

If a family member has celiac disease, you should get yourself tested

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An Australian-first study, [published in the *Medical Journal of Australia*](#), has found significant prevalence of undiagnosed celiac disease among first degree relatives of people already diagnosed.

The study findings support existing overseas recommendations for screening of first degree relatives for celiac disease.

Celiac disease is an autoimmune disorder where the ingestion of gluten in genetically susceptible individuals causes [gastrointestinal symptoms](#). Delayed diagnosis of the condition can lead to serious health complications such as osteoporosis, infertility, and small bowel cancer.

"We undertook this [research](#) to look at the value of finding cases of celiac disease in people at high risk of celiac disease in Australia," lead author Dr. James Daveson said.

Researchers at the Wesley Research Institute in Brisbane set out to determine the prevalence of undiagnosed celiac disease among first degree relatives of people who had been diagnosed.

A total of 202 first degree relatives ([children](#), siblings or parents) of 134 people with celiac disease were invited to undergo testing for celiac disease. Testing included HLA-DQ2/8/7 [polymerase chain reaction](#) genotyping for celiac disease risk alleles and, where possible, small bowel biopsy.

The study found seven of 62 [child](#) first degree relatives of people with celiac disease had biopsy-confirmed disease, yielding an estimated prevalence of 11%. For those with celiac disease susceptibility haplotypes, the prevalence was 14%.

Dr. Daveson hopes the study will emphasize to health practitioners the importance of family screening for celiac disease. "Defining such a high rate of undiagnosed ... high risk children with celiac disease is important for Australian health practitioners," Dr. Daveson said.

"The most important outcome of this study will be if it highlights the

need for first degree relatives of people with celiac disease to be screened for celiac disease themselves."

More information: Richard Muir et al, Undiagnosed coeliac disease identified by active case finding in first degree relatives of people with coeliac disease in Australia: a prospective observational study, *Medical Journal of Australia* (2023). [DOI: 10.5694/mja2.52105](https://doi.org/10.5694/mja2.52105)

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