

Diagnosing coeliac disease – the key facts

Coeliac disease is common and treatment improves outcomes

- Coeliac disease is an autoimmune disorder occurring in genetically susceptible individuals that results in an abnormal immune response to dietary gluten
- Over 1 in 70 Australians are affected, but the broad clinical presentation means that coeliac disease is often overlooked – 4 out of 5 Australians remain undiagnosed
- Symptoms often go unrecognised or patients may be truly asymptomatic. Targeted screening of at-risk patients is the most effective way to detect coeliac disease
- Untreated coeliac disease is associated with a range of complications, including nutrient deficiencies; premature osteoporosis; abnormal liver function; higher rates of other autoimmune diseases, such as thyroid disease; infertility and poorer pregnancy outcomes; sepsis; and some forms of malignancy, especially lymphoproliferative disorders such as lymphoma
- Strict removal of gluten – a protein found in wheat, rye, barley and oats – can arrest the damaging inflammatory immune response caused by gluten and is important to reduce morbidity and mortality

Symptoms and signs that should prompt testing for coeliac disease:

- Chronic or intermittent gastrointestinal symptoms, such as diarrhoea, constipation, abdominal pain, bloating or flatulence
- Prolonged fatigue (“tired all the time”)
- Iron deficiency anaemia or nutritional deficiency
- Sudden or unexpected weight loss
- Dental enamel defects or mouth ulcers
- Low-trauma fracture or premature osteoporosis
- Infertility, recurrent miscarriage
- Abnormal liver function tests (especially elevated transaminases)
- Peripheral neuropathy, ataxia or epilepsy

High-risk associations that should prompt testing for coeliac disease:

- Family history of coeliac disease (10-20% risk)
- Autoimmune thyroid disease
- Type 1 diabetes
- Other autoimmune disease e.g. Addison’s disease, Sjogren’s syndrome, autoimmune liver disease
- Dermatitis herpetiformis (an itchy, blistering skin condition)
- Immunoglobulin A (IgA) deficiency
- Down’s syndrome
- Turner syndrome

For further information
1300 458 836
www.coeliac.org.au

How to test for coeliac disease:

1. Confirm your patient is consuming a gluten-containing diet for accurate results
(see box below for management if they are already following a gluten free diet).
2. Request **coeliac disease serology**, specifically:
 - i. Transglutaminase-IgA (tTG-IgA) and deamidated gliadin peptide-IgG (DGP-IgG)

OR

 - ii. Transglutaminase-IgA (tTG-IgA) with total IgA level (to exclude the 2-3% of people with coeliac disease who are IgA deficient)
3. In select cases, **HLA-DQ2/8 genotyping** may be performed on blood or buccal scrape.

The HLA DQ2/8 gene test can be useful when screening high-risk individuals, e.g. those with a positive family history, to guide the need for further clinical work-up.

How to interpret these tests:

- If tTG-IgA and/or DGP-IgG is positive refer to a gastroenterologist for confirmatory small bowel biopsy. **Serology alone is insufficient to diagnose coeliac disease.**
- A positive HLA-DQ2/8 gene test is not diagnostic of coeliac disease in isolation (approximately half of the general population are positive).
- **A negative HLA-DQ2/8 gene test has strong negative predictive value (<1% likelihood of coeliac disease being present) and means coeliac disease can be excluded.**
- If coeliac serology is negative but the patient is symptomatic and positive for HLA-DQ2 and/or HLA-DQ8 then consider referral to a gastroenterologist for further work-up.
- A HLA-DQ2 and/or HLA-DQ8 positive relative with normal coeliac serology is at risk for future development of coeliac disease and follow-up is warranted. Repeat screening is recommended if they become symptomatic (*suggestive symptoms indicated over the page*).

If your patient is following a gluten free diet prior to testing:

Option 1 - Recommend a gluten challenge. One option is to recommend 3-6g gluten per day for at least 4 weeks prior to testing. This is equivalent to 2-4 slices of wheat-based bread per day.

Option 2 - If your patient is reluctant or unable to complete a gluten challenge, offer HLA-DQ2/8 gene testing. If HLA DQ2/8 gene testing is negative coeliac disease can be safely excluded. If it is positive, then option 1 is the only feasible diagnostic approach.

Once coeliac disease has been diagnosed:

1. Refer to a dietitian with a special interest in coeliac disease for nutritional education
2. Download the chronic disease management template to guide ongoing follow-up:
www.coeliac.org.au/resources/
3. Provide a Coeliac Australia membership referral letter for ongoing support

References:

1. Anderson RP. *Aust Fam Physician*. 2005 Apr;34(4):239-42;
2. Kenrick K and Day AS. *Aust Fam Physician*. 2014 Oct;43(10):674-8;
3. Steele R et al. *Postgrad Med J*. 2011 Jan;87(1023):19-25;
4. Tye-Din JA et al. *Intern Med J*. 2015 Apr;45(4):441-50;
5. NICE (UK) guidelines: *Coeliac disease: recognition, assessment and management (2015)*. www.nice.org.uk/guidance/ng20