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Case Report

First Clinical Case Report in the South Colombian Region, Newborn with Congenital Glucose-Galactose Malabsorption Syndrome (Sodium/Glucose Cotransporter Type 1 - SGLT1)

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Abstract

Congenital glucose galactose malabsorption syndrome presents with severe osmotic diarrhea of neonatal onset. It is a rare autosomal recessive disorder, consisting of damage to the glucose-coupled sodium cotransporter protein, responsible for its absorption in the enterocytes. We report the case of a previously healthy patient fed with exclusive breast milk, who debuted in the first week of life, with high-expenditure diarrhea, severe malnutrition, and hypernatremia; No response to handling with lactose-free, extensively hydrolyzed, and free amino acid formulas. Once the diagnosis was suspected, a therapeutic test with carbohydrate-free formula was performed, which confirmed the suspicion. During follow-up, the patient presented nutritional recovery and absence of diarrhea. This syndrome is the first case reported in the South Colombian region of our country, according to the literature review, requires a high level of clinical suspicion to perform the therapeutic test and confirm the diagnosis.

Objective: To describe the first clinical case report Newborn with Congenital Glucose-Galactose Malabsorption Syndrome (sodium/glucose cotransporter type 1 - SGLT1).

Keywords: Enteropathy; refractory diarrhea; newborn; colombia

Introduction

Refractory or intractable diarrhea of the neonate is one of the challenges presented to paediatric care due to the varied etiologies and complexities of its management. It presents with frequent stools, of soft consistency and lasting more than two weeks, which evolve in a refractory manner to the usual treatments [1]. Only 1% of patients hospitalized for prolonged diarrheoa evolve refractorily and it is believed that this clinical condition is related to profound changes in bowel function, often independent of the initial cause and the treatment performed. Most cases remain undiagnosed, and the mortality rate is high [2]. Galactose glucose malabsorption syndrome (CGGM, OMIM 606824) is a rare autosomal recessive disorder that presents with severe osmotic diarrhea of neonatal manifestation. It is caused by a defect in the sodium/glucose carrier type 1 (SGLT1) in the enterocyte, responsible for the absorption of these monosaccharides at the brush edge. More than 50 mutations related to the SLC5A1 gene located on chromosome 22q13.1.1

have been described. It is a potentially fatal disease without timely diagnosis and adequate treatment. Restriction of the two carbohydrates leads to complete resolution of symptoms and disappearance of risks related to malnutrition. The objective of this paper is to describe the first report in the South Colombian region, registered in the Neonatal Intensive Care Unit of Clínica UROS, Neiva, Colombia, in order to draw the attention of pediatricians and neonatologists to the importance of estimating this condition in the differential diagnosis of refractory diarrhea in infants and to develop a better management of suspected cases.

Case Presentation

Patient born at term 38 weeks, son of mother 24 years g3p3v3 with 4 prenatal controls, control start at 25 weeks. Born by vaginal delivery presents spontaneous neonatal adaptation Apgar 8-9/10 Silverman Anderson 0 points without respiratory distress, maternal and newborn blood group O positive. Storch study: Treponemal test negative, HIV negative, Agshb negative, Toxoplasma Igm of 3.34 positive for which he received treatment with spiramycin for two months as gestational toxoplasmosis, birth weight of 3 570 grams. It is discharged for good evolution and is fed with exclusive breastfeeding. He entered our institution Clínica UROS in the city of Neiva, Huila, Colombia, referred from a second level hospital as a vital emergency due to acute renal failure secondary to dehydration with indication of dialytic urgency. The mother reports a clinical picture of 4 days of evolution consisting of abundant liquid diarrheal stools with mucus, without yellowish and greenish blood approximately 7 times a day, without vomiting, without fever. Denies respiratory symptoms. The mother reports that she only fed him with breast milk, denies having administered another type of feeding or drink. Consultation to second level hospital for persistence of uncontrollable diarrhea, laboratories are taken that evidence metabolic acidosis with sodium of 192 and creatinine report of 2.9 for which they remit.

A 2-month-old patient who, beginning the second week of life, begins with watery diarrhea of very high expenditure, which leads to severe dehydration, shock, and acute renal failure. In this NICU for about 4 weeks, drawing attention to metabolic acidosis and persistent hypernatremia, on the other hand, high expenditure is confirmed by feces, not by urine, improvement of fecal expenditure with fasting, and no response to hypoallergenic formula. Allergy to cow's milk proteins is ruled out. The diarrhea has been severe, watery, of very high expenditure, it is confirmed that it is osmotic, which has led to severe dehydration, hypovolemic shock, hypernatremia, and malnutrition. Currently with cyclical parenteral nutrition, in hemodynamic compensation, with adequate weight gain, and controlled fecal expenses. A formula based on cow's milk protein, with fructose, without glucose or galactose, was started, with adequate clinical response, which frames a high diagnostic suspicion of malabsorption of glucose - galactose, due to alteration of the sglt-1 transporter, for which assessment is requested by the pediatric gastroenterology service who considers intractable congenital diarrhea, Enteropathy due to severe PLV was ruled out, osmotic diarrhea was confirmed, diagnosis was confirmed, congenital glucose-galactose malabsorption (sodium / glucose cotransporter type 1 - SGLT1). It started, galactomin-19.

It is formula based on cow's milk protein, but free of glucose and galactose, as a source of HCO, fructose, this formula began in terms of diagnostic and therapeutic test, and in this way establish medium and long-term plan; due to the non-availability of this formula in the market and in Colombia, it would begin towards the fourth month of age, artisanal chicken formula. Currently patient in stable conditions, hemodynamically compensated in cardiorespiratory sufficiency, with adequate respiratory pattern and saturations, without supplementary O2 requirements, last gas control with acid-base balance with adequate oxygenation and ventilation with electrolytes and normal lactic acid. At the hemodynamic level, with adequate distal perfusion, without bradycardias, without inotropic support, echocardiogram was taken without hemodynamic repercussion. At the infectious level, without dysthermias with report of blood culture, urine culture, negative stool cultures, now without antibiotics. Report of laboratories blood count within normal limits, with TGO 47 with TGP of 19 normal. Electrolytes with sodium of 137 potassium of 3.6 normal. 3 indirect total bilirubin of 2.8 delta of 0.2, within normal limits. At the active neurological level, reactive, without alterations or neurological risk. Currently patients evolve in stable conditions, progressive weight gain with adequate tolerance to galactomin-19 is expected to start towards the fourth month of age, artisanal chicken formula.

Discussion

Chronic diarrhea of neonatal manifestation is a pathology of low frequency, and its etiology includes a significant number of entities that have in common a very severe clinical presentation, rapid nutritional deterioration, and associated complications. The most frequent hydro electrolyte alteration in high-expenditure diarrhea is hyponatremia. The finding of hypernatremia requires the study of its origin and identify the two pathophysiological mechanisms that produce it: the first, by increase in total body sodium, usually accidental or iatrogenic, which is not the case of the patient; the second, by the decrease in total body water that can be due to low intake, renal losses (diabetes insipidus), gastrointestinal, or through the skin [4]. The initial approach of a patient such as the one presented is to determine whether the diarrhea is osmotic or secretory: if fasting decreases fecal expenditure significantly, it is assumed that the mechanism is osmotic.

Diagnostic possibilities in a neonate with profuse diarrhoea include microvillous congenital atrophy, congenital chlorhydrorhea, intestinal epithelial dysplasia enteropathy, congenital chlorine and sodium malabsorption, bile acid malabsorption, and enterokinase deficiency, which do not present with hypernatremia; Congenital lactase deficiency, which could explain the clinical picture, is ruled out by not improving with lactose-free formula [5]. The CMGG was first described in the sixties by different authors. It manifests as severe watery diarrhea of neonatal onset leading to severe dehydration, in most cases hypernatremic, improves with fasting,

and disappears upon discontinuation of glucose and galactose from feeding [6]. These patients adequately hydrolyse the disaccharides (lactose, sucrose, and maltose) since the defect does not lie in the processes of enzymatic digestion but in absorption [7]. The SGLT1 protein is responsible for the absorption of these monosaccharides by cotrasporte mechanism at the level of the enterocyte.

It uses the sodium electrical gradient generated by the sodium potassium ATPase pump to enter glucose and galactose into the cell. The alteration of the protein causes these monosaccharides to remain in the intestinal lumen, increase osmolarity, cause net loss of water and generate hypernatremia. Sugars are fermented by the microbiota at the level of the colon producing short-chain fatty acids that lower the pH in the feces and generate metabolic acidosis when absorbed [8]. Another function of SGLT1 also altered is to absorb small polar molecules, including water, which contributes to hypernatremia [9]. Unlike glucose and galactose, fructose is independently absorbed via the GLUT5 carrier [10]. With diagnostic suspicion, therapeutic testing with a glucose- and galactose-free formula is indicated; Currently only two pharmaceutical companies have these nutritionally complete and adequate formulas for these patients. The addition of fructose in the formula as the only carbohydrate, without causing diarrhea confirms the diagnosis which occurred in our patient after the start of the formula.

Conclusion

MGG is an exceptional disease, causing severe and early diarrhea at neonatal debut. The diet with fructose contributes to the disappearance of symptoms and maintains an almost normal growth of the affected child. Despite its scarcity, MGG should be known because the diagnosis is easy and has a good prognosis.

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