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When Milk Turns Bitter: A Rare Tale of Congenital Lactose Intolerance

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Abstract

Congenital lactose intolerance is a rare autosomal recessive disorder characterized by complete or near-complete absence of lactase enzyme activity from birth. Affected infants present with severe osmotic diarrhea soon after initiation of lactose-containing feeds. Early diagnosis is essential to prevent dehydration, electrolyte imbalance, and growth failure. We report a 6.5-month-old male infant presenting with chronic watery diarrhea since birth, who showed dramatic clinical improvement following initiation of a lactose-free diet. Reappearance of symptoms after reintroduction of lactose further supported the diagnosis. This case highlights the importance of considering congenital lactose intolerance in infants with persistent diarrhea since birth and emphasizes the diagnostic and therapeutic role of lactose withdrawal.

Keywords: ***Lactose intolerance; Congenital lactose intolerance; Small intestine; Diarrhea***

1. Introduction

Congenital lactose intolerance is an extremely rare inherited disorder resulting from deficiency or absence of lactase-phlorizin hydrolase activity in the brush border of the small intestine. Lactase is responsible for hydrolysis of lactose into glucose and galactose, facilitating intestinal absorption. In affected infants, ingestion of lactose-containing feeds leads to accumulation of undigested lactose in the intestinal lumen, causing osmotic diarrhea, abdominal distension, dehydration, and failure to thrive.

Unlike primary lactase deficiency, which develops later in life, congenital lactose intolerance presents soon after birth with severe diarrhea upon exposure to breast milk or formula feeds. Prompt recognition and dietary modification are critical to prevent life-threatening complications [1,2].

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2. Case Presentation

A 6.5-month-old male infant, born to a non-consanguineous couple, was delivered at term via lower segment cesarean section with a birth weight of 2.1 kg. There was no significant antenatal or perinatal history. The child was appropriately immunized for age.

The infant presented with chronic watery yellowish diarrhea since birth, occurring approximately 20-25 times per day. He had been exclusively breastfed, with no history of pre-lacteal feeds. There was no history of vomiting, blood in stool, fever, or evidence suggestive of superimposed infection.

On examination, the child was developmentally appropriate for age with adequate weight gain and no signs of dehydration. Perianal excoriation was present.

Laboratory investigations revealed mild anemia with normal electrolyte levels. Stool examination showed acidic pH with absence of reducing substances. In view of persistent diarrhea since birth and poor response to medications, carbohydrate malabsorption was suspected.

The child was started on complementary feeds along with a lactose-free formula, following which there was abrupt resolution of diarrhea. A subsequent trial of lactose-containing diet resulted in recurrence of symptoms. The child was discharged on a lactose-free diet and followed up after one month, during which there was complete resolution of diarrhea and satisfactory clinical improvement.

Based on the characteristic clinical history and dramatic response to lactose withdrawal, a diagnosis of congenital lactose intolerance was made.

3. Discussion

Congenital lactose intolerance is a rare autosomal recessive disorder caused by mutations affecting lactase enzyme production. The disease typically manifests during the neonatal period with severe osmotic diarrhea following initiation of milk feeds. Persistent diarrhea can rapidly lead to dehydration, electrolyte disturbances, metabolic acidosis, and malnutrition if left untreated.

The diagnosis is primarily clinical and is supported by stool acidity, presence of reducing substances, and marked improvement after lactose elimination. In resource-limited settings, therapeutic dietary trials remain an important diagnostic tool where genetic testing and intestinal biopsy may not be readily available.

Differential diagnoses include glucose-galactose malabsorption, cow’s milk protein allergy, congenital chloride diarrhea, congenital sodium diarrhea, and infectious causes of chronic diarrhea. The absence of systemic illness, persistence of symptoms exclusively with lactose exposure, and dramatic improvement after lactose withdrawal favored congenital lactose intolerance in the present case [3,4].

4. Management

Management consists of complete elimination of lactose from the diet using lactose-free formulas and ensuring adequate nutritional supplementation, particularly calcium and vitamin D. Early intervention allows normal growth and development and prevents complications.

5. Conclusion

Congenital lactose intolerance, though rare, should be considered in infants presenting with chronic diarrhea since birth, particularly when symptoms are associated with milk feeds. Early recognition and initiation of a lactose-free diet can lead to rapid symptom resolution and prevent severe complications. Lactose withdrawal remains both a diagnostic and therapeutic intervention in such cases.

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