 hours, diarrhea was improved as well as the lab test. We could not observe complication in infant with galactomin 19 formula and after four weeks the patient’s weight increased from 2500 g to 3150 g.

Discussion

Glucose-galactose malabsorption (GGM) is a rare autosomal recessive metabolic disorder which is characterized by the intestine's inability to transport...
and absorb glucose and galactose (simple sugars or monosaccharide). Infants show severe diarrhea resulting in life-threatening dehydration, acidosis, and weight loss, when they are fed with breast milk or regular infant formulas (1-3). The diagnostic criteria for congenital GGM are osmotic watery diarrhea, positive reducing substance in stool, a normal intestinal biopsy (no evidence of enteropathy), flat blood glucose curve or positive glucose breath hydrogen test following oral glucose load and diarrhea promptly stopped after initiating fructose containing feeds (3-5). Fructose is the only carbohydrate that can be given safely to the patients. Therefore, diarrhea immediately ceases when infants are given such a formula (5,6).

Our patient was diagnosed to have GGM because his diarrhea started in early infancy. His stool pH was acidic (osmotic diarrhea) with positive clinitest, flat blood glucose curve following oral glucose load and diarrhea promptly stopped after initiating fructose containing feeds (3-5). Fructose is the only carbohydrate that can be given safely to the patients. Therefore, diarrhea immediately ceases when infants are given such a formula (5,6).

The differential diagnoses of GGM were microvillus atrophy, congenital chloride diarrhea, congenital sodium diarrhea, acrodermatitis enteropathica, disorders villous architecture such as microvillus inclusion disease, tufting enteropathy, milk protein allergy, autoimmune enteropathy, disaccharidase deficiencies, and cystic fibrosis (5-8). GGM is a rare disorder with osmotic diarrhea, dehydration, and acidosis in early infancy that should be considered in patients with diarrhea. It should be noted that after starting glucose-galactose free formula the symptoms were solved (7-10).

**Ethical issues**

The study was approved by the ethical committees of the Mofid hospital.

**Authors’ contributions**

All authors passed four criteria for authorship contribution based on recommendations of the International Committee of Medical Journal Editors (ICMJE).

**References**