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Original Research Article

A Study of the Magnitude of Celiac Disease in Siblings of Children (1–18 Years) with Confirmed Celiac Cases

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Abstract

Background: Celiac disease (CD) is chronic autoimmune disorder triggered by ingestion of gluten in genetically predisposed individuals. It has a wide spectrum of clinical presentations, ranging from classical gastrointestinal symptoms to silent or atypical forms. CD is strongly associated with genetic factors, particularly HLA-DQ2 and HLA-DQ8 haplotypes, and is more common among first-degree relatives of affected individuals. This study focuses on magnitude of CD among siblings of confirmed paediatric celiac patients.

Objective: To determine magnitude of celiac disease in siblings of children diagnosed with CD at tertiary care center in Rajasthan and to assess clinical presentation, serological, and histopathological characteristics in this high-risk group.

Methods: This hospital-based, cross-sectional study was conducted at Sawai Man Singh Medical College and Hospital, Jaipur, Rajasthan, India from May 2021 to May 2022. Fifty siblings of 44 biopsy-confirmed CD patients were recruited. All participants underwent serological testing for anti-tissue transglutaminase (tTG-IgA) antibodies, and those with positive serology were subjected to upper gastrointestinal endoscopy and biopsy. Histopathological examination using Modified Marsh Classification was performed to confirm deign.

Results: prevalence of celiac disease among siblings was found to be 14%, with 7out of 50participants testing positive for tTG-IgA. Among these, 57%were female, and most were asymptomatic or presented with non-classical symptoms such as abdominal pain, short stature, and pallor. Endoscopic and histopathological examination confirmed diagnosis of CD in all serologically positive cases, with Marsh Grade 3a and 3b being most common findings.

Conclusion: Study highlights a significant prevalence of CD among siblings of affected individuals, underscoring importance of targeted screening in this high-risk group. Early diagnosis and intervention, particularly in asymptomatic or non-classical cases, can prevent long-term complications. Routine screening for CD in siblings of confirmed cases should be recommended to ensure timely diagnosis and management.

Keywords: Celiac disease, siblings, gluten, tTG-IgA, biopsy, screening, high-risk group, paediatric.

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Introduction

Celiac disease (CD) is a common autoimmune disorder triggered by the ingestion of gluten—a protein found in wheat, barley, and rye—in genetically predisposed individuals. It primarily affects the small intestine, leading to inflammation and damage of the intestinal mucosa, which results in impaired nutrient absorption.

The genetic predisposition is largely associated with the presence of specific human leukocyte antigen (HLA) haplotypes, particularly HLA-DQ2 and HLA-DQ8. These genetic markers are present in approximately 30-40% of the general population but are seen in over 95% of individuals diagnosed with celiac disease. However, only a small proportion of individuals with these markers develop CD, indicating that environmental factors, especially gluten consumption, also play a significant role in the disease's onset.

CD manifests in various forms, with symptoms ranging from classical gastrointestinal issues, such

as diarrhoea, abdominal pain, and malabsorption, to non-classical or extra-intestinal symptoms like anemia, fatigue, and osteoporosis. A growing number of patients remain asymptomatic, complicating the diagnosis and leading to under-diagnosis or delayed diagnosis. Due to this variability in symptom presentation, CD is often described as an iceberg disease, with many undiagnosed cases hidden beneath the surface.

Globally, CD affects approximately 1% of the population, with a prevalence of 0.5-1.26% across most parts of the world. In India, recent studies estimate the prevalence to be around 1.4%, with a particularly higher incidence in the northern regions where wheat consumption is more prevalent. Rajasthan, a northern Indian state, falls within this high-prevalence zone due to its traditional diet being rich in gluten-containing foods. One of the significant aspects of CD is its familial clustering. Studies indicate that first-degree relatives of individuals with CD, particularly siblings, are at a higher risk of developing the disease due to shared genetic and environmental factors. The risk of CD in first-degree relatives is estimated to be 5-10%, significantly higher than in the general population. Therefore, early screening and diagnosis in this high-risk group are critical to prevent complications arising from untreated CD, such as growth failure, anemia, osteoporosis, and an increased risk of malignancies.

This study aims to assess the prevalence and magnitude of celiac disease among the siblings of confirmed CD cases at a tertiary care center in Rajasthan, India. Early detection of CD through screening in this high-risk group can lead to timely intervention, primarily the initiation of a gluten-free diet, thereby preventing long-term health consequences.

Purpose of the Study: The study seeks to determine the prevalence of CD among siblings of confirmed celiac disease cases, aged between 1 and 18 years, at a tertiary care center in Rajasthan. Identifying the magnitude of CD in this high-risk group will help establish the need for regular screening and early diagnosis, which can prevent complications through timely treatment with a gluten-free diet. The findings could contribute to better health outcomes and inform healthcare policies regarding the screening and management of CD in high-risk populations.

Hypothesis

- **Null Hypothesis**: There is an increased prevalence of celiac disease among siblings of patients diagnosed with celiac disease.
- **Alternative Hypothesis**: There is no increased prevalence of celiac disease among siblings of patients diagnosed with celiac disease.

Aims and Objectives

Aim: To assess the magnitude of celiac disease among siblings of patients with confirmed celiac disease.

Objectives:

1. To determine the proportion of siblings of confirmed celiac disease patients who test positive for CD.
2. To assess the clinical features of CD in siblings, including both symptomatic and asymptomatic presentations.
3. To identify the histopathological and serological characteristics of CD among the sibling population.
4. To recommend screening strategies for siblings of CD patients to facilitate early detection and treatment.

Material and Methods

Study Design: This is a hospital-based, cross-sectional observational study conducted at the Department of Pediatrics, Sawai Man Singh Medical College & Hospital, Jaipur, Rajasthan, India from May 2021 to May 2022.

Study Population: The study recruited 50 siblings of 44 confirmed celiac disease patients, all aged between 1 and 18 years, who were receiving follow-up care at the Paediatric Gastroenterology Department of SPINPH.

Inclusion Criteria

- Siblings of confirmed celiac disease patients (diagnosed through biopsy) aged 1 to 18 years.
- Parents or guardians who provided informed consent for the study.

Exclusion Criteria

- Siblings younger than 1 year or older than 18 years.
- Patients who were already on a gluten-free diet at the time of recruitment.
- Siblings with other gastrointestinal disorders such as inflammatory bowel disease or malabsorption syndromes.
- Siblings or parents unwilling to provide a blood sample or undergo endoscopic biopsy.

Sample Size: Based on previous studies, the expected prevalence of celiac disease in siblings of confirmed celiac cases was approximately 6.6%. At a 95% confidence level and an alpha error of 0.05, the calculated sample size was 50 siblings.

Data Collection and Procedures

Demographic and Clinical Data: A pre-designed proforma was used to collect demographic details, clinical history, and anthropometric data of all participants.

Serological Testing: Blood samples were taken from all siblings for serum tissue transglutaminase (tTG-IgA) antibody testing, which was performed using the Chorus ELISA kit. A tTG-IgA level above 15 AU/ml was considered positive for celiac disease.

Endoscopic Biopsy: Participants with positive tTG-IgA results underwent upper gastrointestinal (UGI) endoscopy, and duodenal biopsies were taken from the second part of the duodenum. The biopsies were analyzed for histopathological changes, and Marsh grading was used to classify the degree of villous atrophy.

Statistical Analysis: Data were analyzed using IBM SPSS Statistics (version 22.0). Means were compared using Student's t-test, while categorical variables were analyzed using the chi-square test or Fisher's exact test where applicable. A p-value of <0.05 was considered statistically significant.

Results

Demographic Characteristics: Among the 50 siblings of confirmed celiac disease patients, 23 were female (46%) and 27 were male (54%). The age distribution showed that 19 (38%) siblings were in the 0-5 years age group, 15 (30%) were aged between 6-10 years, and 16 (32%) were aged between 11-18 years. There was no statistically significant difference in gender distribution among the siblings ($p = 0.353$).

Clinical Features in CD Patients: The clinical features of the 44 confirmed celiac patients showed a wide range of symptoms, with abdominal pain being the most common (50%), followed by pallor (59%) and short stature (47%). Constipation (38%) was observed more frequently than diarrhoea (25%), indicating a shift toward non-classical symptoms.

Prevalence of Celiac Disease in Siblings: Out of 50 siblings, 7 (14%) tested positive for serum tTG-IgA. Of these, 4 were symptomatic, presenting with abdominal pain, abdominal distention, short stature, and pallor. The remaining 3 were asymptomatic. The symptomatic siblings showed a higher prevalence of gastrointestinal symptoms, similar to those seen in confirmed CD patients. The prevalence of CD was significantly higher among female siblings, with a male-to-female ratio of 1:1.3 in tTG-IgA-positive cases.

Endoscopic and Histopathological Findings: All 7 serologically positive siblings underwent upper gastrointestinal endoscopy, which revealed scalloping in 5 cases (71%), reduced fold height in 4 cases (57%), and nodularity in 3 cases (43%). Histopathological examination of duodenal biopsies showed Marsh grade 3b in 2 siblings (28.5%), Marsh grade 3a in 3 siblings (42.8%), and Marsh grade 3c in 1 sibling (14.2%).

Discussion

This study found a prevalence of 14% for celiac disease among the siblings of confirmed CD patients, which is significantly higher than the general population's prevalence of approximately 1%. This is in line with findings from previous studies, such as the meta-analysis conducted by Singh et al. (2015), which reported a pooled prevalence of 8.9% among siblings of celiac patients. The higher prevalence of CD among siblings, particularly female siblings, can be attributed to the shared genetic background and environmental exposure.

The study also highlighted the wide spectrum of clinical presentations in both symptomatic and asymptomatic siblings. Interestingly, many of the symptomatic siblings exhibited non-classical symptoms such as constipation, short stature, and abdominal distention, similar to the trend observed in recent studies where non-classical forms of CD are becoming more common due to better awareness and diagnostic methods.

The endoscopic and histopathological findings of the serologically positive siblings were consistent with CD. The presence of Marsh grade 3a and 3b in most cases indicates significant villous atrophy, further confirming the diagnosis of CD in this high-risk group.

Implications of Early Diagnosis and Management: Early diagnosis of celiac disease in siblings of confirmed cases is crucial for preventing complications such as growth failure, anemia, and osteoporosis. This study reinforces the need for routine screening of first-degree relatives, particularly siblings of patients with celiac disease, given the high prevalence observed in this study.

Early identification through serological screening and confirmatory endoscopic biopsy can facilitate timely intervention, primarily the implementation of a gluten-free diet. Adherence to a strict gluten-free diet can lead to significant improvements in growth, gastrointestinal symptoms, and overall quality of life, especially in children, as their bodies are still developing.

A gluten-free diet has been shown to result in rapid clinical improvement in younger patients with classical CD symptoms, such as diarrhoea and malabsorption, and can also help resolve extra-intestinal manifestations like short stature and anemia. Early diagnosis is especially important in asymptomatic cases, where patients might not exhibit noticeable symptoms but still suffer from the underlying damage caused by gluten intake. By identifying CD before severe symptoms or

complications arise, siblings can avoid long-term health consequences and maintain better overall health.

Challenges in Diagnosis and the Role of Screening Programs: One of the challenges in diagnosing celiac disease is its heterogeneous presentation. As this study has shown, many siblings with positive serology and histopathology for CD were either asymptomatic or presented with non-classical symptoms like short stature, abdominal distention, and constipation. These atypical or silent forms of CD are harder to detect clinically and often go undiagnosed for long periods, increasing the risk of complications such as osteoporosis, iron-deficiency anemia, and delayed puberty.

In this context, the findings from this study suggest that screening programs targeting high-risk populations, such as siblings of confirmed CD patients, are essential. While mass screening of the general population for CD remains controversial due to concerns over cost-effectiveness and adherence to a gluten-free diet, screening among high-risk groups like siblings has a higher diagnostic yield and is more justified from a public health perspective. This is especially important given the genetic predisposition that siblings share, making them significantly more likely to develop CD than the general population.

Moreover, repeated screening over time may be necessary for siblings who initially test negative for CD, as the disease can develop later in life. Guidelines from organizations such as the European Society for Paediatric Gastroenterology, Hepatology, and Nutrition (ESPGHAN) recommend that first-degree relatives of CD patients be screened periodically to account for the potential seroconversion that may occur later in childhood or adolescence.

Study Limitations and Recommendations for Future Research: While this study provides valuable insights into the prevalence and clinical presentation of celiac disease among siblings of confirmed cases, it has certain limitations.

Being a single-center, cross-sectional study, the findings may not be generalizable to other regions or populations with different dietary habits, genetic backgrounds, or environmental exposures. Additionally, the relatively small sample size (50 siblings) may limit the statistical power of the study.

Another limitation is the lack of longitudinal follow-up to monitor the long-term outcomes of the siblings who tested negative for CD during the study period. Celiac disease can develop at any age, and seroconversion might occur years after the initial screening. Future studies should consider long-term follow-up of siblings and other high-risk groups to assess the incidence of delayed-onset CD.

Future research should also explore the psychosocial and economic implications of screening for and managing CD in siblings, particularly in asymptomatic cases. While the medical benefits of a gluten-free diet for individuals with CD are well-documented, the psychological burden of adhering to such a strict diet, particularly in a gluten-rich cultural context, should be studied further. Understanding the challenges faced by these families can help in designing better support systems and education programs to ensure compliance with dietary recommendations.

Conclusion

This study found a significant prevalence (14%) of celiac disease among siblings of confirmed celiac disease patients, highlighting the importance of targeted screening in this high-risk group. The study also demonstrated that many siblings with CD present with non-classical or silent forms of the disease, further emphasizing the need for vigilance and comprehensive screening protocols. Early diagnosis and intervention, through the implementation of a gluten-free diet, can prevent the severe complications associated with untreated CD, such as malnutrition, growth failure, and other autoimmune conditions. Given the higher prevalence of CD in siblings compared to the general population, regular screening for CD in this group is highly recommended, especially for those presenting with mild or non-classical symptoms. Periodic follow-up screenings for those who initially test negative should also be considered, given the potential for delayed disease onset.

In conclusion, the study reinforces the importance of early detection and proactive management of celiac disease in high-risk populations. By implementing routine screening protocols for siblings of confirmed celiac patients, healthcare providers can significantly improve outcomes, reduce morbidity, and enhance the quality of life for individuals who might otherwise go undiagnosed.

Recommendations

Based on the findings of this study, the following recommendations are proposed:

1. **Routine Screening of Siblings**: All siblings of confirmed celiac disease patients should undergo routine screening for CD, even in the absence of symptoms. Serological testing, particularly for tTG-IgA, should be the first line of investigation, followed by endoscopic biopsy if necessary.

2. **Regular Monitoring of At-Risk Populations**: Siblings who test negative for CD should be monitored regularly, as seroconversion can occur later in life. Follow-up screenings should be conducted at regular intervals, particularly during periods of rapid growth or significant changes in dietary habits.
3. **Comprehensive Education and Support**: Families of CD patients, particularly those with multiple affected members, should be provided with comprehensive education about the disease, the importance of a gluten-free diet, and the potential complications of untreated CD. Support groups and counseling services should be offered to help families manage the dietary and psychological challenges associated with CD.
4. **Further Research on Genetic and Environmental Triggers**: While genetic predisposition plays a key role in the development of CD, further research is needed to understand the environmental triggers that lead to the onset of the disease in genetically susceptible individuals. Studies exploring the role of infections, dietary patterns, and other environmental factors could provide valuable insights into prevention strategies.
5. **Policy Implications**: Health authorities should consider implementing guidelines for the routine screening of first-degree relatives of celiac disease patients. Such policies could help improve early detection rates and reduce the burden of untreated CD on the healthcare system.

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