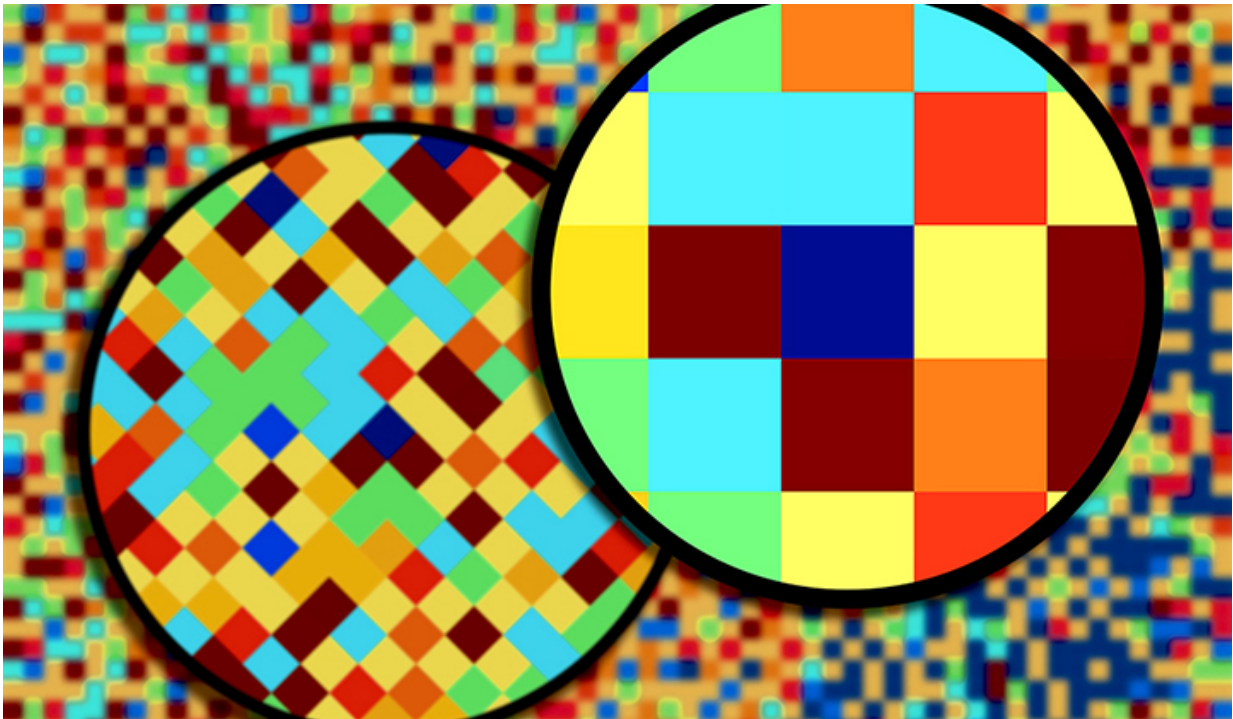


Gene variants linked to multiple sclerosis disrupt key regulator of inflammation

June 11 2015, by Bill Hathaway



Armed with knowledge of the genetic roots of autoimmune diseases like MS, scientists are zeroing in on their molecular causes. Credit: Michael S. Helfenbein

With genetic roots of many autoimmune diseases pinpointed, scientists are zeroing in on the variety of molecular mechanisms triggered by these harmful variants. A team led by Yale School of Medicine researchers has implicated a central regulator of inflammation as a cause of many

cases of multiple sclerosis (MS)—and intriguingly, the researchers note—ulcerative colitis as well.

The study was published in the June 10 issue of the journal *Science Translational Medicine*.

"After identifying the genes that cause MS, we are starting to generate a comprehensive roadmap of the how these genes operate together in allowing immune cells to become activated and attack the myelin," says David A. Hafler, the William S. and Lois Stiles Professor of Neurology and Immunobiology at Yale and chair of Yale's Department of Neurology.

Last fall, a consortium of researchers identified genetic variants that play a role in onset of 21 different [autoimmune diseases](#). Ninety-seven variants were associated with [multiple sclerosis](#). The new Yale research led by Hafler and first author William J. Housley shows that 17 of these MS variants affect the NFkB pathway, which controls a host of immune system responses to environmental threats, and that one variant associated with MS near the NFkB gene profoundly increased gene activity.

The findings illustrate the complexity of individual diseases like MS, in which variants can contribute to small increases in risk of disease through different [molecular mechanisms](#). They also illustrate how same molecular pathways, such as NFkB, can trigger a variety of autoimmune diseases with fundamentally different symptoms—such as MS and [ulcerative colitis](#).

"Identifying these like-minded genes that by themselves contribute only a small risk to disease but together lead to major alterations in immune function may allow a more precise approach in deciding which drug should be used to treat patients," Hafler said.

More information: Genetic variants associated with autoimmunity drive NFκB signaling and responses to inflammatory stimuli, *Sci. Transl. Med.* [DOI: 10.1126/scitranslmed.aaa9223](https://doi.org/10.1126/scitranslmed.aaa9223)

Provided by Yale University

Citation: Gene variants linked to multiple sclerosis disrupt key regulator of inflammation (2015, June 11) retrieved 15 July 2023 from <https://medicalxpress.com/news/2015-06-gene-variants-linked-multiple-sclerosis.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.