

In the News

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RNA Blood Test Can ID Bowel Diseases

A commercial test that looks for genetic material in blood has been shown to have more than 90% accuracy in detecting irritable bowel syndrome and inflammatory bowel disease, researchers have found.



Charles Spurlock, PhD

The RNA biomarker test, from IQuity Inc., is based on work from a group at Vanderbilt University, in Nashville, Tenn., with over 10 years of research funded by the National Institutes of Health.

“We found that there are distinct patterns of RNA expression specific to IBS, Crohn’s and ulcerative colitis,” said Charles Spurlock, PhD, CEO of IQuity and a co-inventor of the test. “We are able to take these RNA signatures and detect the presence or absence of disease at the earliest sign of symptoms. It is this information that is valuable to a physician because it allows them to diagnose and treat the patient earlier in the disease progression.”

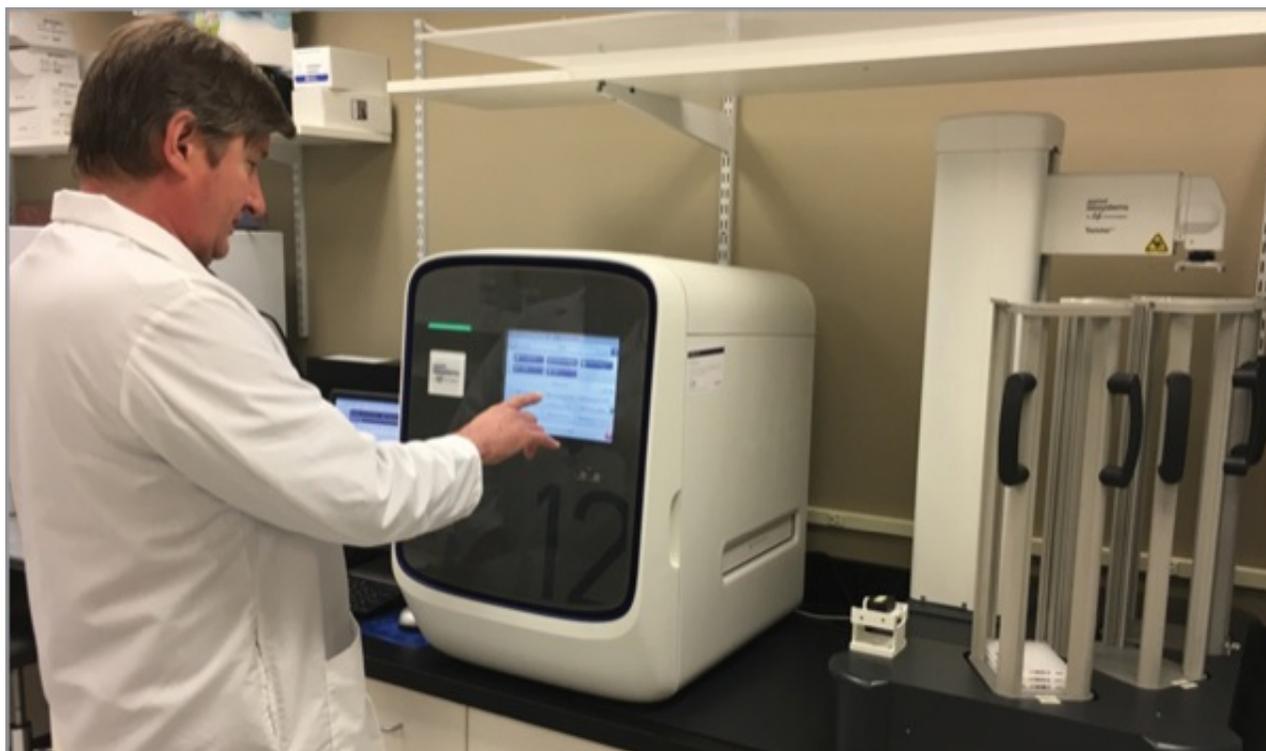
Dr. Spurlock called RNA “a real-time snapshot of what is happening in the cells,” whereas DNA “is not a very dynamic molecule.”

Changes in DNA do not always indicate that a disease will manifest itself, according to Dr. Spurlock. “RNA allows us to take a look at what is happening in the immune system at the time of blood draw. By taking a look at the RNA signatures in the immune cells from both healthy and diseased patients, we have been able to identify a characteristic pattern of expression that we can use ... to differentiate patient samples in each group.”

The latest clinical study included 340 patients who had IBS (n=128), Crohn’s disease (n=89), ulcerative colitis (n=84) and celiac disease (n=39). Another 115 served as healthy controls.

Peripheral whole blood was collected in special tubes and then shipped to a central laboratory in Nashville.

“We look at a panel of RNAs,” said Dr. Spurlock, who was one of the lead investigators of the study, which was presented in November at the 2017 Advances in Inflammatory Bowel Disease (AIBD) conference. “The differences in expression of those RNAs help us determine if a particular blood sample looks similar to one disease or another.”



During the research and development phase, IQiity performed RNA sequencing using whole blood samples to identify gene targets that exhibit the greatest difference between several GI diseases, including IBS, Crohn’s disease and ulcerative colitis.

“We then conducted experiments using quantitative real-time polymerase chain reaction—a gold standard approach for measuring gene expression—to verify the trends we observed in our sequencing studies,” Dr. Spurlock said. “These studies allow us to measure expression of our chosen targets in a larger patient cohort.”

He said different genes “are known to be turned on or turned off, depending on disease state, and these changes or differences analyzed with our machine learning capabilities permit us to distinguish patients with disease from healthy patients.”

IQuity validates its tests by training its algorithms on hundreds of samples from a variety of patient populations, drawn from numerous locations in the United States.

By testing the machine learning algorithm, “we find that this approach can distinguish and capture these differences and correctly classify the individual, based on their RNA profile, thus underlying the robustness of the approach,” Dr. Spurlock said.

The RNA test is intended to support a suspected diagnosis and give providers information that aids their diagnostic workup at the earliest time point possible. It is not intended to replace clinical observation or other tests that a provider would order to give the patient the most accurate diagnosis possible, he said.

For commercial use, test results will be generated and uploaded on a portal for physician review. The turnaround time is approximately seven days from the time the blood sample arrives at the lab.

For clinicians unable to draw blood at their own facility, IQuity will arrange for patients to have their blood drawn by an approved phlebotomist.

“The clinical advantage of our test is that it allows physicians to identify signatures that are consistent with IBS or IBD early in the disease process,” Dr. Spurlock said. “Some doctors report that with colonoscopy, it can be very difficult to visualize certain portions of the small intestine. Similarly, oftentimes it can be challenging to distinguish between IBS from ulcerative colitis and Crohn’s, especially in the early stages of the disease.”

IQuity is currently accepting orders for the test. “Although we are targeting gastroenterologists, we are not eliminating the possibility of including primary care providers as well, because they are often the first line of defense,” Dr. Spurlock said.

“Accurate diagnostic tests able to confidently differentiate IBS from organic gastrointestinal disease represents an unmet need, and efforts to develop such tests continue to progress,” said Brooks D. Cash, MD, chief of gastroenterology and professor of medicine at the University of South Alabama, in Mobile. “Recent examples include tests for postinfectious antibodies as well as stool and serum panels designed to identify common alternative diagnoses in patients with IBS symptoms.”

Although the current study “is intriguing, it is preliminary and additional prospective validation is required in order for this approach to be considered a viable surrogate to replace the current approaches used by clinicians to diagnose IBS,” Dr. Cash said.

—*Bob Kronemyer*

Dr. Cash reported no relevant financial conflicts of interest. He is a member of the editorial board of Gastroenterology & Endoscopy News.