GENETICS

UM research uncovers key link to MS

UM researchers have found a gene linked to multiple sclerosis that could have huge implications in treating the disease that affects hundreds of thousands.

BY JOHN DORSCHNER

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After decades of searching, teams of researchers at the University of Miami and elsewhere have found a gene linked to multiple sclerosis that could lead to breakthroughs in understanding and treating the often-devastating disease.

The news is being released today in related reports in Nature Genetics and the New England Journal of Medicine. The studies examined more than 10,000 DNA samples from MS patients in the United States and Europe to discover the genetic link.

Wolfgang Streit, a University of Florida neuroscientist not involved with the research, called the discovery "somewhat spectacular" because researchers have been searching for a link for 30 years.

Ultimately, researchers hope the link between the gene and the disease can help pharmaceutical companies discover ways to treat and perhaps even avoid the onset of MS, which often occurs between the ages of 20 and 40.

"This is the kind of major development that we're expecting from our genetics research," said Pascal Goldschmidt, UM's medical school dean. He lured more than 50 genetics researchers from Duke University to Miami and persuaded the Legislature to give the school $80 million for genetics research.

"This is one of the reasons we came to Miami, to attack common disorders in this way," said Margaret Pericak-Vance, an author on both of the MS papers. ``This is just the tip of the iceberg."

MS is a chronic inflammatory disease of the central nervous system in which the body's own immune system attacks the spinal cord and other areas. According to the National Institute of Neurological Disorders, most patients experience "muscle weakness in their extremities and difficulty with coordination and balance." At worst, the symptoms can impair walking or standing and might even lead to paralysis.

MS affects about 350,000 persons in the United States. What exactly causes MS is unknown. Some researchers believe it may be a combination of genetic and environmental factors. There is no cure.

In the early 1970s, researchers found a link between MS and certain variations in proteins on the surfaces of cells that allow the immune system to distinguish the body's cells from foreign cells. Having this variant increases the likelihood of a person getting the disease four-fold.

For years, geneticists have been looking for other links to MS and have examined more than 100 genes thought to be connected with the disease before finding the interluekin7 receptor (IL7R) alpha chain gene. Having this gene increases a person's chance of getting MS by 20 percent if passed along by one parent, 30 percent if transmitted by both parents.

Supported by grants from the National Institute of Health, researchers from Vanderbilt, UM, the University of California at San Francisco, Duke and the University of Cambridge participated in both studies being published today.

"The genetics of MS has been very difficult to crack," said Jonathan Haines, a Vanderbilt genetics researcher and co-author of the Nature Genetics paper.

MS is a complex disease. Researchers have long known that it tends to be more prevalent in the Northern Hemisphere, so that location, combined with a genetic predisposition, can be a major factor.

Pericak-Vance, co-leader of the overall project, has personally been on the quest for almost 20 decades -- going back to 1988 when she was at Duke. Because there are hundreds of thousands of variations in a person's DNA, finding genetic links to diseases involves lengthy and complex searches.

Goldschmidt, the UM dean, believes Pericak-Vance and colleagues have the greatest potential for breakthroughs in medicine.

"Genetics is fascinating," he said. ``It cuts across all these specialties and can help them all. There's even the possibility to detect heart problems before they lead to death."