diagnosing coeliac disease

Coeliac Australia

Coeliac disease (pronounced seel-ee-ak) is an immune disease caused by gluten, a component of wheat, rye, barley and oats. When people with coeliac disease eat gluten, an inappropriate immune reaction causes inflammation and damage to the small bowel (intestine). Untreated, coeliac disease can cause a range of symptoms and health problems. Treatment involves lifelong and strict avoidance of gluten in the diet and leads to healing of the bowel and better health.

The lining of the normal small bowel is made up of tiny, finger-like projections called villi, which aid in the digestion and absorption of nutrients from food. Through a microscope, the lining of the small bowel looks rather like a shag-pile carpet, the villi making up the "pile". The entire surface area of a healthy small bowel is very large, comparable in area to a tennis court. In people with untreated coeliac disease, the villi become inflamed, damaged and flattened, like a threadbare carpet. This is referred to as "villous atrophy". As a result of this damage the surface area of the bowel available for nutrient absorption is markedly reduced (to the size of a small tabletop) which can lead to nutrient deficiencies. The bowel inflammation also results in problems that can affect the bones, joints, skin and other organs such as the liver and brain.

Who gets coeliac disease?

People are born with a genetic predisposition to develop coeliac disease. The most important genes associated with susceptibility to coeliac disease are termed "HLA DQ2" and "HLA DQ8". Either one or both of these genes are present in virtually every person with coeliac disease. Various environmental factors are thought to play an important role in triggering or "unmasking" coeliac disease in infancy, childhood, or later in life.

Coeliac disease is not an allergy or an intolerance. Food allergies or intolerances are separate conditions managed differently to coeliac disease, although occasionally they can co-exist.

- Food allergies are an adverse immune response to a component of food and can cause reactions in the skin, respiratory or gastrointestinal tracts. These reactions can be serious and occasionally life-threatening (anaphylaxis). Food allergies are most common in children and can be diagnosed by skin prick or blood (RAST) testing. Treatment is by strict avoidance of the offending food.
- Food intolerance (including "wheat intolerance") is a broad term that refers to unpleasant symptoms brought on by a particular food or food component not due to an allergic reaction. A food and symptom diary in combination with a systematic and supervised elimination and challenge process may be useful in identifying food(s) that precipitate troubling symptoms. Breath hydrogen and methane tests are sometimes used to detect fructose malabsorption or lactose intolerance.

Symptoms

The symptoms of coeliac disease vary considerably. Common complaints include gastrointestinal upset (such as abdominal pain, bloating, diarrhoea, or constipation), lethargy, weight loss, and

Coeliac disease affects approximately 1 in 100 Australians. However approximately 75% remain undiagnosed. This means that around 160,000 Australians have coeliac disease but aren't aware of it.

anaemia (low blood count). Some people suffer severe symptoms, while others are symptom free. Further investigation for coeliac disease should occur if one or more high risk features are present. These include:

- Iron deficiency anaemia
- Gastrointestinal symptoms
- Osteoporosis (thinning of the bones, which increases fracture risk)
- Autoimmune disease (such as type 1 diabetes or autoimmune thyroid disease)
- Weight loss
- Infertility
- A family history of coeliac disease.

Untreated coeliac disease can lead to chronic poor health, osteoporosis, infertility, miscarriage, depression, liver disease, poor dentition and an increased risk of autoimmune disease and some forms of cancer such as lymphoma. Importantly, appropriate treatment with a strict gluten free diet leads to small bowel healing, resolution of symptoms, and a reduction in the long-term risk of these complications.

Although symptoms can vary considerably in coeliac disease, everybody with the condition is at risk of complications if they do not adhere strictly to treatment with a gluten free diet. Since bowel damage can occur in coeliac disease even when symptoms are absent, everybody with coeliac disease, irrespective of the severity of their symptoms, needs to adhere strictly to a gluten free diet.

Diagnosis

As coeliac disease has significant health implications, a definitive diagnosis is paramount. Coeliac Australia's diagnostic postcard provides guidelines for diagnosis of coeliac disease. Contact your state organisation for a copy of the postcard.

The tests for coeliac disease are simple – just follow the steps below…

1. Keep eating gluten

Do not commence a gluten free diet prior to being tested for coeliac disease. If a gluten free diet has already been adopted, the tests used to diagnose coeliac disease are unreliable, and can be falsely negative.

If gluten has been removed from the diet, a normal diet must be resumed for at least six weeks prior to testing. During this 'gluten challenge', four slices of wheat based bread (or equivalent) should be consumed each day (for adults). A gluten challenge can be a daunting prospect for some people who experience unpleasant symptoms. While symptoms may be fairly severe for the first few days of the challenge, they often reduce over time. It is important the gluten challenge is carried out properly to ensure reliable testing results.

2. Blood Tests are used for screening

Blood tests (coeliac serology) are used to screen for coeliac disease. Coeliac serology measures antibody levels in the blood which are typically elevated in people with untreated coeliac disease. The antibodies measured include:

- Anti-tissue transglutaminase antibodies (tTG-IgA).
- Deamidated gliadin peptide (DGP) IgA and IgG. This has superceded the older, less accurate anti-gliadin antibody (AGA) test. The DGP performs similarly to the tTG test. The DGP IgG can pick up individuals with coeliac disease even if they are deficient in IgA antibody. It may also be useful as a screening test in infants.
- Anti-endomysial antibodies (EMA). EMA is similar to tTG but is less commonly tested nowadays.

Total immunoglobulin (IgA) levels are often measured to exclude the 3% of people with coeliac disease who are deficient in IgA.

IgA deficiency renders the tTG-IgA test unreliable. In children under the age of four years the tTG-IgA test is also less reliable. As antibody levels can fluctuate in children, it is suggested the antibody tests be performed on two occasions three months apart.

Importantly, a diagnosis of coeliac disease SHOULD NOT be made on the basis of a blood test alone. Blood tests are prone to error and the presence or absence of coeliac disease should never be based solely on a blood test result. A positive blood test always needs to be followed by a small bowel biopsy to confirm the diagnosis.

While a normal (negative) coeliac antibody result suggests that coeliac disease is unlikely it can miss the diagnosis in up to 20% of cases. Therefore, doctors should consider further investigation of people who have risk-factors (listed under symptoms) for coeliac disease irrespective of their antibody result.

3. A small bowel biopsy is essential

A diagnosis of coeliac disease can only be made by demonstrating the typical small bowel changes of coeliac disease (villous atrophy). This involves a gastroscopy procedure in which several tiny samples (biopsies) of the small bowel are taken. A gastroscopy is a simple day procedure done under light anaesthetic sedation that takes about 10 minutes. In the majority of cases, the bowel damage present in those with untreated coeliac disease is not visible to the naked eye.

The biopsies are examined under a microscope to confirm the presence of villous atrophy.

It is important for patients to keep a record of the pathology report describing the microscopic changes seen by the pathologist in the small bowel biopsies. This information is helpful for doctors when assessing healing of the damage on follow up biopsy, and if there

A quick recap…

- The symptoms of coeliac disease vary considerably
- Do not start a gluten free diet prior to testing
- A definitive diagnosis is important
- Testing is simple
- Coeliac Australia is here to give you support and information – www.coeliac.org.au

is any question about the diagnosis of coeliac disease. The report should indicate the number and quality of the biopsies received by the pathologist.

A repeat biopsy should occur approximately 18 – 24 months after commencing treatment with a gluten free diet to confirm small bowel healing. A healthy looking biopsy is good and means the gluten free diet is being followed adequately – but it does not mean coeliac disease has been cured. Relapse will occur if gluten is reintroduced to the diet. Coeliac disease is for life, and a gluten free diet needs to be followed lifelong to maintain health.

Gene testing (HLA genes)

Gene (HLA) testing is a useful test in select cases when the diagnosis of coeliac disease is unclear. This can occur if the blood or small bowel biopsy results are difficult to interpret, or if adequate gluten was not being consumed to make the test reliable. It is performed on a blood test or cheek (buccal) scraping and can be ordered through your local doctor.

Over 99% of people affected by coeliac disease possess either HLA DQ2, HLA DQ8, or parts of these genes. A negative gene test for HLA DQ2 and HLA DQ8 therefore effectively rules out a diagnosis of coeliac disease. However, since these genes are seen in almost a third of the normal population, their presence only indicates susceptibility to coeliac disease. Just one in 30 people who possess HLA DQ2 or HLA DQ8 will develop coeliac disease. Thus the gene test on its own cannot diagnose coeliac disease and a gluten free diet should never be commenced based solely on a positive gene test. A gluten free diet should only be started after confirmation of coeliac disease by small bowel biopsy.

As the gene test is not dependent on gluten intake, it can be used when people have already commenced a gluten free diet. If the gene test is negative for HLA DQ2 and HLA DQ8 then coeliac

disease can be ruled out. If the gene test is positive for HLA DQ2 or HLA DQ8, then a gluten challenge followed by small bowel biopsy will be required to test for coeliac disease.

Beware of unorthodox diagnosis techniques

There are a number of tests and treatments for allergy, intolerance and coeliac disease that are used in the absence of any scientific rationale. These tests and treatments have been shown to be unreliable when subjected to careful study. Unproven testing methods provide misleading results, delay correct diagnoses and lead to unnecessary and ineffective treatment. The Australasian Society of Clinical Immunology and Allergy (ASCIA) advise against the use of such tests for diagnosis or to guide medical treatment. Such methods may include stool-based tests, Vega testing, iridology, hair analysis or the inappropriate use of tests for food-specific immunoglobulin. For a full list and more information about unorthodox tests and treatments, visit the ASCIA website: www.allergy.org.au.

The benefits of diagnosis

As coeliac disease is a serious medical condition with lifelong implications, a definitive diagnosis is essential. The gluten free diet is not a trivial undertaking and involves lifestyle changes and learning new skills such as reading and interpreting food labels. It should only be undertaken after the diagnosis of coeliac disease has been properly medically established.

- A strict gluten free diet has positive implications for health by reducing the long-term risks associated with coeliac disease. People who have been properly medically diagnosed are more likely to maintain the strictness required to remain healthy.
- By obtaining a proper diagnosis, you can be assured that your symptoms are caused by coeliac disease (and should therefore improve once the gluten free diet is established) and not by another more sinister condition.

- As a genetic condition, there could be implications for your family once a diagnosis of coeliac disease is made. Following a diagnosis of coeliac disease, immediate family members should be screened.
- On diagnosis of coeliac disease, screening for complications and associated conditions such as osteoporosis or autoimmune diseases should occur. Being diagnosed appropriately will ensure this important medical assessment takes place.
- Following a proper diagnosis, a dietitian will be more likely to consider additional food intolerance if symptoms persists despite strict adherence to a gluten free diet.
- In the future, it is likely that any potential non-dietary therapy for coeliac disease, such as a therapeutic vaccine, will only be available to those who have coeliac disease properly proven by biopsy.
- Adjusting to the gluten free diet may seem difficult at first but as your knowledge and confidence grows, managing the diet becomes easier. Advice from a specialist dietitian is invaluable and can greatly enhance the enjoyment to be had from a gluten free lifestyle.

Note: It is recommended that a copy of all coeliac tests including antibody, gene and biopsy tests, be kept by those diagnosed with coeliac disease.

Once you are diagnosed…

Coeliac Australia is here to help you manage your gluten free diet. Your state organisation provides support and information on the disease, the gluten free diet, ingredients, where to buy, cooking and recipes, overseas travel and education and research material. Specific resources for children requiring a gluten free diet are also available.

For more information or to become a member, call **1300 GLUTEN** (1300 458 836) or visit our website **www.coeliac.org.au.**

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Printed June 2011