key points

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Historically coeliac disease was considered an uncommon paediatric condition presenting with diarrhoea, weight loss, and failure to thrive. However, recent population studies show that it affects around 1 in 100 people. The estimated ratio of diagnosed to undiagnosed individuals is 1: 8, and the average delay in diagnosis is reported to be 13 years. The contemporary coeliac patient is diagnosed between the ages of 40 and 60, has normal or even high BMI, and subtle symptoms. Patients are often identified in screening groups which would include those with type 1 diabetes, autoimmune thyroid disease, or a first-degree relative with coeliac disease.

The presence of relevant symptoms is not essential for a diagnosis of coeliac disease. Additionally, positive serological testing alone is not sufficient to confirm the diagnosis. Endomysial antibody (EMA) and tissue transglutaminase (TTG) have a combined sensitivity and specificity of >90% when used in combination in selected populations but false positives occur in conditions such as inflammatory bowel disease, and autoimmune disease. Conversely antibody-negative disease accounts for around 9% of cases. IgA deficiency is another cause of false negatives and levels should be checked at the same time as EMA and TTG. It is important that patients consume a gluten-containing diet for six weeks before serological testing and biopsy if necessary.

All those with positive serological tests, and those for whom clinical suspicion is high even if seronegative, should be referred to a gastroenterologist for a duodenal biopsy to confirm or exclude coeliac disease. Diagnosis of coeliac disease requires the demonstration of villous atrophy in the small bowel which improves when gluten is withdrawn.

Contemporary population studies of coeliac disease show only a modest risk of malignancy and death. The overall hazard ratio for death in coeliac disease is reported to be as low as 1.3 with most of the excess risk occurring in the year after diagnosis. While the risk of small bowel cancer may be 40 times that of the general population, it remains a very rare cancer and absolute risk is low. The prevalence of abnormal bone mineral density is reported to be around 40% and may lead to an increased fracture risk. However, this increased risk is small and bone mineral density usually improves or stabilises once gluten has been excluded from the diet.

Patients with coeliac disease should be followed up on an annual basis to allow assessment of adherence to the gluten-free diet, manage complications and detect any micronutrient deficiencies.