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CONGENITAL GLUCOSE-GALACTOSE MALABSORPTION : CASE REPORT

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The first dignosed case of congenital glucose-galactose malabsorption in The Republic of Macedonia has been presented.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.Further laboratory investigations revealed repeted 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).All these findings and further succsessful, sustained weight gain, established the diagnosis of congenital glucose-galactose malabsorption in our patient. At the age of two years he had normal growth and neurological development.