Clinical Presentations of Coeliac Disease in Children from 2 to 14 Years

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Abstract

Background: To study presentations of childhood coeliac disease (CD) in different paediatric age groups.

Methods: This descriptive study was conducted on two hundred patients of either sex from two to fourteen years of age with suspicion of CD. Anti-endomysial titres (EMA) were done in all the patients. Those with positive EMA titres were subjected to endoscopy. Biopsy was taken for confirmation of CD.

Results: Out of two hundred patients seen, forty patients fulfilled the diagnostic criteria of CD. Mean age at presentation was eight years. Mean duration of symptomatology was 6.5 years. Male to female ratio was 1.6:1. Out of forty patients, 32% presented with chronic diarrhea, 22.5% with short stature. Iron deficiency anaemia was found in 20% and abdominal pain in 14.5%. Family history was positive in 4.55% patients.

Conclusion: Younger children present with intestinal manifestation of CD like chronic diarrhoea. Older children usually have extra-intestinal manifestations like short stature, iron deficiency anemia, abdominal pain and failure to thrive.

Key Words: Coeliac disease, anti endomysial antibody

Introduction

CD has a wide clinical spectrum ranging from malabsorption syndrome to extra-intestinal manifestations. It is induced by the ingestion of gluten, which is a component of wheat, barley and rye. The gluten protein is rich in glutamine. This protein is poorly digested in human upper gastrointestinal tract. The incidence of coeliac disease is estimated to be 0.3% to 1% in North American children.1

CD has a wide variety of clinical presentations. In “Classic form” intestinal symptomatology is prevalent including diarrhoea, abdominal distension and failure to thrive. Infants and young children commonly present with classical form of coeliac disease. Atypical form with predominantly extra-intestinal features, like short stature, neurological symptoms and anemia is seen in older children. “Silent form” includes those with no clinical symptoms. This form is usually present in first degree relatives of coeliac patients.2-4

Diagnosis of CD is based on serological testing and on duodenal biopsy. Anti-endomysial antibody and tissue trans-glutaminase antibodies are positive. Duodenal biopsy shows characteristic findings of intra-epithelial lymphocytosis, crypt hyperplasia and total villous atrophy and positive response to gluten free diet. The diagnostic criteria developed by the European Society for Paediatric Gastroenterology and Nutrition (ESPAGAN) requires only clinical improvement with gluten free diet as villous atrophy may persist despite clinical improvement. However, roughly 10% of cases are difficult to diagnose due to lack of concordance among clinical, serological and histological findings.5-9

Patients and Methods

This prospective study was conducted from March 2009 to February 2012, in department of Paediatrics, Railway Hospital, Rawalpindi. Two hundred patients with suspicion of CD were selected. Inclusion criteria included those with chronic diarrhoea, short stature, iron deficiency anemia not responding to iron therapy for three months, abdominal pain, failure to thrive and family history of CD. Informed consent about routine evaluation of CD was obtained from all patients. In those who had raised anti-endomysial titers conducted through ELISA, endoscopy was carried out with duodenal biopsy. Diagnosis of CD was based on endoscopic findings consistent with CD showing cobble stone appearance, scalloping, reduced duodenal folds or mosaic pattern of intestinal mucosa.

Results

In the two hundred patients with suspicion of CD male to female ratio was 1.6:1. Children from two to fourteen years of age were enrolled for this study. Most common presentation was chronic diarrhoea 28% (n=56) followed by iron deficiency anemia 27% (n=55). Six percent (n=12) had a positive family
history. Abdominal pain was present in 38% (n=76), failure to thrive in 4% (n=8) and 15% (n=31) had short stature. Anti-endomysial antibody titres were done in all patients. Out of two hundred patients, forty patients had positive anti-endomysial antibody titres (Table 1). They underwent distal duodenal biopsy with histo-pathological examination of the biopsy specimens. The findings were consistent with diagnosis of CD. Majority (70 %) showed atrophy of duodenal folds, while 30% patients showed cobble stone appearance on endoscopy. Histopathological examination revealed total villous atrophy in 60% specimens and near normal villous architecture with a prominent intraepithelial lymphocytosis in 40% of biopsy specimens.

Table 1: Serological findings

<table>
<thead>
<tr>
<th>Patients(n=200)</th>
<th>Frequency</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Antiendomysial antibody negative patients</td>
<td>n=160</td>
<td>80</td>
</tr>
<tr>
<td>Antiendomysial antibody positive patients</td>
<td>n=40</td>
<td>20</td>
</tr>
</tbody>
</table>

Mean age of presentation was eight years .Mean duration of symptoms was 6.5 years. The classical manifestation of chronic diarrhoea was the most common presentation and was seen in 32% (n=13) of patients. The next common presentation was short stature and the third was iron deficiency anemia found in 22.5% (n=9) and 20% (n=8) respectively (Table 2).

Fourteen children were under four years of age out of which thirteen presented with chronic diarrhoea and one with failure to thrive. Older patients between the ages four to fourteen presented with iron deficiency anemia, short stature, abdominal pain and failure to thrive. Only two patients had positive family history.

Table 2: Coeliac Disease-Presenting complaints

<table>
<thead>
<tr>
<th>Presenting complaints</th>
<th>Pretesting group (n-200)</th>
<th>Testing group (n-40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chronic diarrhoea</td>
<td>28%(n=56)</td>
<td>32%(n=13)</td>
</tr>
<tr>
<td>Iron deficiency anemia</td>
<td>24%(n=55)</td>
<td>20%(n=8)</td>
</tr>
<tr>
<td>Abdominal pain</td>
<td>38%(n=35)</td>
<td>17.5%(n=7)</td>
</tr>
<tr>
<td>Short stature</td>
<td>15%(n=31)</td>
<td>22.5%(n=9)</td>
</tr>
<tr>
<td>Failure to thrive</td>
<td>4%(n=8)</td>
<td>2.5%(n=1)</td>
</tr>
<tr>
<td>Positive family history</td>
<td>6%(n=12)</td>
<td>4.5%(n=3)</td>
</tr>
</tbody>
</table>

Discussion

The prevalence of CD varies in different parts of the world. Initially, it was thought to be a rare disease, affecting predominantly Caucasians. The disease was initially presumed to be less common in Asian and African populations, but recent reports shows rising trend in diagnosis. Recent surveys from the West show its prevalence to vary from 1 in 100 to 1 in 300 individuals.10 There is no population based data on the incidence or prevalence of the disease in Pakistan. However, CD is not uncommon in the pediatric population of this country presenting with chronic diarrhea.11

The clinical presentation varies considerably from full blown malabsorption syndrome to subtle and atypical symptomatology. Some patients present with combination of symptoms. Younger children present with typical features like diarrhoea, vomiting and failure to thrive. Older patients usually have atypical presentations like anemia, short stature, abdominal pain and osteoporosis. Both first and second degree relatives of coeliac disease have significant (5-15%) risk of developing the disorder.11,12

Findings in this study differs from other local studies describing clinical presentation of coeliac disease in Pediatric population. A study conducted at Services Institute of Medical Sciences and Services Hospital, Lahore found that chronic diarrhoea, failure to thrive and anemia were the commonest clinical features present in 82.6%, 89% and 95.6% of patients.13 Aziz S et al in a study of forty nine patients from Sindh Institute of Urology and Transplantation found chronic diarrhoea, failure to thrive, weight loss and short stature in 69%, 61%, 77% and 32% of patients respectively.14 In our study none of the patients presented with weight loss. In a study from Post Graduate Institute of Medical Education and Research, Chandigarh, India, five hundred and forty nine children with various presentations of coeliac disease were studied. It showed 84% presented with diarrhoea, failure to thrive was seen in 91%, anemia in 84% wasting in 87% and stunting in 60% of cases.15

In a Western study by Mc Gowan KE et al at Department of Pediatrics, University of Calgary, Canada diarrhoea occurred in 30%, short stature in 20%, iron deficiency anaemia in 11%.16 The results of this study coincides with our study. A study conducted at Department of Gastroenterology and Social security Children’s Hospitai, Ankara, Turkey is exactly in line with our study as 50 % of patients presented with chronic diarrhoea, 40% with short...
stature and 38% with iron deficiency anemia. These three presentations are the leading presentation of our study as well.

Majority of the patients with atypical symptoms of coeliac disease do not report to the hospitals in Pakistan. The “atypical form” of the disease is characterized by few, or no gastrointestinal symptoms. It predominantly presents with extra-intestinal features such as neurological, dermatological, hematological, endocrinological reproductive, renal, psychiatric, skeletal, and liver involvement. Taddeucci et al conducted a study on 26 patients with CD and found that 31% had abnormalities in central nervous system. One of the few studies conducted in Turkey describing the dermatologic findings associated with CD showed that cutaneous, mucosal, nail, and hair findings were present in 74.5%, 27.3%, 20.0%, and 7.3% of patients, respectively. Selimoglu performed a study in Italian children and found a high incidence of autoimmune thyroid disease (26.2%) in patients with CD and type 1 Diabetes mellitus was seen in 11% of patients having CD. Silent presentations of CD can be identified through screening program of high risk groups. It is also evident that anti-endomysial antibody is a useful screening test with sensitivity of 90%.

**Conclusion**

1. Coeliac disease may present with a wide spectrum of symptoms and signs which can vary with age.
2. Physicians should be aware of atypical manifestations in high risk populations in order to avoid under diagnosis of the disease.

**References**