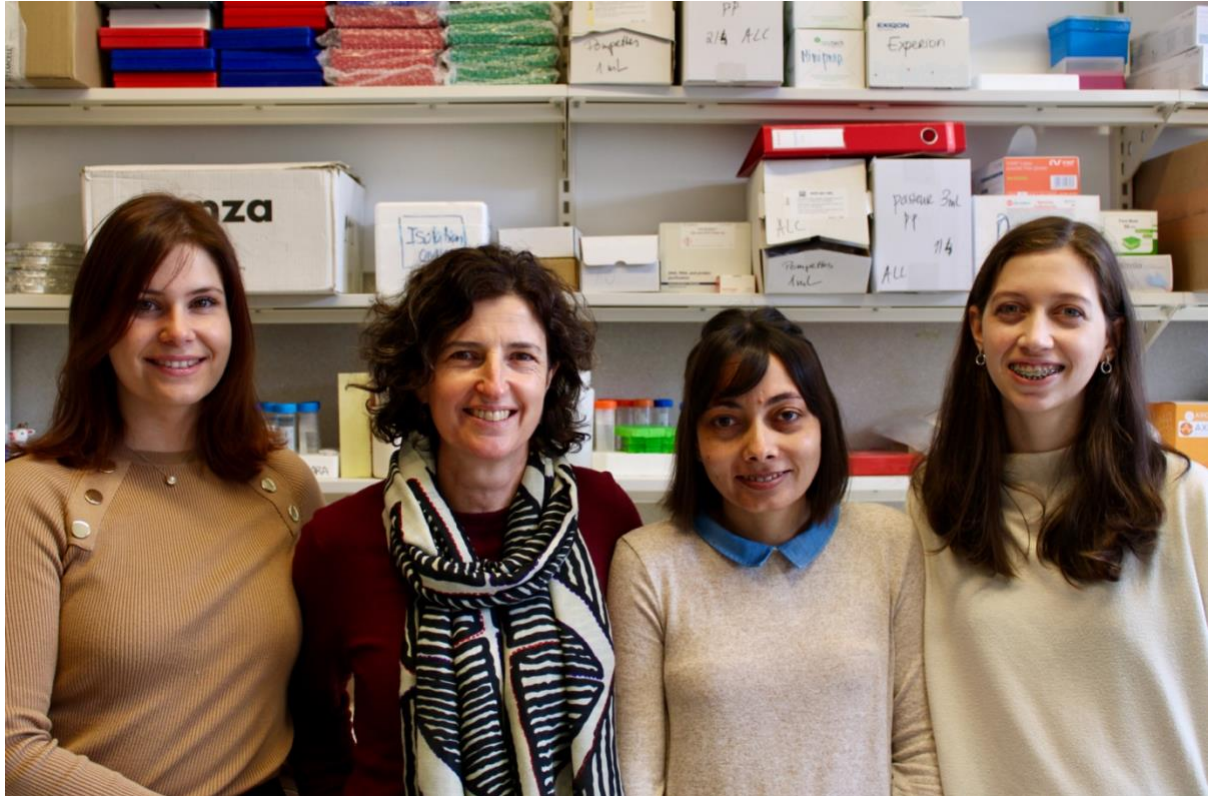


Scientists from the University of Coimbra discover cognitive changes caused by a genetic variant linked to intellectual disability



A study carried out by neuroscientists at the University of Coimbra (UC) reveals altered cognitive abilities and social behavior in mice that express a genetic mutation related to intellectual disability.

Intellectual disability results from changes in neurodevelopment and is characterized by compromised intellectual function, as well as changes in communication and social skills. There are several institutions in Portugal that develop their action within the scope of enabling and integrating people with intellectual disabilities, most of them associated with HUMANITAS – Portuguese Federation for Mental Deficiency.

This neurodevelopmental disorder can have several causes, including genetic and/or environmental ones, with little known about the biological mechanisms that cause the cognitive, behavioral and social changes in these people. Unraveling these mechanisms is essential to design therapeutic and rehabilitative strategies, whenever they appear as necessary.

Genetic changes, associated with neurodevelopmental disorders, such as intellectual disability, are often found in genes that encode proteins present at the synapse, the communication structure between neurons within the brain. One of these proteins is stargazine, which was the subject of a study carried out at the Center for Neurosciences and Cell Biology of the University of Coimbra (CNC), with the aim of **“understanding the impact that a change in the gene encoding stargazine can have on communication and structure of neurons, as well as in animal behavior”**, explains Ana Luísa Carvalho, leader of the study and professor at the Department of Life Sciences (DCV) at the Faculty of Science and Technology of the University of Coimbra (FCTUC).

The work, already published in the journal *Molecular Psychiatry*, had as its starting point a computer simulation that made it possible to predict the impact of the genetic alteration associated with intellectual disability on the structure and molecular dynamics of stargazine. Using cultured rat neurons and genetically modified mice, in which the human genetic alteration was introduced (knock-in animals), the authors of the study found that the genetic variant that leads to the production of a modified stargazine compromises the transmission of information between neurons of the hippocampus, a region of the brain that, in both rodents and humans, is responsible for memory formation and the ability to learn. The genetically modified mice also showed memory and learning difficulties, as well as abnormal social behavior. **“These results show that an alteration in the gene encoding the protein stargazine causes changes in cognitive functions, similar to those observed in people with intellectual disabilities, and causes changes in the transmission of**

information between neurons in the hippocampus as well as in the plasticity of this transmission”, says Ana Luísa Carvalho.

This study **“also allowed us to understand the role of the stargazine protein in neuronal communication in a specific subregion of the hippocampus. It is a study with a dual contribution to the understanding of the functioning of synaptic communication and to the identification of biological mechanisms associated with intellectual disability”,** she concludes.

This work, which was conducted by CNC researchers Gladys Caldeira and Ângela Inácio, is the result of a collaboration between the research group led by Ana Luísa Carvalho and the groups led by Irina Moreira and João Peça, also CNC researchers and DCV teachers at the FCTUC. The study was funded by the Brain and Behavior Research Foundation (USA), the Jérôme Lejeune Foundation (France), the Science and Technology Foundation (Portugal) and the La Caixa Foundation.

The article can be consulted [here](#).

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