

# Understanding coeliac disease: Causes, symptoms, and diagnosis.

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## Abstract

**Coeliac disease is a common but often under diagnosed condition with important complications. It is due to immune-mediated gluten intolerance and may present in a number of ways. It has become more frequently diagnosed due to the recognition of the atypical presentations. In recent years, more sensitive and specific serological markers have been developed but the gold standard of diagnosis remains duodenal biopsy. Compliance with a strict, lifelong gluten-free diet is the cornerstone of management, improving symptoms and reducing complications of the disease a gastroenterologist's responsibility increasingly includes managing celiac disease.**

**Keywords:** Celiac disease, Ultrashort celiac disease, Refractory celiac disease, Collagenous sprue, Q-MARSH

## Introduction

A growing number of referrals are being made as a result of recent prevalence studies' finding that 1% of the general UK population had positive celiac serology results. Today, very sensitive and precise serological tests are used to diagnosis the majority of referrals, which are then quickly followed by endoscopic biopsies. As a result, we are now able to detect a large number of individuals who have no or few clinical symptoms, making the traditional combination of diarrhea, steatorrhea and weight loss very uncommon. Several of the early studies on the clinical features of classical coeliac disease (*i.e.*, those conducted and published prior to might not be relevant to modern-day celiac disease. These modifications to clinical practise include [1].

Coeliac disease is increasing in prevalence, which is currently estimated at one in 100 of the population and may occur de novo in adults. The diagnosis requires a joint clinic pathological approach; the recommended first-line test is serology with Immunoglobulin A (IgA) tissue transglutaminase and IgA endomysial antibodies. These serological tests show high levels of sensitivity and specificity, but biopsy is the gold standard to confirm the diagnosis. It is important that both tests are performed before the introduction of a gluten-free diet. Although the classical histopathology changes of coeliac disease with partial or total villous atrophy are well recognized, the pathology classification of coeliac disease is changing, with recognition that coeliac disease may show minimal pathology (normal architecture and an intraepithelial lymphocyte count/100 enterocytes. This entity is also described as lymphocytic duodenosis, and recommendation of follow-up serology testing is paramount in this condition. Follow-up of patients with coeliac disease is warranted, as normal serology does not predict mucosal recovery. Failure to heal predicts risk of progression to refractory coeliac disease and malignancies. Refractory coeliac disease occurs in 1%-2% of patients and this

diagnosis requires a combined clinical and histopathology approach with immunocytochemistry [2].

Once thought to be a rare disease of childhood, coeliac disease is now recognized as a common but under diagnosed cause of gastrointestinal symptoms in adults. Recent guidelines aim to improve recognition and assessment of this condition and will impact on the workload of all gastrointestinal specialists. In this article, current understanding of coeliac disease and controversies in diagnosis will be discussed. By considering the varied presentation of coeliac disease as well as high-risk groups and common complications, the implications for the role of the specialist nurse will be examined [3].

Coeliac disease is a chronic enteropathy caused by intolerance to gluten proteins. The true prevalence of this condition is greater than previously thought, with increasing numbers of 'silent' cases being diagnosed. Untreated coeliac disease is associated with significant morbidity and increased mortality. There have been a number of advances in our understanding of the pathogenesis of coeliac disease, in particular the mechanisms whereby gluten epitopes are processed, become modified by tissue Transglutaminase (tTG) and then interact with HLA restricted T cells. An improved understanding of the immune response to gluten is likely to lead to the development of novel strategies for the treatment of coeliac disease [4,5].

## Conclusion

Diagnostics will play a central role in addressing the ongoing dramatic rise in global prevalence of coeliac disease, and in deploying new non-dietary therapeutics. Clearer understanding of the immunopathogenesis of coeliac disease and the utility of serology has led to partial acceptance of non-biopsy diagnosis in selected cases. Non-biopsy diagnosis may expand further because research methods for measuring gluten-specific CD4+ T cells and the acute recall response to gluten ingestion in

patients is now relatively straightforward. This perspective on diagnosis in the context of the immunopathogenesis of coeliac disease sets out to highlight current consensus, limitations of current practices, gluten food challenge for diagnosis and the potential for diagnostics that measure the underlying cause for coeliac disease, gluten-specific immunity.

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