Dear Doctor,

A close relative of your patient has been diagnosed with coeliac disease
Have you considered screening your patient?

- 1st degree relatives have a 10% or greater chance of also being affected by coeliac disease.
- Typical symptoms include gastrointestinal upset, lethargy or anaemia, but symptoms often go unrecognised, or the patient may be truly asymptomatic.
- Early diagnosis and treatment of coeliac disease is important to reduce morbidity such as osteoporosis, malignancy, subfertility and general malaise.
- Targeted screening is the most effective way to detect coeliac disease.

How to test:

1. Confirm your patient is consuming a gluten-containing diet for accurate results (*see over page for management if they are following a gluten free diet*).

2. Request the following blood tests:

- Coeliac serology, *specifically*:
  i. Transglutaminase-IgA (tTG-IgA)
  ii. Deamidated gliadin peptide-IgG (DGP-IgG)
- HLA-DQ2/8 genotyping
  (*useful when screening high-risk individuals e.g. those with a positive family history*)

How to interpret:

- 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 the future development of coeliac disease and follow-up is warranted. Repeat screening is recommended if they become symptomatic (*suggestive symptoms indicated over the page*).

Thank you for your care.

Yours sincerely,

Dr Jason Tye-Din *MBBS, FRACP, PhD*
Chair, Medical Advisory Committee of Coeliac Australia

NATIONAL OFFICE
PO Box 271
Wahroonga NSW 2076
Suite 1, 41-45 Pacific Hwy
Waitara NSW 2077
T 02 9487 5088
F 02 9487 5177
E info@coeliac.org.au
W www.coeliac.org.au

The Coeliac Society of Australia Inc trading as Coeliac Australia ABN 60 103 887 670

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

Option 1. Recommend a gluten challenge: your patient needs to return to a gluten containing diet (equivalent of 4 slices of bread per day (2 for a child) for at least 6 weeks before testing).

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.

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
- Abnormal LFTs (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

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 medical follow-up: http://www.coeliac.org.au/health-professionals/
3. Provide a membership referral letter for Coeliac Australia 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 guidelines, UK (2009). https://www.nice.org.uk/guidance/cg86

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