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

The advice of a medical practitioner should always be obtained.



How to test for coeliac disease:

- Confirm your patient is consuming a glutencontaining diet for accurate results (see box below for management if they are already following a gluten free diet).
- 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)
- 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 atrisk 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 8-10g gluten per day for 6 weeks prior to testing. This is equivalent to approximately 4 slices of wheat based bread per day. Our Fact Sheet, *Gluten Challenge*, has more information:

(www.coeliac.org.au/uploads/65701/ufiles/Fact_sheets/GlutenChallenge.pdf)

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