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Research Article

Celiac Disease: Symptoms, Risk Factors, Causes, and Treatment

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Abstract – Celiac disease, also known as celiac sprue, is a chronic inflammatory disorder of the small intestine triggered by the ingestion of gluten in genetically predisposed individuals. This study systematically reviews data on celiac disease and its treatment. The objectives include an overview of the clinical manifestations, diagnostic methods, associated pathologies, and pharmacological and non-pharmacological treatment approaches. The methodology involved a literature review focusing on epidemiology, pathogenesis, clinical presentation, diagnostic tests, and therapeutic management. The results indicate that celiac disease is multifactorial, involving genetic and environmental factors. Genetic testing and serological assays are among the most effective diagnostic tools. The disease is commonly associated with thyroid disorders, liver diseases, osteoporosis, and neurological complications. A strict gluten-free diet is the definitive treatment, although pharmacological interventions are necessary in refractory cases.

Keywords – Celiac Disease, Serologic Tests, Symptoms, Treatments, Diagnosis, Autoimmune Disorders.

I. INTRODUCTION

Celiac disease is an autoimmune disorder resulting from an abnormal immune response to gluten, a protein found in wheat, barley, and rye. It was first described in 1888 by Samuel Gee, with the role of gluten in its pathogenesis identified in 1953. The condition leads to enteropathy, characterized by mucosal damage and malabsorption of nutrients. It manifests as a spectrum of clinical symptoms and is associated with various systemic disorders.

GENERAL INFORMATION

1. **Epidemiology** Celiac disease is one of the most common genetically-based disorders, with a prevalence of 1-2% in Europe and similar rates reported in the United States (Lebwohl et al., 2018). Many cases remain undiagnosed due to varied clinical presentations. The "iceberg model" illustrates that only a fraction of cases present with typical symptoms, while many remain undiagnosed (Ludvigsson et al., 2013).
2. **Small Intestinal Structure and Histology** The small intestine is composed of the duodenum, jejunum, and ileum. It is histologically characterized by villi, crypts, and intraepithelial lymphocytes.

In celiac disease, histological changes include villous atrophy, crypt hyperplasia, and increased intraepithelial lymphocytes (Green & Cellier, 2007).

3. **Genetic Susceptibility** Over 90% of celiac patients carry the HLA-DQ2 or HLA-DQ8 genetic markers. However, the presence of these markers alone is not sufficient for disease development, indicating the role of additional genetic and environmental factors (Maki et al., 2003).
4. **Immune System and Pathogenesis** Celiac disease involves an inappropriate immune response to gluten peptides, leading to chronic inflammation and mucosal damage. This process is mediated by transglutaminase 2, which modifies gluten peptides, making them more immunogenic (Fasano et al., 2015).
5. **Environmental Factors** Early-life dietary patterns and infections, such as rotavirus, have been implicated in disease onset. Other potential contributors include gut microbiota composition and exposure to heavy metals (Lebwohl et al., 2018).

II. OBJECTIVES

- To summarize the clinical manifestations of celiac disease.
- To review various diagnostic methods.
- To analyze comorbidities associated with celiac disease.
- To evaluate pharmacological and non-pharmacological treatment options.

III. MATERIALS AND METHOD

A literature review was conducted using keywords such as "celiac disease," "gluten," "pathogenesis," "immune system," and "enzymatic degradation." Sixty relevant articles were included, encompassing clinical and in vitro studies.

1. **Clinical Presentation**

Celiac disease exhibits diverse clinical phenotypes:

- **Classical form:** Characterized by malabsorption symptoms such as diarrhea, weight loss, and abdominal pain.
- **Atypical form:** Predominantly extraintestinal manifestations, including anemia and osteoporosis.
- **Silent form:** Histological and serological abnormalities without clinical symptoms.
- **Latent form:** Genetically predisposed individuals without current symptoms or histological changes.
- **Refractory form:** Persistent symptoms despite a gluten-free diet, requiring immunosuppressive therapy.

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2. **Associated Conditions** Celiac disease is frequently associated with autoimmune and metabolic disorders:
    - **Type 1 Diabetes Mellitus:** Up to 10% of type 1 diabetics exhibit celiac autoantibodies.
    - **Thyroid Disorders:** Increased prevalence of Hashimoto's thyroiditis and Graves' disease.
    - **Liver Diseases:** Chronic hypertransaminasemia is common.
    - **Neurological Disorders:** Includes epilepsy, ataxia, and neuropathy.
    - **Bone Disorders:** Increased risk of osteoporosis due to vitamin D and calcium malabsorption.

3. Diagnostic Tests

3.1 Serological Tests

- Anti-tissue transglutaminase (tTGA-IgA) and anti-endomysial antibodies (EMA-IgA) are the most sensitive and specific.
- Anti-gliadin antibodies (AGA) are less specific but may be useful in young children.

3.2 Histological Examination

- Biopsy of the small intestine remains the gold standard for diagnosis.
- The Marsh classification is used to grade histopathological findings.

3.3 Genetic Testing

- HLA-DQ2/DQ8 testing is useful in uncertain cases but not diagnostic on its own.

3.4 Gluten Challenge Test

- Involves controlled gluten exposure with monitoring of immune and histological responses.

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4. Treatment Approaches

4.1 Gluten-Free Diet (GFD)

- The cornerstone of treatment; strict adherence leads to mucosal healing and symptom resolution.
- Nutritional supplementation is necessary for vitamin and mineral deficiencies.

4.2 Enzyme Therapy

- Prolyl endopeptidases (e.g., ALV003) are being investigated to degrade immunogenic gluten peptides.

4.3 Modified Grains

- Genetic modification and enzymatic processing to reduce gluten immunogenicity are under study.

4.4 Rho Kinase Inhibition

- Potential therapeutic target for modulating intestinal permeability.

4.5 Immunotherapy

- Strategies include IL-15 blockade and regulatory T-cell enhancement.

IV. RESULTS AND DISCUSSION

Celiac disease is a complex autoimmune condition with a wide range of clinical manifestations. While a gluten-free diet remains the mainstay of treatment, new therapeutic strategies are under investigation. Advances in enzyme therapy, immune modulation, and modified grains hold promise for improved disease management.

V. CONCLUSION AND LIMITATIONS

This review highlights the multifaceted nature of celiac disease and the importance of early diagnosis and management. While dietary adherence remains the primary treatment, pharmacological options are evolving. Future research should focus on alternative therapies to improve patient outcomes.

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