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New subtype of ataxia identified

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Researchers from the Germans Trias i Pujol Health Sciences Research Institute Foundation (IGTP), the Bellvitge Biomedical Research Institute (IDIBELL), and the Sant Joan de Déu de Martorell Hospital, has identified a new subtype of ataxia, a rare disease without treatment that causes atrophy in the cerebellum and affects around 1.5 million people in the world. The results have been published online on April 29 in the journal *JAMA Neurology*.

The cause of ataxia is a diverse genetic alteration. For this reason it is classified in subtypes. The new subtype identified described by the researchers has been called SCA37. The study has found this subtype in members of the same family living in Barcelona, Huelva and Madrid and Salamanca (Spain). The finding will allow in the medium term that these families and all who suffer the genetic alteration identified will have personalized therapies and diagnostics prior to the development of the disease. The study was funded by La Marató de TV3 (the Catalan public TV) in 2009, dedicated to rare diseases.

The cerebellum is a part of the brain located behind the brain that, among other functions, coordinates the movements of the human body. When it is atrophied, movement disorders appear, and when the ataxia evolves, the patients suffer frequent falls and swallowing problems. Progressively, they end up needing a wheelchair. Until now, there have been identified more than 30 different subtypes of ataxia, the first of which was described in 1993 by Dr. Antoni Matill, head of the Neurogenetics Unit, IGTP, and Dr. Victor Volpini, head of the Center for Molecular Genetic Diagnosis at IDIBELL.

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Particular eye movements

The first symptoms of ataxia may develop during the childhood or adult stage, depending on the subtype. The SCA37 subtype, the first cases of which were identified by Carme Serrano, neurologist at the Sant Joan de Deu Hospital, Martorell (Barcelona), is expressed at 48 years on average. One of the features of SCA37 subtype is the difficulty for vertical eye movements. Besides the patients identified in Spain by Dr. Serrano and Germans Trias and IDIBELL researchers, there are evidence of the existence of more people affected with this subtype of ataxia in France, Holland and Britain, and for this reason it seems to be a quite prevalent subtype of ataxia in Europe.

All SCA37 patients have a common genetic alteration in the portion 32 of the short arm of chromosome 1, wherein there are a hundred genes. Currently, researchers are sequencing it with new generation technologies to find the specific mutation that causes ataxia. When it is found it will be possible to make an accurate diagnosis in family members who do not yet have developed symptoms. Also, it will be possible to investigate the biological mechanisms that cause ataxia in order to develop and implement personalized therapies, with drugs or stem cells therapy.

Provided by IDIBELL-Bellvitge Biomedical Research Institute