Coeliac Disease – Diagnosing and Testing

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
- Within the population, about 1 in 70 people are affected, but the broad clinical presentation means that coeliac disease is often overlooked – about 4 out of 5 are left 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

How to test for coeliac disease:

1. Confirm your patient is consuming a gluten containing diet for accurate results (see 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 (DGPIgG)

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.

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. Use a coeliac management plan to guide ongoing follow-up

3. Encourage membership of NZ Coeliac Society for ongoing support

There is also a video guide by Dr Jason Tye-Din (gastroenterologist and leading coeliac researcher) available on-line titled "Diagnosing coeliac disease - a brief guide for GPs" – view at https://vimeo.com/157267575

http://www.coeliac.org.au/resources/#video

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