

Testing and diagnosis of coeliac disease in children

Coeliac disease may cause many symptoms and medical issues in children and adults. Some people with coeliac disease don't suffer obvious symptoms but are nonetheless at-risk for the complications of disease.

Testing for coeliac disease is simple and making the diagnosis is important for improving health outcomes.

See your doctor about coeliac disease testing if your child has any of the following symptoms or issues:

- Persistent, unexplained abdominal or gastrointestinal symptoms
- Faltering growth or failure to thrive
- Prolonged fatigue
- Unexpected weight loss
- Severe or persistent mouth ulcers
- Unexplained iron, vitamin B12 or folate deficiency
- Type 1 diabetes
- Autoimmune thyroid disease
- Dental enamel defects
- Down's syndrome
- Turner syndrome
- First-degree relatives with coeliac disease
- A child who is regularly "out of sorts" or generally unwell

Diagnosing coeliac disease

How to screen for coeliac disease

Initial testing for coeliac disease is a blood test that measures certain antibodies ("coeliac serology").

The test involves the doctor requesting either:

- Transglutaminase-IgA (tTG-IgA) **AND** deamidated gliadin peptide-IgG (DGP-IgG) antibody tests

OR

- Transglutaminase-IgA (tTG-IgA) antibody test **AND** the total IgA level

Positive antibody tests alone are insufficient to diagnose coeliac disease and confirmation of the diagnosis by gastroscopy and small intestinal biopsy is needed (see next page); it is possible to have a positive blood test and not actually have coeliac disease.

These tests are only reliable if your child is regularly consuming gluten. If gluten has been removed from the diet for more than a month prior to testing, the blood test and small bowel biopsy results may be inaccurate or hard to interpret (see Coeliac Australia Fact Sheet: "Gluten Challenge" www.coeliac.org.au/uploads/65701/ufiles/Fact_sheets/GlutenChallenge.pdf).

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

Intestinal biopsy

A small bowel (intestine) biopsy is the gold standard for confirming a diagnosis of coeliac disease. This involves a simple procedure called a gastroscopy (endoscopy) where a flexible tube is inserted through the mouth into the stomach and intestine while the child is sedated (asleep). It is a painless procedure that takes about 10 minutes and allows tiny samples (biopsies) of the lining of the small bowel to be collected. These samples are analysed under the microscope and the presence of typical changes to the lining of the small intestine help confirm the diagnosis of coeliac disease.

This procedure also helps to exclude other diagnoses that may be causing symptoms, e.g. Crohn's disease. Understanding the degree of intestinal damage is helpful for follow-up care and assists in excluding a condition called "potential coeliac disease," where coeliac antibodies are positive but the small intestine is normal.

Gene testing (HLA-DQ2/8)

The "coeliac gene test" tests for the main genes associated with coeliac disease: HLA-DQ2 and HLA-DQ8. At least one of these genes is necessary for coeliac disease to develop, but their presence alone does not mean the person will have (or ever get) coeliac disease.

This test is not always required but may be useful in certain situations, mainly as a way of excluding a diagnosis of coeliac disease. Some guidelines recommend it be used in conjunction with coeliac serology when assessing relatives of a person with coeliac disease to determine their potential risk for developing coeliac disease.

Genotyping is usually performed on a blood sample, although some pathology collection centres can use a buccal scrape (sample from inside the cheek), which avoids the need for a needle.

A negative gene test is very helpful as it means coeliac disease is very unlikely; coeliac disease can be excluded and further testing is not necessary. A positive gene test indicates susceptibility to coeliac disease but does not diagnose coeliac disease. Approximately half of the general population are positive for the coeliac genes but only a small proportion will ever develop coeliac disease.

Frequently asked questions

What if my child has symptoms of coeliac disease but the coeliac antibody blood test is negative?

If your child is symptomatic but serology results are negative, a review by a paediatric gastroenterologist may still be needed. Coeliac serology does not always accurately exclude coeliac disease, i.e. false negative results can occur, so if symptoms are ongoing (and especially if there is a family history of coeliac disease or other risk factors) an appointment with a paediatric gastroenterologist may be warranted. Other causes for the symptoms may also need to be investigated.

The coeliac gene test may be useful in such cases (see section on Gene Testing above).

In cases where symptoms are severe, an urgent referral to a paediatric gastroenterologist is important to achieve a correct diagnosis (coeliac disease or otherwise) and relevant treatment.

What if my child is asymptomatic (has no symptoms) but has other risk factors for coeliac disease, e.g. positive family history?

If your child is symptom free and growing normally but has other risk factor/s for coeliac disease, it is recommended to wait until they are 4 years of age to do initial screening. The antibody blood tests are more accurate from the age of 4 and it can be difficult to justify a blood test earlier if your child does not have obvious symptoms.

If the antibody blood test is normal (negative) at age 4, consider repeating this test in future years, e.g. every 2-3 years during your child's growing years, to ensure a diagnosis is not missed before the adolescent growth spurt.

The coeliac gene test may be useful in such cases (see section on Gene testing above).

Can coeliac disease be diagnosed without small intestinal biopsies?

The European Society for Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN) published diagnostic guidelines in 2012 that suggest coeliac disease can be diagnosed in some children in certain circumstances if the following specific strict criteria are met:

- (i) The child is unwell with symptoms suggestive of coeliac disease
- (ii) The tTG antibody level is more than 10 times the upper limit of normal
- (iii) On a blood sample collected at different points in time, there is a POSITIVE endomysial antibody (EMA) result (another highly specific coeliac antibody test) AND ALSO a POSITIVE HLA-DQ2 and/or DQ8 coeliac gene test result
- (iv) The diagnosis is made by a paediatric specialist/gastroenterologist (not a general practitioner) after detailed consideration of all the circumstances

If these criteria are not all met then the "gold standard" approach based on gastroscopy and small intestinal biopsies is necessary to confirm a diagnosis of coeliac disease.

How well this approach works in Australia and other countries is uncertain due to the limited availability of the EMA test, significant testing variation between laboratories and lack of standardisation of the tTG assay (which means every kit has a different "upper limit of normal"). On this basis, the Medical Advisory Committee of Coeliac Australia recommend further studies to confirm the accuracy of this approach.

The decision to use the above criteria to make a non-biopsy diagnosis of coeliac disease may be appropriate in some situations, but should only be made by a specialist paediatrician (paediatric gastroenterologist).

These non-biopsy guidelines do not apply to adults, in whom small intestinal biopsies are always recommended for confirming the diagnosis.

Does my child with coeliac disease need a follow-up biopsy?

Not usually; if your child with coeliac disease is following a strict gluten free diet, has no troubling symptoms, is growing well, has good nutrient levels and their coeliac antibodies have returned to the normal range, it is very likely they have achieved good healing of their small intestine. A repeat gastroscopy to confirm healing is not required in this situation.

A repeat gastroscopy may be important if your child has ongoing symptoms, persistently abnormal antibody levels or low nutrients, or other issues that need further investigation. Regular follow-up is always important to monitor progress.

A chronic disease management (CDM) template for children has been developed by Coeliac Australia and can be used to guide ongoing care by doctors.