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Group: Neurometabolic Diseases
Institution: IDIBELL, Institut d'Investigació Biomèdica de Bellvitge
http://www.neurometabolic-lab.org/home.jsp
http://www.idibell.cat/modul/neurometabolic-diseases/en

Project Title: Integration of novel NGS methods into the diagnosis of hereditary spastic paraplegias and leukodystrophies

The overarching goal of the Neurometabolic Diseases Laboratory is to improve the current medical practice, from diagnosis to treatment, of inherited diseases of brain white matter. During the last 3 years, we have implemented genetic diagnosis on families affected by leukodystrophies and spastic paraplegias, two overlapping groups of diseases showing a high clinical and genetic heterogeneity. Nowadays, we have access to clinical records, samples, and NGS data from more than 200 different families, in constant growth. However, the genetic background of these diseases is only partly understood, and more than 30% of patients remain without a genetic diagnosis.

Our group is searching for a PhD candidate to work in a project in which we intend to improve the identification of causative variants in known HSP/leukodystrophy genes, as well as finding novel candidate genes. In this project we will apply a systems biology approach to prioritize candidate variants issued from whole exome sequencing (WES) from a well-defined patient cohort, using our own annotation pipeline and in-house developed computing tools. In patients where exome analysis has not detected any candidate variant, we will apply whole genome and/or transcriptome sequencing. Candidate variants will be experimentally tested using molecular and cell biology approaches on patients' samples, cell lines edited with CRISPR-Cas9 methods, or animal models.

Highly motivated candidates with excellent academic records (undergraduate final scores **higher than 8.5**/10 in the Spanish evaluation system) are invited to submit their CV and motivation letter to:
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