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A Rare Association of Celiac Disease with Turner Syndrome

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Abstract

Celiac Disease is an immune mediated enteropathy, with wide spectrum of extra intestinal manifestation and rare autoimmune and syndromic associations. We report one such exclusive extra intestinal manifestation of celiac disease with association of Turner syndrome.

Key words: Celiac, Turner, Gluten

Introduction

Celiac Disease is an immune - mediated enteropathy caused by permanent sensitivity to gluten in genetically susceptible individuals. It develops after dietary exposure to the protein gluten, which is found in wheat and rye[1]. Although it is primarily an enteropathy, it may manifest with a number of extra-intestinal manifestations[2]. We report one such rare presentation of Celiac Disease.

The Case

This 13 year old girl presented with effort intolerance and easy fatigability of eight years duration. Clinically she was found to have severe pallor. However, there was no rash, lymphadenopathy, organomegaly or bone tenderness. Anthropometry revealed severe stunting with proportionate short stature [Weight - 21 kg (45% of 50th centile). Height – 110 cms (less than 3rd centile) but a normal weight for height.She was also found to have short neck and preadolescent features in SMR staging (SMR Stage II). Systemic examination was essentially within normal limits.

Investigations revealed dimorphic anemia [Hb - 4.6 gm%] with normal urinalysis, chest X-ray and tuberculin test. The bone age was between 11 – 13 yrs, which approximates the chronological age but exceeds the height age ($5^1/_4$ years).The results are summarized in Table 1.

Table 1: Summary of Investigations

S.No	Investigation	Results	Remarks
1	Hemoglobin	4.9gm%	PBS – Dimorphic anemia
2	Serum Iron	29 µg/dl	normal range 50 to 120 µg/dL
3	Iron Binding capacity	280 mcg/dl	normal range 250 – 400 mcg/dl
4	Transferrin saturation	11.1%	normal range 25 – 45%
5	Radiology	Bone age - 11 – 13 yrs	which approximates the chronological age but exceeds the height age (51/4 years).
6	Urine analysis	Normal	
7	Tuberculin Test	Negative	
8	CXR	Normal	

In view of the presence of severe stunting and severe anemia, possibility of malabsorption syndrome was entertained and she was investigated accordingly. The anti-tissue trans-glutaminase (TTG) IgA titre was 298 units (normal range - less than 20 units) which was suggestive of Celiac Disease. Small intestinal biopsy was also done which confirmed the diagnosis (flattening of villi with increased intraepithelial lymphocytes).

Considering the proportionate short stature in a girl with short neck, an association of Turner Syndrome was suspected which was confirmed by Chromosome analysis (45, XO). USG abdomen revealed hypoplastic ovaries, echocardiography was within normal limit.

The child is presently being managed with gluten free diet and nutritional supplements with good response.

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Discussion

Celiac disease is an immunologically mediated small intestinal disease with a number of extra intestinal manifestations. The susceptibility is based on genetic and environmental factors. Epidemiologic studies in Europe and United States suggests that Celiac Disease may occur in 0.5% – 1.0% of general population[1]. The prevalence of this disease in India has also found to be same[3].

The mode of presentation of Celiac Disease is quite variable. Acute or chronic diarrhea, abdominal pain and failure to thrive are the usual presentations, though the disease may present with extra-intestinal manifestation only. The common extraintestinal manifestations of celiac disease are short stature, iron deficiency anemia, dermatitis herpetiformis, dental enamel defects, osteoporosis, delayed puberty, arthritis and hepatitis. Other known extra-intestinal manifestations include occipital lobe epilepsy with cerebral calcifications, cerebellar ataxia, myelopathy, autoimmune myocarditis and idiopathic dilated cardiomyopathy[4].

Celiac disease is rarely associated with other autoimmune disorders and genetic syndromes (Table 2)[5, 6, 7, 8, 9].

Table 2: Genetic diseases associated with Celiac disease

S.No.	Syndromes	Association
1	Insulin Dependent Diabetes Mellitus	4.5%
2	Autoimmune Thyroiditis	7.8%
3	Rheumatoid Arthritis	2.0%
4	Down Syndrome	4.6%
5	Sjogren's Syndrome	12%
6	Turner Syndrome	5%

Anti-endomysium antibody IgA and anti-tissue trans-glutaminase (TTG) IgA antibody are both highly sensitive & specific in identifying individuals with Celiac Disease. Anti-gliadin IgA & IgG are no longer recommended due to lack of specificity. Definitive diagnosis of celiac disease requires small intestinal biopsy. Revised criteria published by European Society of Pediatric Gastroenterology, Hepatology and Nutrition children with characteristic histologic features and unequivocal clinical response to gluten withdrawal can be considered definitive for celiac disease. The cornerstone of treatment for this disease remains the gluten free diet[1,10].

Conclusion

Our patient showed several unusual features. Although she was primarily a case of celiac disease, no gastrointestinal manifestation was present. But a number of extra-intestinal manifestations (short stature, delayed puberty & iron deficiency anemia) were present including the rare association with Turner's syndrome. To the best of the authors' knowledge, this has not been described in Indian literature.

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