PhD position in the Neurometabolic Diseases Laboratory, IDIBELL January 12 2021

"Gene Discovery for Rare Brain Diseases: from Genomic Diagnosis to Treatment using Brain Organoids"

A predoctoral position is available to enroll in the PhD student program of Biomedicine at the University of Barcelona, Spain. Work will be carried out in our genomic medicine research team at IDIBELL, Bellvitge Campus (*www.neurometabolic-lab.org*), (http://www.idibell.cat/en/content/neurometabolic-diseases) under the supervision of Dr. Stéphane Fourcade (PhD), Dr. Carlos Casasnovas (MD, PhD) and ICREA Professor Aurora Pujol (MD, PhD).

Our aim is to apply clinical and functional genomics for diagnostics and gene discovery. We are identifying novel disease-causing genes for brain neurodevelopmental disorders, such as myelin diseases or leukodystrophies, motorneuron disorders and cerebellar ataxias.

The proposed project, funded by FIS projects (PI20/00758 and PI20/007) and Marató de TV3 proposes to modeling novel rare metabolic brain diseases starting from patients fibroblasts', derived to iPS and brain organoid or specific motorneurons and astroglial cultures. Multiomic approaches to decipher pathogenesis and targeted drug testing and drug screening will follow. Results will decipher the metabolic needs of the brain in development while solving diagnostic Odysseys for many undiagnosed families.

Self-motivated team players, enthusiastic individuals who wish to acquire solid scientific knowledge *and* impact patients lives during their thesis are encouraged to apply. Previous experience with molecular/cellular neuroscience, genetics/genomics or bioinformatics is preferred. The ideal candidate would be eager to learn both dry and wet lab techniques. Publications are a plus. MSc and excellent English communication skills are required.

Applicants should submit a single PDF file with a cover letter, full CV, university marks and contact information of two references to Dr. Stéphane Fourcade and to Prof. Aurora Pujol (Tel.: +34 932607500 ext 3332, E-mail: sfourcade@idibell.cat, apujol@idibell.cat). The deadline for receiving applications is February 15th 2021, but interviews will start immediately. The selected candidate will apply for PFIS fellowship, although start date may be negotiable.

Selected publications

1. di Domenico A, Carola G, Calatayud C, Pons-Espinal M, Muñoz JP, Richaud-

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Patin Y, Fernandez-Carasa I, Gut M, Faella A, Parameswaran J, Soriano J, Ferrer I, Tolosa E, Zorzano A, Cuervo AM, Raya A,Consiglio A (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, Dorboz I, Schluter A, Fourcade S, Launay N, Joya J, Aguilera-Albesa S, Yoldi ME, Casasnovas C, Willis MJ, Ruiz M, Ville D, Lesca G, Siquier-Pernet K, Desguerre I, Yan H, Wang J, Burmeister M, Brady L, Tarnopolsky M, Cornet C, Rubbini D, Terriente J, James KN, Musaev D, Zaki MS, Patterson MC, Lanpher BC, Klee EW, Pinto E Vairo F, Wohler E, Sobreira NLM, Cohen JS, Maroofian R, Galehdari H, Mazaheri N, Shariati G, Colleaux L, Rodriguez D, Gleeson JG, Pujades C, Fatemi A, Boespflug-Tanguy O, Pujol A. (2019) Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. *J Clin Invest*, 129, 1240-1256. (https://www.ccma.cat/324/descobreixen-una-varietat-duna-malaltia-genetica-infantil-i-la-seva-cura-potencial/noticia/2897252/)

3. García-Cazorla A, Verdura E, Juliá-Palacios N, Anderson EN, Goicoechea L, Planas-Serra L, Tsogtbaatar E, Dsouza NR, Schlüter A, Urreizti R, Tarnowski JM, Gavrilova RH, SHMT2 Working Group, Ruiz M, Rodríguez-Palmero A, Fourcade S, Cogné B, Besnard T, Vincent M, Bézieau S, Folmes CD, Zimmermann MT, Klee EW, Pandey UB, Artuch R, Cousin MA & Pujol A (2020) Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. *Acta Neuropathol*. 140(6):971-975. (https://www.lavanguardia.com/vida/20201005/483870086261/descubren-una-nueva-enfermedad-genetica-rara-del-sistema-nervioso-y-cardiaco.html)

4. Vélez-Santamaría V, Verdura E, Macmurdo C, Planas-Serra L, Schlüter A, Casas J, Martínez JJ, Casasnovas C, Si Y, Thompson SS, Maroofian R, Pujol A (2020) Expanding the clinical and genetic spectrum of PCYT2-related disorders. *Brain* Sep 1;143(9):e76.

5. Verdura E, Fons C, Schlüter A, Ruiz M, Fourcade S, Casasnovas C, Castellano A, Pujol A (2020) *J Med Genet*. Feb;57(2):132-137.

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