Journal of Pediatrics and Neonatology

ISSN 2688-5573

Case Report

Glucose Galactose Malabsorption in a Neonate Complicated by Renal Failure

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Abstract

We report a case of glucose galactose malabsorption in a patient who presented with renal failure. Our aim is to inform pediatricians about the life-threatening complications associated with glucose galactose malabsorption, especially renal complications.

Introduction

Glucose Galactose Malabsorption (GGM) is a rare autosomal recessive disorder with an *SLC5A1* gene mutation that encodes the sodium/glucose cotransporter [1-3]. This gene is present in the intestinal mucosa, proximal renal tubules, heart, and parotid and submandibular salivary glands [2].

Cases usually present in the first few days of life with intractable diarrhea and hypernatremic dehydration [2]. A diagnosis may be made clinically with improvement after starting on a fructose-based formula. Early diagnosis and appropriate management can prevent life-threatening complications, including renal failure.

Case Presentation

The patient was a six-day-old baby girl, full-term, spontaneous vaginal delivery, birth weight 3.7 kg, with no history of neonatal ICU admission, regular antenatal follow-up, and a history of urinary tract infection during the last trimester. No treatment was administered.

There was a positive family history of consanguinity; there was no history of chronic diarrhea, renal disease, or metabolic disease. She has three healthy siblings. Immunization was up to age.

She was well on the first day, but on day 2 of life, she started to have a fever of 38°C with watery diarrhea, moderate in amount with no mucus or blood, 3-4 times per day. She was managed by Oral rehydration solution and was on exclusive breastfeeding. The next day, she showed improvement.

Then, after 1 day, the diarrhea started again with fever, decreased feeding, and lethargy. The family sought medical advice at the referring hospital, and the investigation discovered creatinine 600 mg/dL and urea 28 mg/dL. She received an IV fluid bolus and was referred to our hospital as a case of renal impairment. Upon examination, she looked unwell, lethargic, dehydrated, with mottled skin, and tachypneic. Other vital signs were normal, capillary refill time of 3 seconds.

She weighed 3.2 kg with a normal systemic examination. The impression was to rule out sepsis, congenital renal anomalies, or metabolic disease. She was given a normal saline bolus (20 ml/kg), kept NPO, and started on ampicillin and cefotaxime after obtaining bacterial cultures.

Laboratory tests showed metabolic acidosis (pH=7.14, pCO_2=16 mmHg, HCO_3=7.1 mmol/L, lactate=4.6 mmol/L), hyperglycemia (430 mg/dL), Urine dipstick (positive 3 glucose and positive 3 protein), hyperkalemia (6.7 mEq/L), hypernatremia (183.6 mEq/L), hypochloremia (146.3 mEq/L), and renal impairment (Creatinine of 837 mg/dL and BUN of 50 mg/dL). The bone profile, full blood count, and coagulation profile were within the normal range. She had a normal chest X-ray, an Echo, and a brain and abdomen ultrasound. After 14 hours of rehydration, she was still oliguric with metabolic acidosis, abnormal electrolytes, and renal impairment. She became edematous, and repeated chest X-rays showed lung congestion. An ultrasound of her abdomen showed mild ascites. Nephrology advised a Lasix challenge test and dialysis.

In the PICU, she was electively intubated and dialysis was started urgently; she was also receiving Lasix, sodium bicarbonate, salbutamol, and sodium polystyrene for hyperkalemia.

Several days later, the patient started to improve; metabolic acidosis resolved, blood sugar normalized, and electrolyte balance and renal function were improving. The septic workup was negative, so antibiotics were stopped.

As she improved, feeding was started gradually through a nasogastric tube with regular formula, and dialysis eventually stopped. On her second day in the regular ward, she started to have watery diarrhea again. Electrolyte balance and renal function were improving with sodium (170 mEq/L), urea (15.5 mg/dL), and creatinine (93.4 mg/dL). Dialysis was resumed in the PICU. The gastroenterologist's opinion was that the diarrhea was secondary to infection versus renal causes. After the patient showed a clear relationship between the reappearance of diarrhea and feeding two times between NPO and resumed feeding, the diagnosis was suspected to be GGM or allergy to cow's milk. She was started on a fructose-based formula (Galactomin 19). The patient improved dramatically after that, and the diarrhea stopped. Investigations were normalized after several days, and dialysis was stopped. The diagnosis of GGM was confirmed.

She was discharged in good condition on special formula and

Citation: Sheikh SB, Al-Rehaili K, Al-Harbi DL. Glucose Galactose Malabsorption in a Neonate Complicated by Renal Failure. J Pediatr Neonatol. 2023;4(2):1034.

Publisher Name: Medtext Publications LLC

Manuscript compiled: Aug 23rd, 2023

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regular follow-up and is gaining weight with normal development.

Discussion

GGM is a rare metabolic disease inherited in an autosomal recessive manner. The gene responsible is *SLC5A1*, located at chromosome 22q12.3, which encodes for Sodium-Glucose Cotransporter 1 (SGLT1) [2,3], which is present mainly in the intestinal brush border but may also be found in the heart and kidney [2].

Patients usually present in the first few days of life with intractable diarrhea and hypernatremic dehydration [2]. Our patient presented with a septic shock-like picture, hypernatremic dehydration, and acute kidney injury, which made the diagnosis at first seem like sepsis instead of a congenital renal anomaly.

When the relationship between diarrhea and feeding became apparent, GGM was suspected. The diagnosis was confirmed after improvement with a fructose-based formula.

A genetic study may be done to support the diagnosis [2], but it is not mandatory and was not done in our case.

Management is based on dietary restrictions on glucose and galactose. Complications associated with GGM may be serious and life-threatening, including renal failure [1], nephrolithiasis [4], nephrocalcinosis [4], renal tubular acidosis [5], heart failure [2], recurrent infection, and sepsis with opportunistic infections [6].

Renal failure and dialysis, as happened to our patient, are caused by severe dehydration. This complication was also reported by Al-lawama et al. [1] however, their case had irreversible renal failure.

Nephrolithiasis and nephrocalcinosis can present at the time of diagnosis or manifest later during follow up [4]. Nephrolithiasis is thought to be caused by chronic diarrhea, dehydration, and urine concentration [4]. Nephrocalcinosis is usually associated with hypercalcemia, hypercalciuria, and renal tubular defects [4]. Distal renal tubular acidosis is also reported in some cases and is associated with persistent metabolic acidosis [5].

Heart failure can happen because of severe acidosis, which leads to cardiac compromise and dysfunction [2].

High clinical suspicion leading to early diagnosis and appropriate treatment of this rare disease can prevent life-threatening complications.

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