TITLE

Congenital glucose- galactose malabsorption in a male infant

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OBJECTIVES

Congenital glucose-galactose malabsorbtion is a rare autosomal recessive disorder of intestinal transport of glucose and galactose. It is characterized by watery diarrhea, dehydration, failure to thrive, or early death without appropriate dietary treatment.

METHODS

The patient was 15 days old when he was admitted to the hospital because of continued, severe, watery, acidic diarrhea and hypernatremic dehydration. The abnormal stool looses were recorded within 4 days of birth. They were followed by abdominal distension, with no vomiting, and persistent, osmotic, watery diarrhea for the next two months. Despite management with lactose-free semielemental formula, and periodic administration of total parenteral nutrition during hospitalization, severe malnutrition occured.

RESULTS

Laboratory investigations revealed repeated low blood sugar levels, slight intermitent glycosuria, low stool Ph, and presence of reducing substances in the feces. Oral glucose tolerance test showed flat blood glucose response. Diagnostic evaluation ruled out infectious etiology of the diarrhea, cystic fibrosis, familial chloride diarrhea, and lactose intolerance. The X-ray examination of the intestinal tract revealed no abnormality. The clinical history of the patient and performed laboratory investigations were stongly suggestive of congenital glucose-galactose malabsorption. Dramatic ceasure of the diarrhea followed when the patient was treated with a commercial glucose and galactose-free formula – Galctomine 19 (specialized fructose-based formula).

CONCLUSIONS

All these findings and further succsessful, sustained weight gain, established the diagnosis of congenital glucose-galactose malabsorption in our patient. At the age of 12 years he had normal growth and neurological development.

REFERENCES

Turk, E., Zabel, B., Mundlos, S., Dyer, J., Wright, E. M. **Glucose/galactose malabsorption caused by a defect in the Na(+)/glucose cotransporter.**Nature 350: 354-356, 1991. [PubMed: 2008213, related citations] [Full Text]
12.
Wright, E. M., Martin, M. G., Turk, E. **Familial glucose-galactose malabsorption and hereditary renal glycosuria.In: Scriver, C. R.; Beaudet, A. L.; Sly, W. S.; Valle, D. (eds.) : The Metabolic and Molecular Bases of Inherited Disease. Vol. III.** New York: McGraw-Hill (8th ed.) : 2001. Pp. 4891-4908.
Scheepers A, Joost HG, Schurmann A. The glucose transporter families SGLT and GLUT: molecular basis of normal and aberrant function. JPEN J Parenter Enteral Nutr. 2004;28:364-71.
Stelzner M, Chen DC. To make a new intestinal mucosa. Rejuvenation Res.2006;9:20-25.

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