

Food allergy-related disorder linked to master allergy gene

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Scientists have identified a region of a human chromosome that is associated with eosinophilic esophagitis (EoE), a recently recognized allergic disease. People with EoE frequently have difficulty eating or may be allergic to one or more foods. This study further suggests that a suspected so-called master allergy gene may play a role in the development of this rare but debilitating disorder.

EoE is characterized by inflammation and accumulation of a specific type of immune cell, called an eosinophil, in the esophagus. Symptoms of EoE vary with age: In young children a major symptom is spitting up food, while in older children and adults, the condition may cause food to become stuck in the esophagus. These symptoms may improve when a person with EoE is restricted to a liquid formula diet that contains no protein allergens or is placed on a diet that lacks six highly allergenic foods (milk, soy, eggs, wheat, peanut and seafood). EoE is not the same as more common food allergies, which also have serious consequences. Little is known about what causes EoE, but the disease runs in families suggesting that specific genes may be involved.

Investigators led by Marc Rothenberg, M.D., Ph.D., at Cincinnati Children's Medical Center Hospital, and supported by the National Institute of Allergy and Infectious Diseases and the National Institute of Diabetes and Digestive and Kidney Diseases, both part of the National Institutes of Health, performed a genome-wide association analysis in children with EoE and healthy children. This type of study detects markers of [genetic variation](#) across the entire human [genome](#) and allows researchers to zero in on a region of a chromosome to identify genes that influence health and the development of disease.

In this study, the investigators identified changes in genes within a region on chromosome 5 that were highly associated with EoE. One of the genes in this region encodes a protein called thymic stromal

lymphopoietin (TSLP). When the investigators measured the expression levels of this gene in children with EoE, they found it was more highly expressed than in [children](#) without the disorder. This result suggests that TSLP plays some role in EoE.

TSLP is made by epithelial cells, which line internal and external surfaces of the body. It has already been described as a master switch that may turn on other [allergic diseases](#), such as asthma and atopic dermatitis (eczema).

Future research is needed to determine if these findings might lead to a genetic test for TSLP and whether drugs that block the production or function of TSLP might be useful in treating EoE.

More information: ME Rothenberg et al. Common variants at 5q22 associate with pediatric eosinophilic esophagitis. *Nature Genetics* [DOI: 10.1038/ng.547](#) (2010).

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