

Researchers uncover 48 new genetic variants associated with multiple sclerosis

September 30 2013

Scientists of the International Multiple Sclerosis Genetics Consortium (IMSGC) have identified an additional 48 genetic variants influencing the risk of developing multiple sclerosis. This work nearly doubles the number of known genetic risk factors and thereby provides additional key insights into the biology of this debilitating neurological condition. The genes implicated by the newly identified associations underline the central role played by the immune system in the development of multiple sclerosis and show substantial overlap with genes known to be involved in other autoimmune diseases.

Published online September 29 in the journal *Nature Genetics*, the study, "Analysis of immune-related loci identifies 48 new susceptibility variants for [multiple sclerosis](#)," is the largest investigation of multiple sclerosis genetics to date. Led by the University of Miami Miller School of Medicine, this study relied upon an international team of 193 investigators from 84 research groups in 13 countries and was funded by more than 40 local and national agencies and foundations.

Multiple sclerosis (MS) is a chronic disabling neurological condition that affects over 2.5 million individuals worldwide. The disease results in patchy inflammation and damage to the central nervous system that causes problems with mobility, balance, sensation and cognition depending upon where the damage to the central nervous system occurs. Neurological symptoms are often intermittent in the early stages of the disease but tend to persist and progressively worsen with the passage of time for the majority of patients. The risk of developing multiple

sclerosis is increased in those who have a family history of the disease. Research studies in twins and adopted individuals have shown that this increased risk is primarily the result of genetic risk factors.

The findings released in this study nearly double the number of confirmed susceptibility loci, underline the critical role played by the immune system in the development of multiple sclerosis, and highlight the marked similarities between the genetic architecture underlying susceptibility to this and the many other autoimmune diseases.

The present study takes advantage of custom designed technology known as ImmunoChip—a high-throughput genotyping array specifically designed to interrogate a targeted set of genetic variants linked to one or more autoimmune diseases. IMSGC researchers used the ImmunoChip platform to analyze the DNA from 29,300 individuals with multiple sclerosis and 50,794 unrelated healthy controls, making this the largest genetics study ever performed for multiple sclerosis. In addition to identifying 48 new susceptibility variants, the study also confirmed and further refined a similar number of previously identified genetic associations. With these new findings, there are now 110 genetic variants associated with MS. Although each of these variants individually confers only a very small risk of developing multiple sclerosis, collectively they explain approximately 20 percent of the genetic component of the disease.

Explaining the significance of the work and the nature of the collaboration, the Miller School's Jacob McCauley, Ph.D., who led the study on behalf of the IMSGC, said, "With the release of these new data, our ongoing effort to elucidate the genetic components of this complex disease has taken a major step forward. Describing the genetic underpinnings of any complex disease is a complicated but critical step. By further refining the genetic landscape of multiple sclerosis and identifying novel genetic associations, we are closer to being able to

identify the cellular and molecular processes responsible for MS and therefore the specific biological targets for future drug treatment strategies. These results are the culmination of a thoroughly collaborative effort. A study of this size and impact is only possible because of the willingness of so many hard working researchers and thousands of patients to invest their time and energy in a shared goal."

More information: Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis, [DOI: 10.1038/ng.2770](https://doi.org/10.1038/ng.2770)

Provided by University of Miami

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