Postdoc positions at the Neurometabolic Diseases Laboratory, IDIBELL, Barcelona, Spain
April 2022

Genomic Medicine for Rare Brain Diseases: from Genomic Diagnosis and Gene Discovery to Therapies through Brain Organoid modelling

Two postdoctoral /staff scientist positions available at the Neurometabolic Lab at IDIBELL (http://www.idibell.cat/en/content/neurometabolic-diseases) , Bellvitge Campus, Barcelona, Spain, led by ICREA Professor Aurora Pujol. We apply Genomic Medicine from diagnostics and gene discovery to therapeutics for rare brain metabolic diseases.

Position 1 is for a **molecular geneticist** with deep knowledge of genome analysis and interpretation, who aims at solving diagnostic Odysseys beyond conventional genomic diagnosis, using whole and long-read genomes, epigenomes, and transcriptomes. Ideal candidates will master bioinformatics tools for integrative multiomics analysis.

Position 2 is a for a **cell biologist** interested in organellar crosstalk, bioenergetics, immunology, aging and neuroscience, eager to work with brain organoids, in collaboration with Consiglio lab at Idibell. Multiomic approaches to decipher pathogenesis and the metabolic needs of the brain in development will be applied to find cures for rare metabolic diseases.

Enthusiastic and self-motivated team players with strong record of publications with first author publications and excellent English communication skills are encouraged to apply. Please submit a single PDF file with a cover letter, full CV and contact information of two references to Prof. Aurora Pujol (apujol@idibell.cat). Deadline is May 1st 2022, but interviews will start immediately. Follow us on Twitter: @PujolLab @consiglioLab

Related publications:

1. di Domenico A et al (2019). Patient-Specific iPSC-Derived Astrocytes Contribute to Non-Cell-Autonomous Neurodegeneration in Parkinson's Disease. *Stem Cell Reports*. 12(2):213-229.
2. Pant DC et al. (2019) Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. *J Clin Invest*, 129, 1240-1256.
3. García-Cazorla et al (2020) Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. *Acta Neuropathol*. 140(6):971-975.
4. Verdura et al (2021) Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. *Brain.* 2021 Oct 22;144(9):2659-2669
5. Schlüter A et al (2022) Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. *Neurology.* Mar 1;98(9):e912-e923.
6. Guasto A et al (2022) Biallelic variants in SLC35B2 cause a novel chondrodysplasia with hypomyelinating leukodystrophy. *Brain.* Mar 24:awac110.

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