Discovered a cause of mental retardation and autism

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The term intellectual disability covers a large number of clinical entities, some with known cause and others of uncertain origin. For example Down syndrome is due to an extra copy of chromosome 21 and Rett syndrome is in part caused by a mutation in the control switch gene called MeCP2.

In other cases the mechanisms by which they are produced are not clearly identified. It is the case of most of those disorders classified under the large umbrella of autism. An study published in the journal *Genetics in Medicine*, by Manel Esteller, director of the Program Epigenetics and Cancer Biology (PEBC) of the Bellvitge Biomedical Research Institute (IDIBELL), ICREA researcher and Professor of Genetics at the University of Barcelona , has discovered a mechanism that identifies a cause of intellectual disabilities in these puzzling cases.

"We have analyzed the genome of 215 patients with mental retardation, autism or Rett syndrome, in which they had not found any genetic alteration in the genes classically associated with these clinical conditions, to see if we could find a molecular cause. And this process has allowed us to detect a new mutated gene that could be causing these disorders. "He explained Manel Esteller.

"Specifically, the identified gene is called JMJD1C (Jumonji Domain Containing 1C) and is an epigenetic gene which its normal function is to control the activity of other genes. Only a small percentage of mental retardation of unknown origin is due to mutation of this gene. This

finding suggests that many genes with low frequency disturbance are responsible for cases with unknown cause. They have demonstrated that this gene joins the MeCP2 gene so it could also contribute to cases of atypical Rett Syndrome "says Esteller.

"This study has been possible thanks to the support received from the European Union, the Catalan and Spanish associations of Rett syndrome, a campaign of crowdfunding in Verkami and Foundations Daniel Bravo and Finestrelles. Demonstrating the involvement of civil society in research" concludes the researcher.

More information: *Genetics in Medicine*, doi: 10.1038 / gim.2015.100, 2015.

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