

Medical Update Memo

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Two Studies Report Finding New Genes and Gene Regions that Make People Susceptible to MS

Summary

Predisposition to MS seems to be influenced by environmental and genetic factors. Certain variations of a number of genes have been associated with a higher risk of developing MS, most of them being related to the immune system. In this study, two previously unidentified regions of the DNA, located on chromosomes 12 and 20, have been associated with the risk of developing MS. These DNA regions have also been associated with the risk of developing other autoimmune diseases. Identifying all MS genes will likely lead to the development of more effective ways to treat the disease, and open the door to uncovering the cause of MS, which may lead to its prevention. **Nat Genet. 2009 Jun 14**

Details

The IMSGC study:

A major effort has been under way for over a decade to search for the genetic underpinnings of MS – the inherited set of genes that make people susceptible to the unknown triggering factor or factors that lead to the disease. If successful, it would give scientists a roadmap to the cause of MS, as well as to concrete targets for new therapies and possibly even ways to prevent the disease. This painstaking search, involving analysis of the genome (all the genes within the body) and the banking of thousands of DNA samples from patients and family members, was jumpstarted when the International MS Genetics Consortium, (IMSGC) used cutting-edge gene-chip technology to complete the largest replicated whole genome scan for MS yet in 2007. Now, only two years later, the IMSGC and other groups are taking full advantage of these technologies to identify more and more genes that seem to predispose people to MS.

This international group pooled together data from three separate genome-wide studies in people with MS, including 895 people with MS genotyped in its original scan. Genetic material from a total of 2,624 people with MS and 7,220 controls without MS were analyzed, and replicated in a sample of 2,215 people with MS and 2,116 controls without MS. The study confirms that three new genetic variations are associated with MS; these variations exist in the genes for interferon regulatory factor 8, tumor necrosis factor receptor superfamily member 1A, and CD6 – immune messenger proteins. The data may indicate that these proteins are involved early on in the development of MS, note the authors. With support from the Wellcome Trust, the National MS Society and other sponsors, the IMSSGC is conducting a very large scale genetics project to validate their discoveries and identify all remaining genes that exert significant effects on MS susceptibility.

ANZgene study:

The Australian group compared genetic material from 3,874 people with MS and 5,723 people without MS in search of gene variations that appeared to make people more susceptible to developing the disease. They confirmed several gene variations that had been identified in previous studies, including the immune system-related genes CD58, HLA-DR15, IL2RA, and IL7R. The team also identified two loci on Chromosomes 12 and 20 that have been associated with other autoimmune diseases such as rheumatoid arthritis, but not yet with MS. Further study is necessary to determine the exact nature of the genetic variations at work in MS within these loci, but the authors note one interesting candidate -- CYP27B1 on chromosome 12, a gene that instructs vitamin D. There is growing evidence that vitamin D may help protect against the development of MS.

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