



Institut Hospital del Mar
d'Investigacions Mèdiques *Press release*

Study published in Circulation Research

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

- Patients with certain variants of the *PATJ* gene have less ability to recover after suffering a stroke
- 7 out of every 10 patients who have this genetic variation are left with serious consequences three months after having a stroke, compared to less than half of sufferers who do not present this variation
- These variants could indicate targets for future treatments that improve stroke prognosis, and enable personalised rehabilitation strategies
- This has been revealed by the first multicentre study in this field, using data from more than 2,000 individuals and involving 12 centres around the world. The work was led by researchers from the Hospital del Mar Medical Research Institute as well as doctors from Hospital del Mar, with Genestroke Consortium groups and the collaboration of the International Stroke Genetics Consortium. The study was funded by La Marató de TV3

Barcelona, 19th November, 2018. - Having certain **specific variants of the *PATJ*** gene predisposes to worse recovery from ischemic stroke. **7 out of 10 patients** with these variants suffer severe sequelae three months after having a stroke, in other words, they are in a situation of dependence, compared to less than half of patients who do not present these variants. This is 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. The study was carried out thanks to the help of the 2010 edition of [La Marató de TV3](#).

Dr. Jordi Jiménez Conde, coordinator of the study and attending physician at the Hospital del Mar's Neurology Department, explained that ***"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."*** They analysed the degree of disability after three months of more than 2,000 patients with ischemic stroke —caused by the blockage of a cerebral artery and which represents 88% of total strokes— by studying multiple clinical factors and genetic data. Specifically, more than 5 million genetic variants were studied 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 that has a strong presence in nervous tissue and which has already 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", stresses 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***



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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."

At the same time ***"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"***, notes Dr. Jiménez Conde.

The study also involved Dr. Israel Fernández, co-researcher in the study and participant in its design and coordination. He is currently at the Hospital de la Santa Creu i Sant Pau Research Institute but at the time of the study was part of the Vall d'Hebron Research Institute (VHIR) and Mutua de Terrassa Neurovascular Research Laboratory. Dr. Fernández comments that ***"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 towards developing new drugs that are truly effective."***

Also vital in the study were Dr. Raquel Rabionet, from the Centre for Genomic Regulation (CRG), currently at the University of Barcelona and linked to the Sant Joan de Déu Research Institute, and Dr. Cristòfol Vives, from the Son Espases University Hospital.

The impact of stroke

Stroke, or cerebrovascular disease, is the leading cause of disability in adults in the world, with 15 million people affected each year, of which 5 million are permanently disabled. The degree of functional recovery has a major impact on the quality of life of patients and families, as well as public health systems. The cost of this disability is estimated at between €12,000 and €17,000 per patient/year. In Catalonia, where 13,000 people are hospitalised due to this annually, the figure totals €54 million euros each year.

Reference article

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Further information

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