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Genetic Marker for Painful Food Allergy Points to Improved Diagnosis, Treatment

Researchers have identified a genetic signature for a severe, often painful food allergy – [eosinophilic esophagitis](#) – that could lead to improved diagnosis and treatment for children unable to eat a wide variety of foods.

The scientists, from Cincinnati Children's Hospital Medical Center, report in the *Journal of Allergy and Clinical Immunology* that they have pinpointed a dysregulated microRNA signature for eosinophilic esophagitis (EoE), a disease that also may cause weight loss, vomiting, heartburn and swallowing difficulties.

Interestingly, the dysregulated microRNA was reversible with steroid treatment, according to the study's senior investigator, Marc E. Rothenberg, MD, PhD, director of Allergy and Immunology and the Center for Eosinophilic Disorders at Cincinnati Children's. MicroRNAs are short segments of RNA that can regulate whether genetic messengers (mRNAs) are degraded or translated into protein.

“The identification of biomarkers specific to EoE is a significant advancement for both the diagnosis and treatment of the disease,” explains Rothenberg. “The microRNA signature provides an opportunity for more precise analysis of esophageal biopsies.”

Rothenberg said children with EoE now undergo anesthesia and invasive endoscopy to diagnose and monitor the allergy. The ability to determine the presence and status of EoE with a noninvasive method, such as blood test that measures microRNAs, would have a positive impact on individuals and families.

In the current study, investigators analyzed esophageal microRNA expression of patients with active EoE, steroid-induced EoE remission, patients with chronic (non-eosinophilic) esophagitis and of healthy individuals. Additionally, they assessed plasma microRNA expression of patients with active EoE, remission of EoE remission and of healthy individuals.

The researchers found that EoE was associated with 32 differentially regulated microRNAs and distinguishable from the non-eosinophilic forms of esophagitis (such as reflux disease). Esophageal eosinophil levels correlated significantly with expression of the most increased microRNAs, miR-21 and miR-223, and most decreased, miR-375. MiR-223 was also one of the most increased microRNAs in the plasma, along with miR-146a and miR-146b.

Notably, the expression of microRNAs dysregulated in patients with active EoE was normalized in patients with EoE who responded to steroid treatment. This suggests a

significantly specific microRNA signature for disease activity points to its promise for use as a biomarker for EoE.

Only recently recognized as a distinct condition, the incidence of EoE has been increasing over the past 20 years, as have other allergies. Rothenberg and his laboratory team pioneered research showing EoE's reported incidence is estimated to be at least one in 1,000 people. Its hallmark is swelling and inflammation in the esophagus, accompanied by high levels of immune cells called eosinophils.

EoE can affect people of any age, but is more common among young men who have a history of other allergic diseases, such as asthma and eczema. EoE is often first discovered in children with feeding difficulties and failure to thrive, but it is often misunderstood and not well known, delaying proper diagnosis and treatment.

Several organizations provided funding support for the study, which was released online March 3. These include the National Institutes of Health, the Campaign Urging Research for Eosinophilic Disease ([CURED](#)), the Food Allergy Initiative (FAI), and the Buckeye Foundation.

About Cincinnati Children's

Cincinnati Children's Hospital Medical Center is one of eight children's hospitals named to the Honor Roll in U.S. News and World Report's 2010-11 Best Children's Hospitals. It is ranked #1 for digestive disorders and highly ranked for its expertise in pulmonology, cancer, neonatology, heart and heart surgery, neurology and neurosurgery, diabetes and endocrinology, orthopedics, kidney disorders and urology. Cincinnati Children's is one of the top two recipients of pediatric research grants from the National Institutes of Health. It is internationally recognized for quality and transformation work by Leapfrog, The Joint Commission, the Institute for Healthcare Improvement, the federal Agency for Healthcare Research and Quality, and by hospitals and health organizations it works with globally. Additional information can be found at www.cincinnatichildrens.org.

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