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Coeliac disease: is it time for mass screening?

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Screening studies indicate a prevalence of coeliac disease (CD) of up to 1% in populations of European ancestry, yet the majority of cases remain undiagnosed. Serological markers for CD now available have high sensitivity and specificity, offering the option of mass population screening. The principles of disease screening as set out by Wilson and Jugner can be applied to CD to predict whether this is appropriate. CD is an important health problem for the individual and the community because of high prevalence, associated specific and non-specific morbidity, and long-term complications of which the most important are gut malignancy and osteoporosis. However, recent studies indicate that the prevalence of malignancy and the health impact of osteoporosis are much less than previously supposed, so the prophylactic benefits of early diagnosis through screening may be low. While CD has an accepted and effective treatment, dietary gluten exclusion, this is difficult for the individual and asymptomatic cases may be poorly motivated to comply. Diagnosis of CD is by histological confirmation on duodenal biopsy. We now recognise milder degrees of gluten sensitive enteropathy without villous atrophy (Marsh I, II lesions) and the benefits to the individual by identifying these early lesions through screening is unknown: whether to treat such individuals needs to be agreed before programmes commence. Screening with serum antibodies is relatively non-invasive but may have to be repeated during each individual's lifetime. HLA typing beforehand to identify the 30% of the population with DQ2 or DQ8, who are at potential risk of CD, will allow one-off exclusion of a large percentage of the population but like all genetic testing has ethical implications. The economic costs of screening and treatment versus morbidity prevented have not been calculated.

Key words: coeliac disease; biopsy; diagnosis; enteropathy; gluten; HLA typing; screening; epidemiology; prevalence; lymphoma; osteoporosis.

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Practice point

- Case finding of symptomatic patients with CD appears to be effective in improving health and preventing complications, but it is not clear whether mass screening has similar benefits

Research agenda

- Extensive research using limited screening programmes in well-defined regions is required to assess the benefits of mass screening. Issues of particular interest include the optimum screening strategy, i.e. serology vs. HLA typing followed by serology, screening age and frequency, approach to milder degrees of enteropathy, and compliance with diet, quality of life and health outcomes in those diagnosed by screening

Coeliac disease (CD) is a hidden public health problem worldwide. Many studies have shown that though CD affects 0.3–1.0% of European or European ancestry populations[1–14] (Table 1), most cases remain undiagnosed. Assuming a prevalence of 0.5%, there are approximately 2.5 million people with CD in Europe alone. The prevalence of CD thus exceeds by far that of a number of diseases for which screening programs are currently applied such as congenital hearing loss (1/1000), congenital hypothyroidism (1/3400) and phenylketonuria (1/18 000). Mass screening is the only way to identify the majority of people with CD.

Table 1. Screening studies for celiac disease in different populations.

Country	Screening method[a]	Number	Prevalence	Reference
Italy[b]	AGA EMA	3351	1:328	1
Northern Ireland[c]	AGA EMA	1823	1:122	2
Finland[c]	EMA	1070	1:130	3
The Netherlands[b]	EMA	6127	1:198	4
Sahara[b]	EMA[d]	989	1:18	5
Spain[c]	AGA EMA[d]	1170	1:389	6
Australia[c]	EMA	3011	1:251	7
Sweden[c]	TGA EMA	1850	1:205	8
Argentina[c]	AGA EMA	2000	1:167	9
Brazil[c]	EMA	2371	1:183	10
USA[c]	AGA EMA	4126	1:133	11
Finland[b]	EMA	3654	1:99	12
England[c]	EMA[d]	7550	1:87	13
The Netherlands[c]	EMA[d]	1440	1:288	14

[a] Determination in serum of IgA antibodies against gliadin (AGA), endomysium (EMA) and tissue transglutaminase (TGA).
[b] Children.
[c] Adults.
[d] Diagnosed not confirmed by small bowel biopsy.