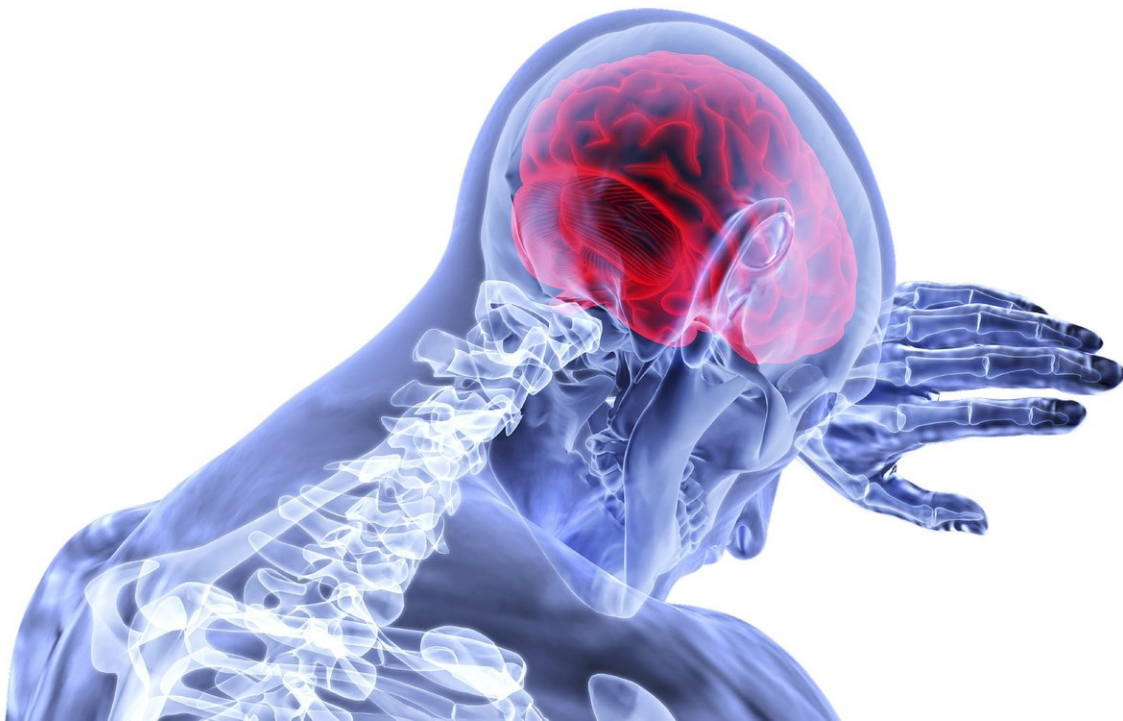


Gene vital for post-stroke recovery identified for the first time

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Having certain specific variants of the PATJ gene predisposes ischemic stroke victims for worse recovery. Seven out of 10 patients with these variants suffer severe sequelae three months after having a stroke and are thus placed in a situation of medical dependence, compared to less than half of patients who do not present these variants.

This is according to data from an international, multicentre study coordinated by researchers at the Hospital del Mar Medical Research Institute (IMIM) and doctors from the Hospital del Mar, published in the journal *Circulation Research*. This is the most important research carried out so far in the field of genetics and stroke prognosis, and the first published. It uses data from more than 2,000 patients and involves 12 centres from around the world.

Dr. Jordi Jiménez Conde, coordinator of the study and attending physician at the Hospital del Mar's Neurology Department, said, "It is the largest study published to date on the genetics and prognosis of stroke, and the first that has found consistent results and been replicated in different countries."

Ischemic stroke is caused by the blockage of a cerebral artery and represents 88 percent of total strokes. The researchers analysed the degree of disability after three months of more than 2,000 patients with [ischemic stroke](#) by studying multiple clinical factors and [genetic data](#). Specifically, they analyzed more than 5 million genetic variants for each individual.

"The PATJ gene shows several variants that significantly influence recovery in [patients](#)," says Dr. Jiménez Conde. It is a gene involved in cell binding, is strongly expressed in nervous tissue, and has been associated with sleep disorders and obesity.

Genetic influence on recovery

"In this study, we identified a set of genetic variants that are relatively common in the population, and which are associated with worse recovery from stroke after three months," says Dr. Marina Mola-Caminal, first author of the study and a researcher in the IMIM's Neurovascular Research Group. This leads the way to studying the

mechanisms used by the PATJ gene to influence this process. "In the future, these variants could be used as biomarkers for stroke sufferers, and, depending on the presence of risk alleles (alternative forms of a gene) in each individual, rehabilitation strategies could be personalised."

Dr. Conde says, "We are able to indicate a region of the genome that is heavily involved in neuroplasticity and neuroregeneration processes, and perhaps if we understand the pathways, we will be able develop new treatments that use this gene as a therapeutic target and help improve patient prognosis."

The study also involved Dr. Israel Fernández, co-researcher in the study and participant in its design and coordination. He says, "All neuroprotective drugs tested to improve post-[stroke](#) recovery have failed. For that reason, this study, utilising mass genetic strategies, may be the first step toward developing new drugs that are truly effective."

More information: PATJ Low Frequency Variants Are Associated with Worse Ischemic Stroke Functional Outcome: A Genome-Wide Meta-Analysis, *Circulation Research* (2018).

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