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# Multiple Faces of Celiac Disease in Same Family

Vani Malhotra, Parveen Malhotra, Anuj Sharma, Yogesh Sanwariya, Vikash Singh, Ahsan Khan

*Department of Obstetrics & Gynecology Medical Gastroenterology , PGIMS, Rohtak, Haryana & Safdarjung Hospital and VMCC Medical College, New Delhi, India*
*Address for correspondence- Vani Malhotra,128/19, Civil Hospital Road, Rohtak, Haryana, India (124001). E-*

***Abstract***
*Celiac disease is immune related disorder of small bowel which is seen in genetically predisposed people and is due to permanent intolerance to wheat gliadins and other cereal prolamins also known as gluten-sensitive enteropathy or non-tropical sprue. In 1888. It was first described by Dr. Samuel Gee and in Greek means koiliakaos-abdominal. Dicke in 1950 highlighted association between the consumption of bread, cereals and diarrhea which improved after stoppage of wheat intake. The diagnosed cases of celiac disease represent just tip of iceberg and rest 90% are hidden and undiagnosed. epidemiology of CD has iceberg characteristics with more undiagnosed cases. Celiac disease is confirmed by clinical symptoms, serology, endoscopy, histopathological diagnosis and resolution of symptoms after gluten restricted diet. As celiac disease requires life-long gluten restriction, thus compliance rate varies between 50% to 90%, and is slightly more in elderly & females. As per available data, there are multiple presentations of celiac disease but to be seen in same family, it is rare*

***Keywords:*** *Celiac Disease, Endoscopy, Gluten, Dental enamel, Hypothyroidism, Transaminitis, Anaemia*

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## I. INTRODUCTION

Celiac Disease (CD) is a immune-mediated disorder of small bowel that is seen genetically predisposed people **[1]**. Wheat, rye, oat and barley prolamins are the major culprit due to prescence of glutamine and proline content in significant amount **[2]**. In the past CD presented in majority of patients with typical gastrointestinal symptoms but now half of cases present with variety of atypical symptoms or even without any symptoms **[3]**. Marsh classification is used for histologic changes and vary from prescence of intraepithelial lymphocytes to severe villous atrophy. The anti-tissue transglutaminase antibodies are the most sensitive test for CD **[2].** Various etiological factors are considered for CD including genetic (HLA class II antigen) and environmental risk factors **[4]** including GI infections **[5]**. The transglutaminase auto antibodies play a role in disease pathogenesis **[6]**. The prevalence of CD worldwide and in India is globally 1% **[7].**

## II. CASE SERIES

**CASE 1-** A fourty two-year-old male presented with unexplained transaminitis for last one year and after ruling out all other causes, he was diagnosed to be celiac disease on basis of serological, endoscopic and histopathological examination of duodenal biopsy. His serum IgATTG antibody level was raised to 68 I.U. and duodenal biopsy was suggestive of Marsh Grade 2 classification. He was put on strict gluten restricted diet and within three months transaminases became normal and after one year of follow up on strict gluten restricted diet, his transaminases are normal.

**CASE-2-** As per protocol, his family was screened for celiac disease and his wife was asymptomatic largely and was on treatment for last three months with 100 micrograms of thyronorm for hypothyroidism for last three months. Her all-other biochemical labs and ultrasonogram was found to be normal. In view of family history of celiac disease and hypothyroidism which has 5% association with celiac disease, she was advised for serum IgATTG antibody level which was increased to level of 45 I.U. and duodenal biopsy was suggestive of Marsh Grade 1 classification. She was immediately put on strict gluten restricted diet and after three months of same, repeat serum IgATTG antibody level were found to be in normal range and thyroid level were also well under control.

**CASE-3**- The only child of both these parents who were diagnosed to be suffering from celiac disease was seven-year-old boy who was asymptomatic largely except for dental enamel changes, showed significant rise of serum

IgATTG antibody level of 110 IU/ml, whereas normal range was 0-20 IU/ml. He was subjected to upper gastro-intestinal endoscopy which showed moderate scalloping of duodenal folds and on histopathological examination proved it to be celiac disease with Marsh Grade 3 stage. On general physical examination, the teeth of this child had dental enamel defect selectively in upper incisor and canines which parents admitted that it was for last two years. His rest examination including systemic one was normal. All his biochemical tests including hemogram, blood sugar, thyroid profile was normal except for slightly low level of serum calcium and serum Vitamin D3 level. He was immediately put on strict gluten restricted diet, along with calcium and vitamin D3 supplementation. After six months of gluten restricted diet, his Serum IgATTG antibody, Calcium, Vitamin D3 levels were found to be normal but there was no improvement in teeth. He was reviewed again after a gap of one year but still there was no change in dental enamel defect in upper incisors and canines.

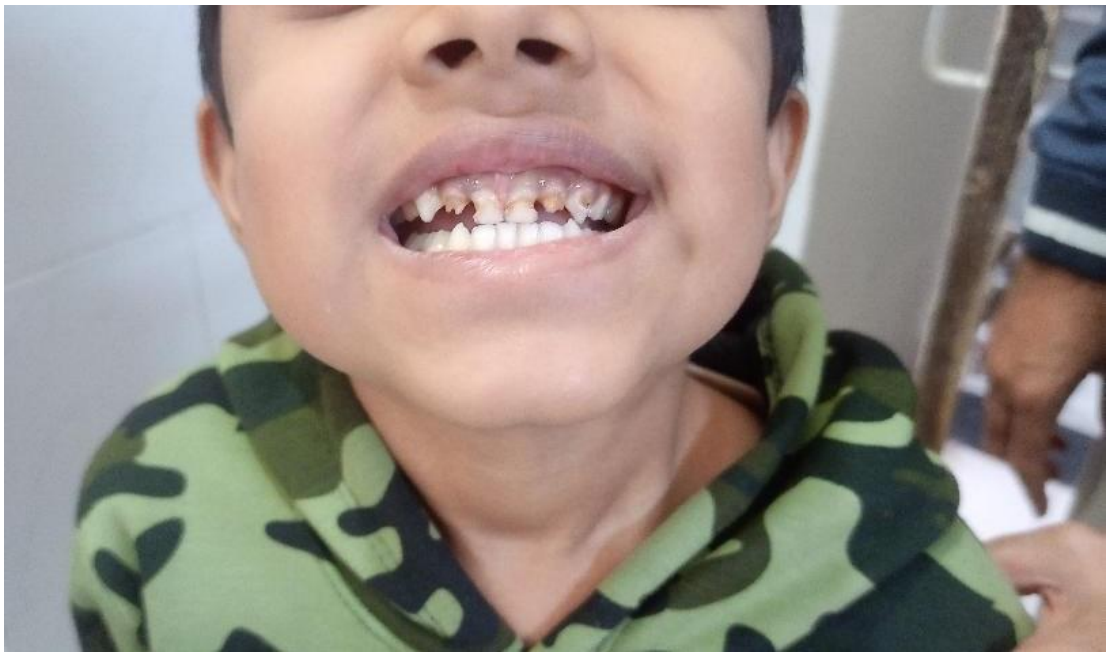

**Figure 1: -**Showing Dental Enamel Defect in Celiac Disease.

**CASE-4-** The sister of the index patient had symptoms of generalized weakness for last few months but was never investigated or treated for the same. In view of family screening for celiac disease when she was clinically examined and investigated, then she turned out to be having iron deficiency anaemia and rest all her biochemical labs and ultrasonogram abdomen was found to be essentially normal. Her cardiological, respiratory, neurological & gynaecological examination was essentially normal but serum IgATTG antibodies were raised to 80 I.U./ml (normal being 0- 20 I.U./ml) and endoscopy showed moderate scalloping of duodenal folds in second part of duodenum and on histopathological examination, Marsh grade 2 celiac disease was diagnosed. She was advised gluten restricted diet which was rigorously followed, thus symptomatic recovery started and she had complete resolution of symptoms within six months, along with increase of haemoglobin to 13 gm% and generalized weakness subsided completely.

## III. DISCUSSION

Celiac disease has varied clinical presentations both typical and atypical. The typical one are diarrhoea, constipation, short stature etc and atypical one includes diabetes mellitus, hypothyroidism, osteoporosis, unexplained transaminitis, iron deficiency anaemia, down syndrome & dental enamel changes **[8,9,10]**. The initial belief of presentation of celiac in majority of patients between 5-15 yrs of age has also been changed and now it is seen in children and adults in equal proportion. Family screening is must because in many family members despite being asymptomatic may have celiac in form of atypical presentation, silent or latent celiac. Our case series typically re-emphasizes the need of family screening and awareness about different presentations of celiac disease.

## IV. CONCLUSION

Celiac disease has many faces and associations with other diseases. It has same tip of iceberg phenomenon of diagnosed cases like hypertension, thus merits more vigil among treating doctors of various specialties for early diagnosis and prevention of development of complications like blood cancer and colon malignancy.

**CONFLICT OF INTEREST**- No conflict of interest and prior permission from patient and relatives was taken before publishing the case report.

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