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

Coeliac Australia is not a medical organisation. Persons reading this material should not act solely on it. The advice of a medical practitioner should always be obtained.
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:**