



Coeliac Disease

Autoimmune & Genetic Testing

- For diagnosis, request antibodies to tTG (Tissue Transglutaminase) rather than endomysium or gliadin.
- For monitoring dietary compliance, request tTG or Gliadin antibody.
- For disease exclusion in high-risk patients, request genetic tests: when gluten diet is refused; or tTG antibody/small bowel biopsy is negative.

A HIGH-RISK PATIENT IS ONE WHO PRESENTS WITH:

1. A genetic predisposition i.e. family history of Coeliac Disease.
2. Classic symptoms of malabsorption:
 - Iron deficiency anaemia
 - Chronic/recurrent diarrhoea
 - Irritable bowel syndrome
 - Weight loss, failure to thrive
 - Pubertal delay
 - Folate, Vitamin E or K deficiency
 - Osteoporosis
 - Hypocalcaemia, Vitamin D deficiency, secondary hyperparathyroidism.
3. Non-classic symptoms arising from associated diseases:
 - Autoimmune liver disease, elevated transaminases
 - Autoimmune endocrinopathy: Type 1 diabetes, Hashimoto's and Addison's disease
 - Sjogren's syndrome
 - Inflammatory bowel disease
 - Neurologic disorders: peripheral neuropathy, epilepsy, ataxia
 - Arthritis of unknown etiology
 - Infertility
 - Down and Turner syndromes.

HOW COMMON IS COELIAC DISEASE?

With a prevalence of ~1% in Caucasians, it is a much more common disease than previously thought. India and the Middle East have a similar prevalence but the disease seems uncommon in East Asia including Japan. Onset is in childhood and in adults with a female:male ratio of 2:1. There is a familial tendency towards Coeliac Disease, with 10-15% of first-degree relatives developing the disease.

HOW DOES COELIAC DISEASE DEVELOP?

Coeliac Disease develops in >99% of persons with HLA DQ2 or DQ8 haplotypes. tTG alters dietary gluten in wheat, barley and rye. Altered gluten peptides form molecular complexes with DQ2 or DQ8 molecules that activate T cells. This leads to inflammation and loss of intestinal finger-like projections (villi), resulting in reduced nutrient absorption.

HOW DOES COELIAC DISEASE PRESENT TO THE CLINICIAN?

Onset of clinical symptoms represents the tip of the 'Coeliac Pyramid' (see Fig. 1). The disease may present with classic malabsorption symptoms, or more typically remains silent, latent or becomes apparent with a wide variety of non-classic, extra-intestinal symptoms arising from pathology in multiple organs as listed earlier. Hence, the diagnosis is likely to be missed unless Coeliac Disease is considered. Undiagnosed disease carries the risk of nutrient deficiency and gastro-intestinal malignancy including enteropathy-associated T cell lymphoma.

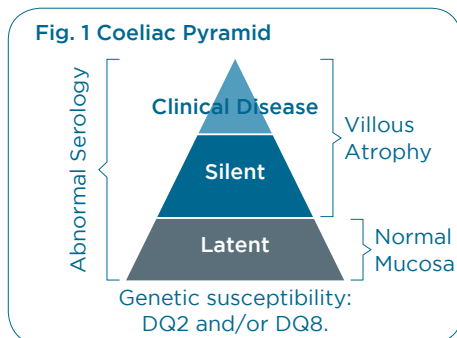


Fig. 2 Coeliac antibody tests

	Endomysia	tTG	Gliadin
Specificity	-100%	97%	70%
Sensitivity	97%	98%	70%
	(adult)	(adult)	
	90%	96%	
	(child)	(child)	

tTG = Tissue transglutaminase (human recombinant)
 Roston et al, Gastroenterology. 2005, 128 (4 Suppl 1): S38-46

WHAT ANTIBODY TESTS SHOULD BE REQUESTED FOR HIGH RISK PATIENTS

- IgA antibody to tissue transglutaminase (tTG)
- IgG antibody to tTG or to deaminated gliadin for patients with IgA deficiency.
- IgG antibody to deamidated gliadin may also identify patients who test negative for tTG antibody.
- Antibody to native gliadin not recommended.

Note that small bowel biopsy remains the gold standard for diagnosis.

WHEN SHOULD GENETIC TESTING FOR DQ2 AND DQ8 BE REQUESTED?

- Antibody to tTG is positive but small bowel biopsy shows a normal mucosa.
- The tTG test is negative in high-risk patients.
- The patient refuses a gluten-containing diet.

Negative testing for DQ2/DQ8 virtually excludes Coeliac Disease.

40% of Caucasians will test positive for DQ2 and DQ8 but only 3% of these are at risk of Coeliac Disease.

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