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IP International Journal of Medical Paediatrics and Oncology

Journal homepage: https://www.ijmpo.com/

Case Report

Extraintestinal manifestation of Celiac disease: A case series

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ARTICLE INFO

Article history:
Received 02-12-2022
Accepted 14-12-2022
Available online 11-01-2023

Keywords:
Celiac disease
Extraintestinal manifestations

ABSTRACT

Celiac disease (CD) is a multisystem chronic inflammatory disorder with an autoimmune component in genetically susceptible individuals elicited by gluten and related prolamines. CD is becoming a common disorder arising at any age with a broad spectrum of symptoms ranging from intestinal to extraintestinal manifestations. Extraintestinal symptoms being more common after 2 years of life, can affect various systems and organs of the body. Knowledge of the extraintestinal manifestations of celiac disease is of utmost importance for the diagnosis of CD. Owing to the high risk of complications among the undiagnosed cases of CD, patients with extraintestinal manifestations should undergo appropriate diagnosis and treatment for CD.

1. Introduction

Celiac disease (CD) has a wide range of clinical situations ranging from the presence of gastrointestinal symptoms to failure to thrive in children, prolonged fatigue, unexpected weight loss and anemia.

It is present in approximately 1% of the population. Diarrhea, once known to be the presenting feature of celiac disease, has now become a less common presentation (< 50% of cases). Common extraintestinal presentations include iron refractory iron-deficiency anemia, osteoporosis, dental enamel defect, arthritis, short stature, dermatitis herpetiforme, delayed puberty, and neurologic disorders, mainly peripheral neuropathy and gluten related ataxia. Celiac disease is known to be associated with other autoimmune disorders more frequently (three to ten times more) than in the general population. A gluten-free diet is the standard of treatment, although its effect on some of the extraintestinal manifestations remains to be determined.

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2. Case Series

2.1. Patient 1

A 5 year old girl presented to our opd with complaints of abdominal distention and paleness of whole body for 15 days. There was no h/o any blood loss nor any previous blood transfusion nor any previous hospitalization.

On examination: pallor was present. Her anthropometry was HFA <1 percentile, WFA < 1 percentile, WFH was between 1^{st} and 3^{rd} percentile.

On investigation: Basic investigation (CBC, PBS, Reticulocyte count, LFT, KFT), iron profile, thyroid profile, celiac profile, ultrasound of whole abdomen, x-ray left wrist AP view was advised. Her reports were suggestive of microcytic hypochromic anemia with normal WBC and platelet count. Her bone age was 4 years. tTG was 174.29.

2.2. Patient 2

A 10 year old girl child came with complaints of paleness of whole body for 2 months, abdominal distention for 20 days. There was no h/o any fever, rash, bleeding from any site,

https://doi.org/10.18231/j.ijmpo.2022.037

any previous blood transfusion or hospitalization.

On examination: pallor was present, splenohepatomegaly was present. Her anthropometry revealed WFA $<3^{rd}$ percentile, HFA $< 3^{rd}$ percentile.

On investigation: Basic investigation was suggestive of bicytopenia with normal platelet count with microcytic hypochromin anemia. LFT, KFT were normal. HPLC was normal. DCT was negative. Vit b12 levels were normal. Bone marrow revealed trilinage haematopoiesis with erythroid hyperplasia. Infective etiology (malaria, kala azar, scrub typhus) were ruled out. UGI Endoscopy was suggestive of pangastritis and duodenal biopsy was suggestive of partial villous atrophy and crepts hyperplasia. Celiac profile was sent which turned out to be positive.

2.3. Patient 3

7 year old girl child came with complaints of not gaining height x 5 years with dryness of skin. No h/o diarrhea, vomiting, any chronic illness, previous hospitalization.

On examination: eczema was present. Her anthropometry revealed HFA $<1^{st}$ percentile, WFA $<3^{rd}$ percentile.

On investigation: Her basic investigations including thyroid profile were normal. Her bone age was 4 years 5 month. On clinical suspicion of extraintestinal manifestation of celiac disease relevant investigations were sent, which turned out to be positive.

3. Discussion

Celiac disease is a common multi systemic immune mediated disorder elicited by gluten and its related prolamines, in genetically susceptible individuals.

CD is a T cell mediated chronic inflammatory disease along with an autoimmune component.

Symptomatic: CD patients present with frank GI signs and symptoms (FTT, chronic diarrhea, weight loss) or extra intestinal manifestations (anemia, hypertransaminasemia, neurological disorders, arthralgia, aphthous stomatitis).

Silent: These patients are asymptomatic, mainly seen in first degree relative of CD patient or at risk groups, are diagnosed with serologic screening or histologic evidence of villous atrophy.

Latent: these patients have normal intestinal histopathology but at some time have shown gluten dependent enteropathy.

Potential: These subjects have positive celiac serology test without any abnormal intestinal histopathology. These patients may or may not be symptomatic and may or may not show gluten dependent enteropathy any time in life.

Celiac disease may be associated with other autoimmune disorders like type 1 DM, autoimmune thyroiditis, rheumatoid arthritis, autoimmune hepatitis, autoimmune cholangitis, primary biliary cholangitis, Addison disease, sjogren syndrome. Their relation with CD is not well understood.

Other conditions associated with CD are Down syndrome, William syndrome, Turner syndrome, IgA deficiency.

Common extra intestinal manifestations include:

Neurological: Peripheral neuropathy, ataxia, seizures, demyelinating CNS lesions.

Endocrine: Delayed puberty, short stature, impotency, infertility, amenorrhoea, secondary hyperparathyroidism.

Hematology: Anemia, thrombocytosis, haemorrhage.

Cutaneous: Dermatitis herpetiforme, ecchymosis, petechiae, follicular hyperkeratosis and dermatitis.

Hepatic: Isolated transaminitis, autoimmune hepatitis.

Muscular: Atrophy, tetany, weakness.

Skeletal: Osteoporosis, osteopenia, osteomalacia, osteoarthropathy, pathological fractures.

Other: Anxiety, aphthous stomatitis, enamel hypoplasia.

In western world, the prevalence has been reported as 0.8 to 2.67%.[1] In India, CD is prevalent in 1.54% by serologically and 1.04% by histologically.[2] Anemia and short stature were more common cause of CD in Western studies.[3] Short stature as a manifestation of CD in India is less common as published in a study a decade ago.[4] This could be due to lack of study or due to lack of knowledge about the extraintestinal intestinal manifestation of celiac disease leading to the low index of suspicion. This highlights the need to increase the awareness about extra intestinal manifestations of CD so as to enable early diagnosis and prompt treatment with gluten free diet.

The serological prevalence of CD ranges from 0.3% to 1.4% worldwide, A Study from North India has reported a frequency of celiac disease to be 1 in 310 children[5] whereas a study from SKMCHI, Jaipur reported a frequency of 1:166.[6]

A study conducted at SPMCHI, Jaipur, has shown a prevalence of about 35.7% among children with FTT, short stature, anaemia, constipation and unusual manifestations.[6]

In the previous Indian series on short stature, none of them had reported Celiac disease as a manifestation of short stature expect Bhadada S, Bhansali A et al, at PGI Chandigarh who reported CD as the most common cause of short stature (15.3%) among 176 children.[7]

In another study of selected population of short children by Rosenbach Y et al, reported celiac disease in 48% of the study.[8] Cacciari et al, reported another study in 1985 among 88 children with short stature suggested a total of 16 of 88 short children with Probable celiac disease as the cause of short stature.[9] "symptomless celiac disease is therefore a commoner cause of short stature than is hypopituitarism." Bhadada S, Bhansali A et al, studied 176 children who fitted the criteria for short stature, out of total children studied celiac diseases was found in 27 (15.3%) of them, making it the single most common cause of short stature.[7]

In a study conducted at SKMCHI, Jaipur, they noted that seronegative groups had significant low height as compared to seropositive group.[6] Emphasising to the point that symptom less celiac disease is therefore a commoner cause of short stature.

The Agency for Healthcare Research and Quality (AHRQ) report found a prevalence of CD in children by biopsy of 0.5% to 1.6% and by serology of 0.3% to 1.9%. Even though. The gold standard for the diagnosis of CD is duodenal biopsy, recent reports have shown that tTG alone is a sensitive market for CD, yet non-invasive. tTG is the investigation of choice for CD mass screening and helpful in identifying patients who can benefit from gluten free diet and follow up.

Celiac Disease can lead to short stature by causing autoimmune hypothyroidism, resistance to growth hormone, and malabsorption of protein, calcium and vitamin D. Additionally, Celiac Disease can lead to hypogonadism which inhibits the pubertal growth spurt.[10]

4. Conclusion

Celiac disease is one of the common disease in India, where wheat is‘the tip of iceberg. The common conception that celiac disease of west dT in many studies In conclusion, the prevalence of Celiac Disease is country specially among children with short stature. Celiac Disease must be considered as an important cause as an extra intestinal manifestation of CD should be screened for utilizing the available, simple, very sensitive and specific serological test which is much cheaper and less invasive than doing the dynamic growth hormone testing to start with.

5. Conflict of Interest

None.

6. Source of Funding

None.

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Cite this article: Kumari M, Kishore S, Kumar A. Extraintestinal manifestation of Celiac disease: A case series. *IP Int J Med Paediatr Oncol* 2022;8(4):160-162.