



Case Report

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Celiac disease in an obese child with down syndrome

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ABSTRACT

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Patients with Down syndrome are predisposed to autoimmune disease and have a higher risk for type 1 diabetes, autoimmune thyroid disorders, alopecia, vitiligo, hypoparathyroidism and celiac disease. Diarrhea and malabsorption represent the typical presentation of celiac disease, while obesity or weight gain are uncommon in young children. We report an 8-year-old obese girl with Down syndrome who was diagnosed as celiac disease in the absence of signs and symptoms. We would suggest that even in absence of signs and symptoms, the children with Down syndrome should be screened annually for celiac disease even owing to obesity.

Keywords:

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1. Introduction

The prevalence of autoimmune diseases such as autoimmune thyroiditis, type 1 diabetes mellitus, and celiac disease is higher in patients with Down syndrome than in the general population (Kinik et al., 2006). Many studies originating from different centers have reported a prevalence of celiac disease in Down syndrome ranging from 4.6%-13% (Book et al., 2001). It is estimated that 6.3% of children with Down syndrome in Turkey have celiac disease (Cogulu et al., 2003). George et al. (1996) found a frequency of 7.0% of celiac disease in 115 screened children with Down syndrome. Recently a prevalence of 17% of celiac

disease in Swedish children with Down syndrome was reported (Jansson and Johansson, 1995).

The clinical presentation of celiac disease in children is very variable and differs with age. Diarrhea and malabsorption represent the typical presentation of celiac disease in young children, while abdominal pain, vomiting, and constipation are atypical gastrointestinal symptoms more common in older children and teenagers. Recent reports have been published that there were children with overweight/obesity at the time of celiac disease diagnosis (Balamtekin et al., 2001; Arslan et al., 2009).

However, there is no report as complaint with

obesity in a case with Down syndrome who was diagnosed as celiac disease.

2. Case report

An 8-year-old girl with Down syndrome was admitted to our Pediatric Endocrinology Unit owing to obesity. She also had constipation, abdominal pain, swollen abdomen and weight loss (4 kg) for two weeks. Her medical history revealed an uneventful birth history and normal cardiac function. However, she has a history of general developmental delay and truncal hypotonia with brisk reflexes and a poor gross motor coordination.

Physically, she was obese and had mild hypotonia and a marked dysmorphic face appearance. The patient weighed 32 kg (90th percentile) and measured 120 cm in height (10th percentile). Body mass index was calculated as 22.6 kg/m² (>95th percentile, Body mass index-standard deviation score: 1.99). She weighed rapidly (6 kg) in last year. Systolic and diastolic blood pressures were 90 mmHg and 60 mmHg, respectively. She had phenotypic features of Down syndrome and in addition she had a swollen abdomen. Her early developmental milestones were delayed. Other systemic examinations were normal. Laboratory studies including erythrocyte sedimentation rate, electrolytes, liver and renal functions were all within normal limits. She had mild iron deficiency anemia (hemoglobin 11.5g/dL, MCV 65 fL, ferritin 5.7 ng/L) and treated with oral iron supplements (ferrous sulfate 6 mg/kg twice in a day). In addition, due to malabsorption of vitamin B12 and folic acid levels were revealed as 261 pmol/L and 8 nmol/L which were within normal limits. Metabolic screening, thyroid function tests, and glucose metabolism were normal. We screened with various autoimmune disorders, and celiac disease screening by tissue transglutaminase antibodies (IgA) were found to be positive. Duodenal biopsy showed focal villous blunting and atrophy with mildly expanded lamina propria with lymphocytes and plasma cells, consistent with celiac disease. Biopsy for celiac disease was found

to be compatible with (Marshall Classification Modified Tip 3b) (Fig.1). The case was treated with gluten-free diet for 6 months, and lifestyle changes were advised, including body weight control with diet modification to reduce their intake of sodium, refined sugars, and fat. Her complaints have disappeared completely after institution of a gluten-free diet. No other signs or symptoms of the other autoimmune disorders have appeared to date.

3. Discussion

In literature, there are no case reports describing the co-occurrence of celiac disease and obesity in Down syndrome patients. Down syndrome may be associated with various autoimmune disorders and may present at any age (Kinik et al., 2006). The reason for the high prevalence of celiac disease in Down syndrome patients is not completely known.

Celiac disease might present in an atypical manner with a predominance of extra-intestinal manifestations such as anemia, short stature, recurrent stomatitis, infertility, Graves' disease, type 1 diabetes mellitus, and various musculoskeletal signs and symptoms. Interestingly, many reports indicate that celiac disease can be associated with overweight or normal weight; hence, malnutrition is not always present at celiac disease presentation. Therefore, celiac disease and obesity can coexist during both childhood and adolescence (Balamtekin et al., 2001; Arslan et al., 2009). However, the pathogenesis and clinical implications of the coexistence of classic malabsorption (e.g., celiac disease) and overweight/obesity remain unclear. The first published report described a 5-year-old girl with obesity, short stature, and recurrent abdominal pain (Conti et al., 1987). Venkatasubramani et al report 5% of patients had a body mass index >95th percentile among 143 patients with celiac disease diagnosed between 1986 and 2003 (Venkatasubramani et al., 2010). In celiac disease patients, atrophy determines the loss of normal intestinal function. This

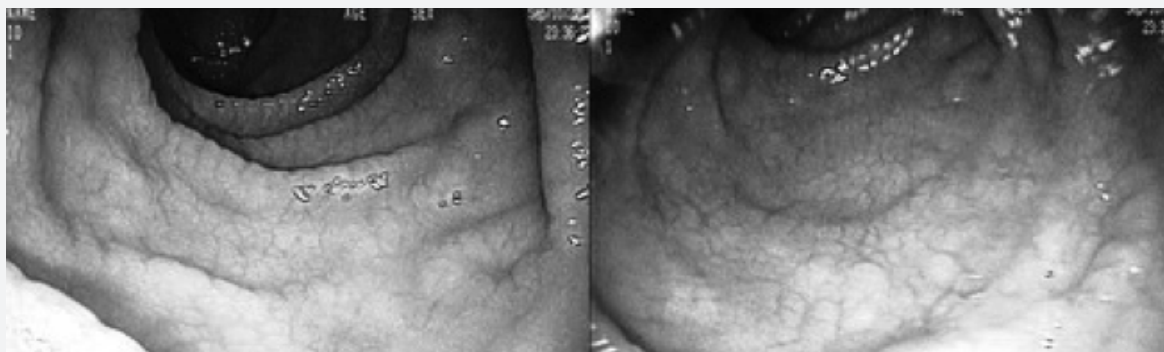


Fig. 1. Endoscopic markers of celiac disease in the descending duodenum (scalloping, mosaic pattern, and decrease in number or height of duodenal folds)

can hypothetically induce increased absorption of the functionally preserved intestinal tract. If this process, it could lead to the extraction of energy exceeding the child's needs, thus increasing the risk of overweight/obesity (Semeraro et al., 1986). Among the obese patients, the most common symptoms at onset were abdominal pain, diabetes, and diarrhea. Similarly, the presented patient was obese, however; in last days she had some intestinal complaints suggesting celiac

disease. However; if she has not diagnosed as Down syndrome, her antibodies have not screened for celiac disease because of the obesity.

We suggest that overweight or obesity can occur in Down syndrome in combination with celiac disease, and obese children with Down syndrome should be evaluated for various autoimmune disorders even the absence of typical signs and symptoms, but further studies are necessary.

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