CIC bioGUNE
Biozientzietako Ikerkuntza Kooperatiboko Zentroa
Centro de Investigación Cooperativa en Biociencias

PRESS RELEASE

A prestigious American foundation is to fund research at Biogune into Angelman Syndrome

- *The group led by Dr. Ugo Mayor aims to identify the protein substrates responsible for this genetic disorder by using a new strategy for capturing neuronal ubiquitin conjugates in the fruit fly.*
- *The March of Dimes Foundation, which will provide 150,000 USD over two years, is a health-related, United States non-profit organisation whose goal is to improve infant health by preventing birth defects and infant mortality.*
- *Angelman Syndrome is a rare disease that affects 1 out of every 15,000 newborns. Infants with AS present delayed development, limited language skills and poor motor coordination.*

(Bilbao, 13 February 2012).- The prestigious American March of Dimes Foundation is to fund a research project at the Basque centre Biogune, led by Dr. Ugo Mayor, which aims to identify the protein substrates responsible for the Angelman Syndrome, a genetic disease that affects newborns, by using a new strategy for capturing neuronal ubiquitin conjugates in the fruit fly model system.

The March of Dimes Foundation is a health-related, United States non-profit organisation whose goal is to improve infant health by preventing birth defects and infant mortality. Founded in 1938, March of Dimes provides funding for research programmes, community services, education and promotion.

Mayor's research group at the Biogune bioscience research centre, located in the Bizkaia Technology Park, is devoted to studying the role played by a specific molecular mechanism known as ubiquitination in the development and functioning of the nervous system.

In their most recent paper, published in May 2011, the group reported the active presence of various proteins responsible for catalysing the incorporation of ubiquitin into the nervous system of the fruit fly, a model system which is widely used due to its

genetic similarity to humans and having a relatively complex nervous system that is nevertheless fairly easy to study.

One of the proteins identified was found to be homologous to the product of the gene responsible for Angelman Syndrome (AS), a genetic disorder with a prevalence of approximately 1 out of every 15,000 newborns. Infants with AS present delayed development, limited language skills and poor motor coordination, as well as balance and movement problems. People affected with this syndrome also have a tendency to laugh frequently, present attitudes that transmit overwhelming happiness and have a high capacity for receptive, non-verbal communication.

First described in 1965, the cause of AS was subsequently identified in 1997 as the mutation of a gene known as UBE3A (located on chromosome 15q), which codes for an ubiquitin ligase that is responsible for binding ubiquitin molecules to other proteins.

In the opinion of the project leader, Dr. Ugo Mayor, “the substrate proteins modified by this ubiquitin ligase in the brain are still unknown, therefore their identification could help us to understand how the syndromes described come about”.

In light of this, Mayor's team, which uses an experimental model that they consider to be suitable for solving this problem, decided to submit a project to the March of Dimes Foundation. The main goal of this project is to identify the substrates of the protein responsible for Angelman Syndrome using a new strategy for capturing neuronal ubiquitin conjugates in the fruit fly model system.

The March of Dimes Foundation will provide 150,000 dollars worth of funding for this research project over the next two years.

“It is plausible that, even when we finally identify the neuronal substrates of UBE3A, we will be unable to develop a treatment for Angelman Syndrome, but if even a small possibility exists, this will only become clear once we understand the molecular mechanisms associated with this disease”, concludes Dr. Mayor.

For more information regarding neuronal ubiquitin pathways, browse to laboratory’s website.