New approach to celiac testing identifies more Australians at risk

August 27 2013

Dr. Jason Tye-Din from the Walter and Eliza Hall Institute and colleagues have developed a new approach to detecting celiac disease, revealing this immune disorder is far more common than previously recognized. Credit: Walter and Eliza Hall Institute

Australian researchers have developed a new approach to detecting coeliac disease, revealing this immune disorder is far more common than previously recognised.

In a study of more than 2500 Victorians the researchers combined traditional antibody testing (measuring the immune response to gluten) with an assessment of specific genetic risk markers. They found more than half of Australians had genetic risk factors for developing coeliac disease. The research is published online today in the journal *BMC Medicine*.

Dr Jason Tye-Din from the Immunology division at the Walter and Eliza Hall Institute and Dr Bob Anderson, chief scientific officer at US biotechnology company ImmusanT, worked with Barwon Health, Deakin University, Healthscope Pathology and the University of Queensland Diamantina Institute to develop and trial the new diagnostic approach.

Dr Tye-Din said the new approach of combining the genetic test with a panel of antibody tests would increase the accuracy of testing, decrease overall medical costs by reducing invasive diagnostic tests, and avoid medically unnecessary use of a gluten-free diet.

"Currently, bowel biopsies are recommended for anybody with positive antibody tests," he said. "In this study the inclusion of a simple genetic test helped identify a substantial number of people whose antibody tests were falsely positive and who did not actually require a bowel biopsy to test for the possibility of coeliac disease."

Coeliac disease is caused by an inappropriate immune response to dietary gluten. Gluten can be found in wheat, rye, barley and oats. When gluten is consumed, it can cause a wide range of complaints from chronic tiredness, iron deficiency, osteoporosis, itchy rash, and headaches to various digestive symptoms. Coeliac disease damages the

lining of the small intestine and can lead to significant medical complications such as autoimmune disease, infertility, liver failure and cancer. Coeliac disease usually develops in childhood and is life-long, but early diagnosis and treatment can reduce the risk of adverse health complications.

Dr Tye-Din said the newly developed testing strategy showed coeliac disease potentially affected at least one in 60 Australian women and one in 80 men. Previous estimates had the number of Australians with coeliac disease as no more than one in 100. Although this study is the first to reveal that more than half of Australians have genetic risk factors for developing coeliac disease, it is not yet known why the disease develops in only some people.

Dr Tye-Din, who is also a gastroenterologist at The Royal Melbourne Hospital, said the findings were surprising and shed new light on the medical burden of coeliac disease in Australia. "It is concerning that a significant number of people in the community with coeliac disease have not been diagnosed," he said. "Accurate and timely diagnosis is important for the health of patients with coeliac disease. Making a diagnosis based on a blood test alone or commencing a gluten-free diet without a confirmatory bowel biopsy is inappropriate and can impose an unnecessary and lifelong treatment.

"Although small bowel biopsy is needed to confirm coeliac disease it is costly and invasive. Reducing unnecessary procedures is better for patients, eliminating an invasive test, and better for the economy by reducing healthcare costs," Dr Tye-Din said. "This study provides a strategy to improve the diagnosis of coeliac disease in the community by combining the benefits of antibody and genetic testing."

Provided by Walter and Eliza Hall Institute