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A Case of eosinophilic gastroenteritis in a Patient with glycogen storage disease type 1a

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Abstract

We present on the case of a pediatric patient with glycogen storage disease type Ia (GSD1a) who was previously well controlled on routine management including frequent cornstarch regimen. Following multiple admissions for hypoglycemia and related symptoms, an esophagogastroduodenoscopy revealed eosinophilic gastroenteritis (EGE) which had resulted in small bowel inflammation and malabsorption of cornstarch. After an extensive work-up, he was found to be sensitive to multiple food proteins, including corn. His symptoms improved after a brief course of corticosteroid, a multiple-food elimination diet, and replacement of cornstarch with tapioca starch. In patients with GSD, slow digestion and absorption of cornstarch is essential to maintaining blood glucose levels. In patients with EGE, mucosal injury results in poor absorption of nutrition. When the two occur together, EGE has an uncharacteristically acute presentation and GSDIa is difficult to treat with adjustments of cornstarch dose.

Keywords

vonGierke disease; GSD1a; esophagitis gastroenteritis; hypoglycemia; lactic acidosis

Introduction

First described by von Gierke in 1929, Glycogen Storage Disease Type I (GSDI) is an inherited, autosomal recessive metabolic disorder. It is caused by defects in the glucose-6-phosphatase (G6PC) system located in the liver and kidneys. Both Glycogen Storage Disease Type Ia (GSDIa) and Glycogen Storage Disease Type Ib (GSDIb) prevent the hydrolysis of glucose-6-phosphate to glucose leading to disruption of the last enzymatic step of both glycogenolysis and gluconeogenesis. This disruption can be clinically manifested as severe fasting ketotic hypoglycemia, lactic acidosis, hyperlipidemia, hyperuricemia, transaminitis, and hepatomegaly all of which can be seen at diagnosis which is usually around 6 months of age, when the fasting period starts to increase [1,2].

Case Presentation

GSD1a and GSD1b share similar presentations and clinical features, but the defect is at different levels of glucose-6-phosphateprocessing. In GSD1a, the original case described by von Gierke, the glucose-6-phosphatase is deficient, therefore limiting the hydrolysis of glucose-6-phosphate to glucose.

In GSD1b, the glucose-6-phosphate transporter is deficient, resulting in a limitation of glucose-6-phosphate available for hydrolysis [1,3]. Long-term complications, such hepatocellular adenomas and carcinomas, nephrocalcinosis, renal failure, growth failure, and osteoporosis can be seen with both. The transporter affected in GSDIb is also thought to participate in neutrophil differentiation and immune modulation. As a result, neutropenia with frequent infections are part of the phenotype, and an increase in immune mediated conditions such as an inflammatory bowel disease, similar to Crohn's Disease, are only seen in GSDIb [1,2,3].

Dietary management of GSDI focuses on avoiding hypoglycemia with frequent meals, continuous nocturnal nasogastric feedings in infancy, and later the addition of complex, slowly absorbed carbohydrates in the form of cornstarch. Fructose and galactose are restricted to minimize hyperlactacidemia [1,4].

Eosinophilic gastroenteritis (EGE) is characterized by gastrointestinal symptoms associated with an immune-mediated response in the gastrointestinal tract to various proteins found in food [5,6]. It is a mixed immune response as eosinophils are seen throughout the GI tract, but the inflammation is due mainly to a T-cell mediated response despite the predominant eosinophilic involvement. The gastrointestinal eosinophilia is not associated with parasitic infection or other systemic processes. Symptoms include abdominal pain, nausea, vomiting, diarrhea, dysphagia, and weight loss. In exceptional cases, intestinal obstruction and perforation can also be seen. Protein-losing enteropathy, bleeding, and malabsorption are common. These symptoms are generally chronic and insidious in onset with only rare acute presentations. Treatment includes corticosteroids and dietary interventions such as food elimination and reintroduction, with attempts to identify the causative foods. Steroid-sparing treatments (sodium cromoglycate, ketotifen, monetleukast) have been used with mixed success [5,6].

Case Report

The patient was an 8-year-old with a history of GSD1a (glucose-6-phosphatase deficiency, von Gierke disease MIM:232200). Prior to presentation, he was adequately maintained on a sucrose, fructose, and galactose-restricted diet, along with frequent cornstarch feedings every four hours. Over a period of 6 weeks, he was admitted multiple times for episodes of acute deterioration, frequent vomiting, hypoglycemia, and lactic acidosis. After receiving continuous dextrose-containing crystalloid solution, the patient would improve, only to return days later with comparable metabolic derangements. His cornstarch dose was increased after each hospitalization. Physical exam was only remarkable for hepatomegaly, with the liver 7 cm bellow the costal margin, and laboratory abnormalities during hospitalization were significant for ketotic hypoglycemia, lactic acidosis, hypertrygliceredemia, hyperuricemia, and a gradually increasing eosinophil count from 800, at his initial presentation, to a peak of 5,800.

Pediatric gastroenterology was consulted when the patient developed abdominal pain. Endoscopy was performed. The duodenum was grossly normal (Figure A), but histology revealed eosinophilic duodenitis (eosinophils >100 per HPF, Figure B) as well as eosinophilic esophagitis (>40 per HPF) and eosinophilic gastritis (>30 per HPF, Figure C). Stool studies were negative for infectious etiologies. Radioallergosorbent testing (RAST) revealed allergen-specific IgE to corn, egg white, peanut, soybean, and wheat; while skin allergy testing showed a strong reaction to corn and weak reactions to

soybean and rice. The patient was placed on a 2 month steroid taper with dietary modification consisting of elimination of corn, soy, fish, wheat, and egg and replacement of cornstarch with tapioca starch every three hours. Repeat endoscopy showed resolution of eosinophilia, and most recent peripheral eosinophilic count was 200. The patient has remained in remission for more than 24 months, and is currently on a corn-free diet. Wheat and fish were reintroduced with no problem.

Discussion

EGE in the setting of GSDIa has not been described in the literature to date. The mucosal involvement and subsequent malabsorption in EGE, in this case led to poor absorption of glucose from cornstarch, resulting in recurrent episodes of hypoglycemia and severe lactic acidosis, as is typically seen in poorly controlled GSDIa. Further complicating our case was corn acting as a contributing allergen. While minimal, commercially available cornstarch has a protein content of 0.3 g per 128 g of cornstarch, which explains the allergic reaction seen in our patient. Dietary interventions were effective in this case with cornstarch included in the elimination diet due the large quantities of cornstarch needed on a daily basis. Corn starch was changed to tapioca starch which has trace protein content, and a glucose content of 87 g per 100 g, compared to 93gr of glucose per 100 gr seen in corn starch [7]. Tapioca starch facilitated glucose control of our patient, but he required higher and more frequent doses of tapioca starch due to the lower carbohydrate content compared to corn starch. Elimination of corn and other allergens helped with improvement of symptoms of EGE, specially the recurrent episodes of hypoglycemia, explained by poor carbohydrate absorption. In our cohort of patients with GSD, we also have another patient with GSD type0 who developed EGE and corn allergy, and is also doing well with tapioca starch.

Identification of corn as a main allergen had important treatment implications for our patient. Currently, an artificial corn- based, long acting form of starch is a globular molecule that has been use with success in treatment of patients with GSD1a after the age of 5 [8]. It is an alternative treatment for patients with GSD1a allowing the increase in the fasting period at night time (6-8 hours), impacting positively their quality of life. Our patient was in the age range when this artificial corn starch is indicated, but due to his proven corn food allergy, he was not a candidate for it. Because of this, his treatment required continuation of tapioca starch, with fasting periods no longer than three hours at night, even shorter than those seen for regular corn starch.

Compared to GSD1b, patients with GSD1a do not commonly have GI problems. In our patient, the first presenting sign of poor glucose malabsorption, due to increase mucosal inflammation, was increased frequency of hypoglycemic episodes despite adequate compliance to his corn starch regimen. In contrast, in patients with GSD1b, an increase corn starch dose, associated or not with GI symptoms, suggests the possibility of GSD-associated inflammatory bowel disease (GSD-IBD), which is a relatively common feature of this condition.

As illustrated by our case, recurrent episodes of hypoglycemia in patients with GSD1a, in the face of previously good control with a corn starch regimen, should alert the clinician to the possibility of carbohydrate malabsorption, requiring further investigation. EGE was the etiology for the hypoglycemic episodes in our patient, but additional GI causes in other cases could be contemplated, underscoring the need for a thorough gastrointestinal evaluation.

Figures

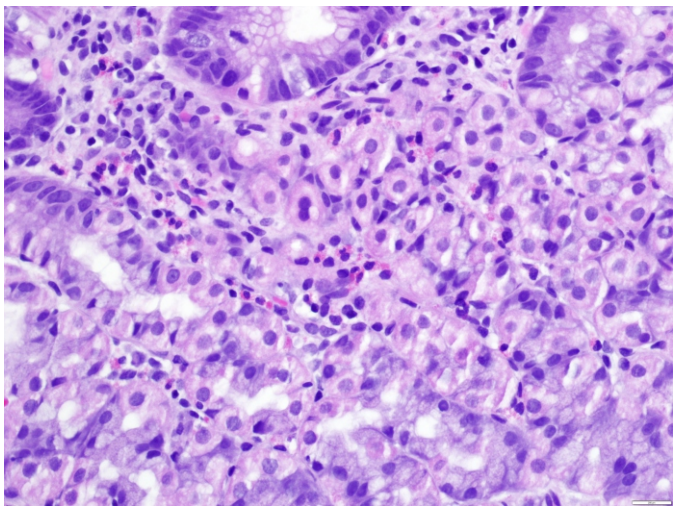

Figure 1

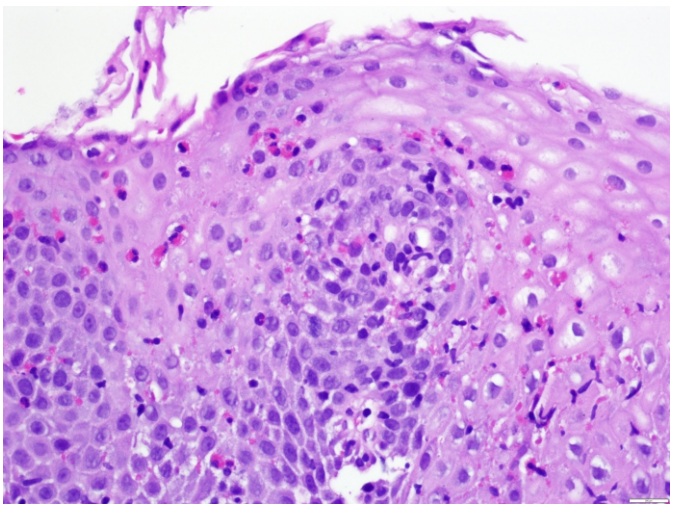

Figure 2

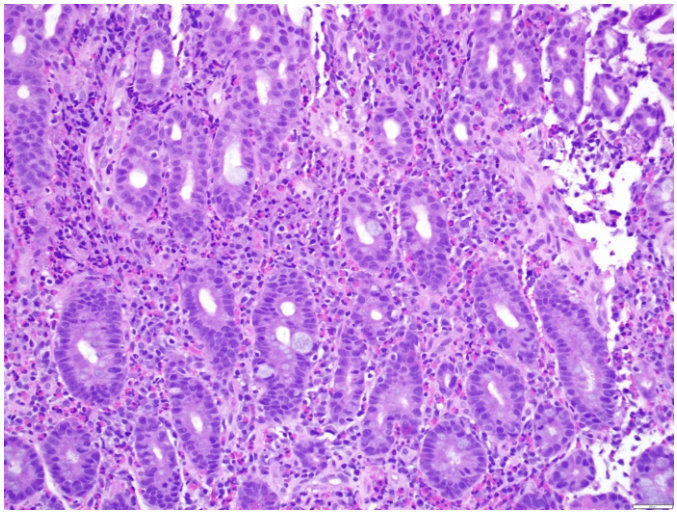

Figure 3

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