

Researchers develop first molecular test to diagnose eosinophilic esophagitis

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Researchers at Cincinnati Children's Hospital Medical Center have developed the first molecular test to diagnose eosinophilic esophagitis (EoE), a chronic upper gastrointestinal disorder. The incidence of EoE has skyrocketed since it was first characterized two decades ago.

The test, based on a 96 gene expression profile, "offers an unprecedented opportunity to improve diagnosis and treatment, and a platform approach for other inflammatory diseases," says Marc Rothenberg, MD, PhD, director of allergy and immunology at Cincinnati Children's and senior author of the study. Up to now, [gene expression profiling](#) has not yet been well applied to [inflammatory diseases](#), he says.

The study is published in the Dec. issue of the journal *Gastroenterology*. The lead author of the study is Ting Wen, PhD, a researcher in the Rothenberg lab at Cincinnati Children's.

The researchers used bioinformatics tools to develop the EoE diagnostic panel, which is composed of a 96 gene PCR array – considered the most reliable tool for analyzing the expression of a focused panel of genes.

The panel was developed from a set of esophageal biopsy samples from children diagnosed with EoE and from some children who do not have EoE. The researchers applied the panel to 148 children and adults with EoE and found it identified patients with approximately 96 percent sensitivity and 98 percent specificity. The diagnostic panel was also able to distinguish patients in remission from those who did not have EoE.

The researchers have preliminary evidence that the panel could identify patients likely to have disease relapse following treatment.

"We aimed to provide a reliable next-generation

diagnosis of [eosinophilic esophagitis](#), and to offer a component of personalized medicine to each patient," says Dr. Wen. "We hope to ease the suffering of every child with EoE."

EoE is characterized by a large number of white blood cells called eosinophils in the esophagus – the tube that connects the mouth to the stomach. The disease is caused by food allergies and possibly airborne allergens as well. Symptoms typically resemble gastro esophageal reflux but do not get better with anti-reflux medications. These symptoms may include trouble swallowing, vomiting, abdominal pain, and food getting stuck in the esophagus.

In an accompanying editorial, journal section editors Anson W. Lowe and Richard H. Moseley remark that "these findings provide proof of principle for the use of a tissue based molecular test in the diagnosis of EoE and highlight the advantages of such techniques over histologic analysis."

Cincinnati Children's has filed for several patents on the technology, one of which has been issued. The medical center also has entered into an option agreement with Diagnostivus LLC, a specialized molecular diagnostic company focused on delivering personalized diagnostic information and services to physicians treating patients suffering from rare, orphan and less frequent diseases. The option will allow the company to validate the technology and potentially enter into a license agreement.

In addition to Dr. Rothenberg and Dr. Wen, other authors include scientists in the divisions of allergy and immunology; gastroenterology, hepatology and nutrition; and pathology.

Provided by Cincinnati Children's Hospital Medical Center

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