Discovered a genetic biomarker that detects Lewy body dementia

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The Germans Trias i Pujol Health Sciences Research Institute (IGTP) and the Universitat Autònoma de Barcelona (UAB) have discovered the first genetic biomarker to detect Lewy body dementia (LBD), a disease that can be confused with Alzheimer's. This biomarker is found in 20% of cases and differentiates one of the sub-groups of the pathology. Licensed to the Grifols company, it will lead to more precise diagnosis and treatment.

Lewy body dementia (LBD) is the second cause of dementia after Alzheimer's disease. The symptoms of both diseases are very similar, since in both cases there is a gradual deterioration in mental capacity, affecting memory, thought processes, behaviour and physical activity. These similarities mean that some patients with LBD are wrongly diagnosed and treated with the usual drugs for Alzheimer's. But this treatment causes adverse reactions in approximately half of these patients, making the disease much worse in some cases.

Currently there is no specific test to diagnose LBD. In practice, various neurological and neuropsychological tests are used to detect the disease and its possible overlap with other disorders, but clinical diagnosis of LBD is not very accurate.

The research, conducted by the IGTP and the UAB, has led to the discovery of the first genetic biomarker, found in 20% of LBD cases, and differentiating between one sub-group of LBD and Alzheimer's disease. "Although this marker only detects a certain number of LBD

sufferers, it significantly increases diagnostic sensitivity to the disease and these patients can get an accurate diagnosis and therefore the right treatment", explains Dr Katrin Beyer, head of the research project and belonging to the Group of Structural and Molecular Pathology, Department of Pathology at the Germans Trias Hospital and Institute.

The researchers first detected the marker through a study of post mortem brain samples, in which they observed an alteration in the expression of the enzyme butyrylcholinesterase (BCHE) in the brains of patients with LBD. These data indicated that there could be genetic alterations in the BCHE gene promoter, causing changes in the expression of the gene. In fact, they found four polymorphisms in the LBD promoter region that, in certain combinations, are associated with LBD. These findings, which have been patented, make it possible to determine if a patient has LBD, distinguishing it from Alzheimer's disease.

Currently, the patent is in its last stage of validation, which is being carried out in collaboration with neurologists from the Neurodegenerative Disease Unit of the Germans Trias Hospital and Bellvitge Hospital.

The licensing agreement with the Grifols company means the results can be applied, thus providing a simple, rapid, and effective procedure for diagnosing LBD in hospitals. Moreover, the marker can also be used to design clinical studies to help identify groups of patients with a more accurate diagnosis, removing, for example, LBD cases from a group of Alzheimer's patients.

Grifols is a global company that for over 70 years has been providing therapeutic treatments with plasmatic proteins, technology for clinical diagnosis and pharmaceutical products for hospital use. It is now the third worldwide producer of biological drugs derived from plasma, is

present in over 100 countries and is a world leader in plasma supplies, with 150 blood donation centres in the United States.

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