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CASE REPORTS

Difficulties in Celiac Disease Diagnosis in Children – A case report

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ABSTRACT

Diagnosis of celiac disease in a patient with lactose intolerance has special importance having implications for the treatment of both diseases. The authors present the case of a 2 years old girl, first diagnosed with enterocolitis, but her clinical evolution revealed a complex situation: both celiac disease and secondary lactose intolerance. We present the case as a special situation in clinical pediatric practice that must be taken into account more often.

Keywords: celiac disease, lactose intolerance, serology

INTRODUCTION

Celiac disease and lactose intolerance are relatively common diseases (1), with symptoms occurring after ingestion of certain food components. In celiac disease, gluten, a protein storied in wheat, rye or barley, ingested induce intestinal inflammatory disease in susceptible individuals (2), causing gastrointestinal (diarrhea, weight loss, abdominal pain, anorexia, lactose intolerance, abdominal distension, irritability) and extra intestinal symptoms (e.g. anemia, dermatitis herpetiformis, chronic fatigue (3). Diagnostic criteria include serological tests (e.g. endomysial antibodies – EmA-IgA), characteristic small intestinal histology (lymphocytes infiltration, villous atrophy) and genetic testing, e.g., determining human histocompatibility antigens HLA DQ2/DQ8. In

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current medical practice, the first two are more common. Therapy is a strict gluten free diet throughout life.

A child's classic signs of celiac disease are abdominal pain, diarrhea or constipation, vomiting, flatulence, and regurgitation. Recent studies show that these clinical signs may be accompanied by an imbalance of the neurovegetative system with a predominantly sympathetic tone that prevails throughout the diet without gluten (4). ❑

CASE REPORT

RC, 2 years old girl, has a first hospitalization (April 2010) for diarrheic stool and intermittent vomiting, symptoms of insidious onset several weeks ago. Patient from the burden of physiological development was born at nine months, with weight 3100g, height 51cm, Apgar score 10.

Personal pathological histories were insignificant.

The patient had a natural diet for 6 months, and then properly diversified food during the next six months and ablactate at 9 months. It was diagnosed as diarrhea disease and followed a diet that excluded milk. Normalization of the stools was achieved. At home, gradually milk diet was reintroduced, with favorable evolution. Two weeks later (May 2010), mother noticed recurrence of vomiting, for each meal containing milk or cheese.

Physical examination at admission noticed a relatively good general condition, poor nutritional status, slightly pale skin, adipose tissue diminished at all levels, cardiovascular and respiratory balanced, increased abdominal volume which was meteoritic, umbilical hernia point.

Her appetite is capricious, and is manifesting abdominal pain and weight loss. Curve weight was decreasing: in February 2010 weighed 11.6 kg, in April 2010 -10 kg and hospitalization 9.8 kg (5th percentile), size of 87 cm (60th percentile) and body mass index 13kg/m^2.

Laboratory findings revealed Hb 12 g / dl, MCV 78.2 fL, MCHC 29.6 g / dl, Fe 18 mg / dl, AST 58 U / l, ALT 27 U / l, CRP 0.4 mg / dl. Stool examination for digestion showed muscle fibers partially digested and rarely intracellular starch.

Positive diagnosis

Clinical signs and symptoms are not enough for a diagnosis of lactose intolerance. Many other conditions including gastroenteritis and irritable bowel syndrome can give similar symptoms. In infants, diarrhea can be a sign of allergy to milk proteins, together with eczema, irritability and weight gain deficit. Therefore, to confirm the diagnosis, a lactose load test was performed (specificity 96%), which presented a payment curve (Figure 1).

Abdominal ultrasound revealed increased peristalsis, more relaxed bowel, filled with liquid, without wall thickening. Liver and spleen had no pathological changes.

Lactose intolerance can sometimes be transient, due to diseases that damage the small intestine casing, with lactase, the most common reason being celiac disease.

Celiac disease screening was performed with EmA-IgA determination, in indirect immunofluorescence on monkey esophagus, which were positive, but that requires confirmation with a biopsy duodenal. For this reason upper digestive endoscopy was performed. This presented the esophagus, stomach, duodenal bulb and duodenal D2 of normal endoscopic aspect. Therefore, at histopathology of D2 we found: examined material consists of two pieces of duodenal mucosa poorly targeted; villous not appear at all the material, surface epithelium was only focally presented. At this level there is a pathological level exocytosis; among crypts (cuts occurring across) can be observed an intense lymphoplasmacytic infiltrate, containing eosinophils. Cryptic exocytosis was normal. Histopathological examination has concluded: the described appearance does not exclude celiac disease but may be present in other enteropathic pathologies.

The final diagnosis was made: celiac disease (diagnosis supported by EmA-IgA positivity but with mention to repeat EmA-IgA, intestinal biopsy and HLA DQ2/DQ8 typing to the next medical examination), secondary lactose intolerance, protein-energy malnutrition, hypochromic microcytic anemia. ❑

TREATMENT

It was recommended: (a) exclusion from food preparations based on gluten and lactose-free milk consumption, (b) medication: Liv52 2x5

ml / day for 6 weeks. Evolution was favorable: has not presented vomiting, abdomen was reduced in volume, normalized intestinal transit and weight became upward curve. ❑

DISCUSSIONS

In this circumstances we had to deal with two different diagnoses attitudes, namely: because cryptic exocytose was normal, histopathology was not relevant to celiac disease and thus to support diagnosed celiac disease, duodenal biopsy should be repeated at 6 months and 12 months or it could bring faster gluten free treatment and support celiac disease diagnosis based only on EmA-IgA positivity. These two different attitudes raise many discussions in celiac disease diagnosis.

Duodenal biopsy on the American continent remains the gold standard in diagnosing celiac disease, according to North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (5), protocol, which also joined the European Society of Pediatric Gastroenterology, Hepatology and Nutrition (6). This view is found in the present in a group of American researchers, Parizide et al. (7); which argue that serological screening tests are used primarily to identify those who need a duodenal biopsy as a diagnostic test. Due to the complexity of this disease, Catassi et al. (8) recently proposed a new diagnostic algorithm, which could confirm diagnosis of celiac disease if at least 4 of the following 5 criteria are positive: 1) typical symptoms of celiac disease, 2) serological positivity of immunoglobulin classes, 3) a class of celiac disease-specific antibodies at a high titer, 4) human leukocyte antigens DQ2 or DQ8, 5) diagnosis of celiac disease by the biopsy of the small intestinal mucosa and the response to a gluten-free diet.

However, the need for rapid diagnosis of celiac disease by avoiding intestinal biopsy and the benefits of the introduction of gluten-free diet persists in the current studies (9-11), studies that are too numerous to go unnoticed. In Europe, in 2005, stands a group of Finnish researchers, Collin et al. (12), that in a multicenter European study, argue that in view of the high percentage of poor histological changes duodenal biopsy, celiac disease diagnosis should not depend only on biopsy. Their opinion was kept constant until today. Kaukinen et al. (13), noticed that celiac disease occurs before the appearance of pathological changes in villous duodenal biopsy and increased levels of EmA-IgA in a patient with normal intestinal villi guide us towards developing celiac disease. Otherwise, EmA-IgA is a predictive marker of celiac disease occurrence. In another prospective study published in 2010 in "The Journal of Pediatrics", Kurppa et al. (14), from Tampere University in Finland, the Pediatric Research Center, recommends a reassessment of diagnostic criteria for celiac disease only from the serology presence, because children who are positive for EmA-IgA can get early treatment. Mustalahti et al. (15) conducted a study of 29,212 participants in four European countries (Finland, Germany, Italy and UK) and recommended performing intestinal biopsy only in those patients positive for serology testing. ❑

CONCLUSIONS

This case report brings into question the importance of rapid and certain diagnosis in celiac disease associations, in this case lactose intolerance, given by serological tests compared with duodenal biopsy, a more invasive, children sometimes unpleasant, hard accepted by the family especially when it is necessary to repeat the maneuver and long waiting time results.

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