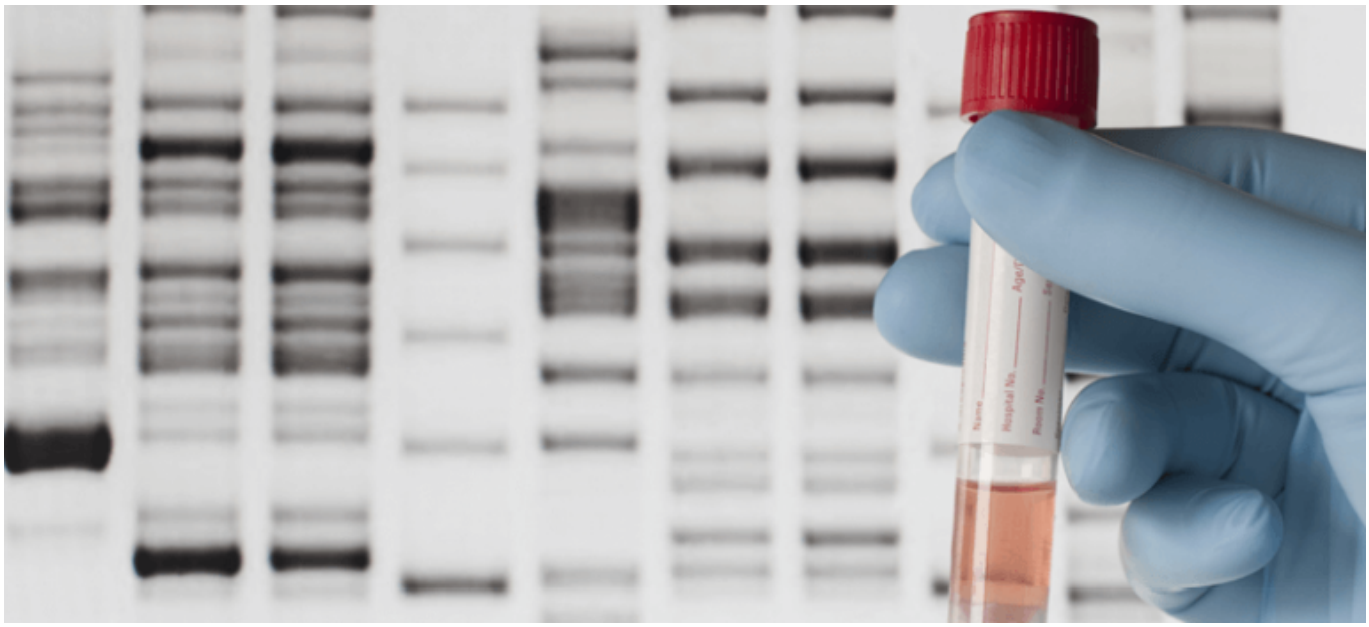
A genetic sleuth: Dr. Philip De Jager



The 2014 Barancik Prize winner is teasing out why some people whose genes predispose them to MS develop the disease while others don’t.

by Vicky Uhland

For more than a decade, Dr. Philip De Jager has been conducting large-scale genetic research designed to solve two of multiple sclerosis’ greatest mysteries: who is most likely to acquire the disease, and how to stop the disease from progressing. Dr. De Jager’s research is so groundbreaking that he’s been awarded the second annual National MS Society’s Barancik Prize for Innovation in Multiple Sclerosis.

Dr. Philip De Jager, winner of the 2014 Barancik Prize, has created an algorithm that uses genetic and environmental factors to identify people who are most likely to develop multiple sclerosis. Photo courtesy of National MS Society

The $100,000 international prize is the largest award ever given to recognize MS research, and is funded by the Charles and Margery Barancik SO Foundation. The Baranciks have been major supporters of MS research projects for more than 20 years, and developed the Barancik Prize to celebrate exceptionally innovative and original scientific research that has the potential to help treat or cure MS.

“We’re thrilled to present the 2014 Barancik Prize to Dr. De Jager for his visionary approach toward understanding the genetic architecture of MS,” says Dr. Timothy Coetzee, Chief Advocacy, Services and Research Officer at the Society. “Dr. De Jager has leveraged his deep understanding of the clinical context of MS with his background in molecular genetics and immunology to design new ways of approaching and answering challenging MS questions.”

Dr. De Jager’s research into MS began in 2003, when he was still a neurology resident at the prestigious Brigham and Women’s Hospital in Boston. He became a founding member of the International Multiple Sclerosis Genetic Consortium (IMSGC), which then tapped him to lead a massive study designed to identify variations in genes that could make someone susceptible to contracting MS. Using data from more than 34,000 people with MS and another 40,000 people without the disease, Dr. De Jager and the IMSGC have to date identified more than 150 different genetic variants that are associated with MS susceptibility.

Dr. De Jager’s next step was to develop resources and tools to help him discover more about these genetic variants. “The variants are all present in the general population, but we wanted to find out why only a small number of people who have them develop MS,” he says.

So Dr. De Jager created the PhenoGenetic Project—a group of more than 1,700 people who donate blood samples several times a year for his and other scientists’ genetic research. Then he developed an algorithm—or mathematical formula—that uses genetic and environmental factors to identify people who are most likely to contract MS. To test the algorithm, he started the Genes & Environment in MS (GEMS) study. This ambitious study is expected to span at least 20 years and include more than 5,000 people who have a family member with MS—which Dr. De Jager says makes them 30 percent more likely than the

general population to develop the disease.

Currently, the only way to monitor people who have a high susceptibility of developing MS is through magnetic resonance imaging (MRI) scans that can identify MS-like lesions. “But at that stage, it’s too late to prevent the disease,” Dr. De Jager says. “We need to find other signs that people are on the way to developing MS.”

Dr. De Jager believes genetic data and the algorithm he developed will help researchers identify those signs. It’s already known that certain blood proteins and the genetic signposts are different in people with MS, so his theory is that over time, the blood and immune-system profile of people who are most susceptible to developing MS will change—but before they develop the disease.

The GEMS study could “open up the understanding of the transition from health to MS and, ultimately, the development of strategies for primary prevention of MS,” says Dr. Dennis Selkoe, a Harvard Medical School professor who nominated Dr. De Jager for the Barancik Prize. “This is an ambitious goal, but Phil has the rare ability to translate a compelling vision into a realizable study.”

Dr. De Jager is also dedicated to developing a reliable, effective way to measure the progression of MS. He’s conducting research analyzing tools like websites and smartphone apps that people with MS can use to track symptoms such as gait changes and sleep patterns that are difficult to measure in a clinical setting.

“We’re working on capturing data in real time in the patient’s natural environment, and then we can do an average measurement to see how the patient is behaving over time,” he says. “This can help us with studies that give new insights into MS progression.”

Dr. De Jager believes that ultimately, finding new ways to measure MS susceptibility and progression will help scientists better understand the biology of the disease and identify treatments.

“Basically,” he says, “I want to find a way to stop MS in its tracks, especially for people who are most at risk.”

Vicky Uhland is a freelance writer and editor in Lafayette, Colorado.

To learn more about the Barancik Prize, visit nationalmssociety.org/Barancik.

Read more about last year’s Barancik Prize winner, Dr. Jonah Chan.