

A Case Report of Glucose-Galactose Malabsorption in Iranian Child

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Abstract

Introduction

Glucose-Galactose Malabsorption (GGM) is an autosomal recessive and rare disorder of intestinal transport of glucose and sodium-glucose cotransporter type (SGLT1).

Case Report

Our patient is a 32-day-old boy who was examined for severe diarrhea and acidosis and was treated with GGM diagnosis. A number of laboratory tests were performed on this patient as well as positive test for reduced substance of stool and positive hydrogen breath test. On the other hand, the improvement of diarrhea with fasting and the initiation of a glucose and galactose free formula (fructose-based formula [galactomin B-19]) was instructed. He was treated and followed with diagnosis of GGM.

Conclusion

In summary, careful clinical observation, laboratory tests, and the character of the external cues may provide indications of GGM.

Key Words: Case Report, Child, Diarrhea, Glucose-Galactose Malabsorption.

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1- INTRODUCTION

Glucose-Galactose Malabsorption (GGM) is an autosomal recessive and rare disorder of intestinal transport of glucose and Sodium-glucose cotransporter type 1 (SGLT1) (1, 2). This disorder leads to severe watery diarrhea that is osmotic and dehydration, failure to thrive, or early death (3, 4). This is due to a mutation in the gene encoded for the intestinal brush border of sodium-glucose co-transporter (5). In this gene more than 30 different mutations have been found that cause disturbances in transporter (1, 5). Genetic testing for defects is difficult because a large number of mutations are known today (6). Therefore, many doctors use the glucose or galactose hydrogen breath test for diagnosis. The treatment involves the removal of glucose and galactose from the diet (5, 7). Our patient was a 32-day-old boy who was examined for severe diarrhea and acidosis and was treated with GGM diagnosis.

2- CASE REPORT

The first case of GGM from Iran was a two month-old boy with a history of watery, acidic diarrhea on the third day of his life that was reported in 2007 (8). Our patient was an Iranian 32-day-old boy in our area hospital (in Semnan, Iran), admitted with diarrhea, poor feeding, fever and severe dehydration. He was born at 37 weeks of pregnancy without any prenatal problem and with a birth weight of 2,900 gr, but after that his weight dropped to 2,780 gr. He was the first child of his parents. He was fed with breast milk after birth, and from the second day of his birth, he had severe diarrhea, and was once admitted to the hospital due to the severe dehydration. Recently he was admitted to the intensive care unit (ICU) of our hospital due to diarrhea, fever, severe dehydration, lethargy and confusion. He had watery diarrhea (10-14 times/day and approximately 100-150 ml/kg, measured

by weighing the napkin) that was developed within two weeks after birth. In physical examination, he had dehydration, peeling skin, sunken eye and his fontanel was depressed; heart and lungs were normal, abdominal examination was normal but Moro, grasp and sucking reflex was decreased. Cytomegalovirus, Human immunodeficiency virus, toxoplasmosis, syphilis, varicella zoster, parvovirus B19 and Rubella, cytomegalovirus and herpes infection (TORCH) study were done and results were in a normal range. Stool pH was 5.3, and stool sugar testing by Clinitest was positive (2+). No fat droplet was detected in stool examination. Stool osmolality was compatible with osmotic diarrhea (osmotic gap: 129 mosm/kg). Stool exam and Stool culture were normal (**Table.1**). Thyroid function tests, sweat tests and LFT were normal. Abdominal and brain ultrasonography and echocardiography were normal as well.

Ophthalmologic test was normal (Cataract in the field of galactosemia was ruled out). Immunoglobulin flow cytometry and metabolic tests were normal. With 0.5 g/kg oral glucose loading test showed 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 82, 86, 113, 95, and 99 mg/ dL. For treatment we started 3×20 cc/kg NS, then acidosis and dehydration were improved. Water diarrhea and dehydration started again after breastfeeding, so we started the lactose-free formula for the patient; but diarrhea and acidosis did not improve. Then, after starting treatment and correcting electrolyte impairment, in the next step, we started the fasting for the patient. After 24 hours, infant diarrhea improved, and in the next step, nutrition began with fructose-based formula (Galactomin 19). For this patient Hydrogen breath test was done and above 20 ppm (parts per million). A number of

laboratory tests were performed on this patient as well as positive test for reduced substance of stool and positive hydrogen breath test. On the other hand, the improvement of diarrhea with fasting and the initiation of a glucose and galactose-free formula (fructose-based formula: Galactomin B-19) were instructed. He was

treated and followed with diagnosis: glucose galactose malabsorption. In this case, after 72 hours, diarrhea acidosis and other lab tests were improved and in follow up after two weeks the patient's weight increased from 2,780 gr to 3,300 grams.

Table-1: Laboratory test of the patient

Lab (unit)	At admission	After Galactomin 19
pH (mmhg)	7.24	7.41
HCO ₃ (mmol/L)	13.7	19.5
PCO ₂ (mmhg)	32	46
BE (mmol/L)	-12.4	-1
Na (mmol/L)	174	147
K (mmol/L)	4.1	4.7
Mg (mg/dL)	2	2.1
Ca (mg/dL)	10.1	10
P (mg/dL)	4.2	4.5
Cl (mmol/L)	108	110
BUN (mg/dL)	42	10
CR (mg/dL)	2.4	.4
WBC (n/ml)	17500	10000
HB (g/dL)	9.5	10.9
PLT (n/ mm ³)	130000	210000
S/E (rate)	NL	NL
S/C	NL	NL
Reduced substance	3+	Neg.
LFT	NL	NL
TFT	NL	NL
ESR (mm)	49	16
CRP (mg/L)	2+	Neg.

Abbreviations: BMI: Body mass index; Hb: Hemoglobin; WBC: White blood cell; PLT: Platelet; BS: Blood sugar; P: Potassium ; Na: Sodium; CL: Chloride; BUN: Blood urea nitrogen; Mg: Magnesium; Cr: Creatinine; ABG: Arterial blood gas; PCO₂: Partial pressure of carbon dioxide; S/E: Stool exam; S/C: Stool culture; LFT: Liver function test; TFT: Thyroid function test; ESR: Erythrocyte sediment rate; CRP: C-reactive protein.

3- DISCUSSION

Glucose-galactose malabsorption is an autosomal recessive and rare metabolic disorder that results from inability of the intestine to transmit and absorb glucose and galactose. In this disorder, an infant who is fed with breast milk or formula suffers from a life threatening watery diarrhea which leads to severe dehydration, acidosis and weight loss (1, 3,

9). The disease is characterized by watery diarrhea, a positive test for reduced substance of stool and a normal biopsy of the intestine (no evidence of enteropathy), and flat blood glucose curve or positive glucose breath hydrogen test following oral glucose load that stops after feeding with fructose containing formula (3, 5). When this formula is given to the patient, diarrhea stops and so fructose is the only

carbohydrate that can be given to him (10). Other diagnoses that should be differentiated from GGM are congenital chloride diarrhea, microvillus atrophy, acrodermatitis enteropathica, congenital sodium diarrhea, disorders villous architecture such as tufting enteropathy, microvillus inclusion disease, autoimmune enteropathy, disaccharidase deficiencies, milk protein allergy, and cystic fibrosis (10, 11). Family history of osteogenic diarrhea in a neonate period in an older sister and sister who continues with fluid therapy can suggest this diagnosis (12).

Prognosis of GGM in the medium term is usually good. When these patients become older, they can sometimes tolerate a small amount of glucose without diarrhea. By colonic bacteria the unabsorbed glucose is fermented. The main concern in these patients is that they will not tolerate these particular formulas in the long term and will have consequences for the diet containing high protein and fat (13).

Our patient had severe diarrhea from the first days of birth. In the lab test, his stool was acidic and he had a positive Cline test, and the blood glucose curve was flat after oral glucose loading. Diarrhea did not respond to the lactose free diet, but with the starting of fructose-based formula (Galactomin 19) diarrhea was discontinued. Therefore, it was treated with GGM and discharged. Due to lack of access to the genetic test it was not performed for the patient.

4- CONCLUSION

In summary, careful clinical observation, laboratory tests, and the character of the external cues may provide indications of GGM. Early diagnosis and initiation of appropriate nutrition with fructose-based formula will prevent morbidity and mortality.

5- CONFLICT OF INTEREST: None.

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