Accuracy of Serologic Tests and HLA-DQ Typing for Diagnosing Celiac Disease

What is the problem and what is known about it so far?
Celiac disease is caused by a reaction to gluten, a protein found in wheat, rye, and barley. People with celiac disease who eat food with gluten develop an immune response to the protein. The immune response causes damage to the lining of their small intestine, which makes it difficult to digest food and absorb nutrients. Classic symptoms of celiac disease are abdominal pain, diarrhea, and weight loss. Some people with the disease have no symptoms at all. Some have only mild symptoms that are nonspecific, such as mild fatigue and weakness. Treatment is careful avoidance of foods that contain gluten.

Doctors diagnose celiac disease by taking a tissue sample of the intestinal lining (called small-bowel biopsy). Blood tests for antibodies can also be used to help diagnose the disease. The antibody tests are not a replacement for biopsy, but they can help decide which patients need biopsy. Because celiac disease is hereditary, genetic blood testing may also be useful. It is not clear whether combining antibody and genetic testing is more useful than either test alone.

Why did the researchers do this particular study?
To see whether the combination of antibody and genetic testing is more useful than either test alone for diagnosing celiac disease.

Who was studied?
463 people who were referred to a gastroenterology clinic for testing because they had symptoms that suggested celiac disease.

How was the study done?
The researchers obtained biopsy specimens from all participants. Then they took blood for antibody and genetic tests. Using the biopsy result to diagnose celiac disease, they calculated the proportion of people with the disease who had positive and negative antibody test results and the proportion of people without the disease who had positive and negative genetic test results. Then they compared the results.

What did the researchers find?
Only 16 participants had celiac disease. Some antibody tests were more accurate than others. A positive genetic test result did not always mean that a person had celiac disease. However, a negative genetic test result accurately excluded disease. The combination of antibody and genetic testing was about as accurate as either test alone.

What were the limitations of the study?
The small number of patients with celiac disease made it difficult to compare tests. The findings apply primarily to people referred by their primary care doctors to secondary health care providers. They do not apply to people seeing primary care providers for their symptoms.

What are the implications of the study?
The combination of antibody and genetic testing is about as accurate as either test alone for diagnosing celiac disease. The tests can help doctors decide who needs a small-bowel biopsy. However, neither test is a substitute for biopsy.