



Master project 2021-2022

Personal Information

Supervisor	Andrés Ozaita
Email	andres.ozaita@upf.edu
Institution	UPF
Website	https://www.upf.edu/neurophar
Group	Laboratory of Neuropharmacology

Project

Computational systems biology

Project Title:

Intellectual disability can be caused by genetic mutations

Keywords:

intellectual disability, treatment, synaptic proteome, synaptic transcriptome, splicing

Summary:

Synaptic Intellectual disability may derive from specific genetic alterations, as found in neurodevelopmental disorders such as fragile X syndrome (FXS) and Down syndrome (DS), both disorders associated to relevant alterations in synaptic plasticity. Mouse models of these disorders mimicking the genetic alterations found in humans have demonstrated relevant tools to understand the physiopathology of the disorders and to test pharmacological approaches that may improve cognitive performance. In the lab we have described an approach to improve cognitive performance in models of FXS and DS, but the impact of these treatments in the biology of the synapse has not been addressed. We are now investigating, using high throughput proteomic and transcriptomic analysis of sorted synaptic contacts, the characteristics of pathological synapses, and the effects that pharmacological treatments have in improving synaptic plasticity in both models of intellectual disability. Landmarks of intellectual disability

References:

Navarro-Romero A, Vázquez-Oliver A, Gomis-González M, Garzón-Montesinos C, Falcón-Moya R, Pastor A, Martín-García E, Pizarro N, Busquets-García A, Revest JM, Piazza PV, Bosch F, Dierssen M, de la Torre R, Rodríguez-Moreno A, Maldonado R, Ozaita A. Cannabinoid type-1 receptor blockade restores neurological phenotypes in two models for Down syndrome. *Neurobiol Dis.* 2019 May;125:92-106. doi: 10.1016/j.nbd.2019.01.014. Epub 2019 Jan 25. PMID: 30685352. Busquets-García A, Gomis-González M, Guegan T, Agustín-Pavón C, Pastor A, Mato S, Pérez-Samartín A, Matute C, de la Torre R, Dierssen M, Maldonado R, Ozaita A. Targeting the endocannabinoid system in the treatment of fragile X syndrome. *Nat Med.* 2013 May;19(5):603-7. doi: 10.1038/nm.3127. Epub 2013 Mar 31. PMID: 23542787.

Expected skills::

Bioinformatics

Possibility of funding::

To be discussed

Possible continuity with PhD: :

To be discussed
