

## **Researchers account for the complex symptoms of Angelman syndrome**

## April 20 2018

A research group at the Faculty of Science and Technology of the UPV/EHU-University of the Basque Country has managed to identify the changes in the proteins altered by the UBE3A enzyme, which is responsible for Angelman syndrome. This disease causes problems in intellectual and motor development, epilepsy, difficulties in communication, and insomnia.

The group, led by the Ikerbasque professor Ugo Mayor of the UPV/EHU's Department of Biochemistry and Molecular Biology, has just published an explanation of the mechanisms affected by Angelman syndrome in the journal *Human Molecular Genetics*. The researchers identified the changes in the proteins altered by the UBE3A enzyme, the malfunctioning of which leads to <u>brain disease</u>.

According to these new results, UBE3A is responsible for regulating the function of proteasome, a kind of shredding machine that regulates the balance of the other proteins in the cells. In this indirect way, UBE3A is responsible for the stability of a huge number of processes that take place within the cells. When there is a fault in the UBE3A, these processes do not take place correctly. This explains the complexity of the syndrome that emerges when the UBE3A enzyme fails to perform its function properly. The genetic origin and symptoms of this disease have been studied previously, but until now, no explanation has been forthcoming regarding how a single gene was capable of creating so many alternations in brain function.



Angelman syndrome is a disease affecting one in every 15,000 newborn babies. It causes complex problems in intellectual development, epilepsy, as well as difficulties in communicating, lack of motor coordination, and problems in balance and movement accompanied by extremely few hours of sleep. All this is caused by the failure of the UBE3A gene in the brain.

**More information:** Juanma Ramirez et al, Quantitative proteomics reveals neuronal ubiquitination of Rngo/Ddi1 and several proteasomal subunits by Ube3a, accounting for the complexity of Angelman syndrome, *Human Molecular Genetics* (2018). DOI: 10.1093/hmg/ddy103

## Provided by University of the Basque Country

Citation: Researchers account for the complex symptoms of Angelman syndrome (2018, April 20) retrieved 26 April 2024 from <u>https://medicalxpress.com/news/2018-04-account-complex-symptoms-angelman-syndrome.html</u>

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