Universitat Pompeu Fabra Barcelona | Master in Bioinformatics for Health Sciences

Master project 2024-2025

Personal Information

Supervisor	Marta Melé
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Institution	Barcelona Supercomputing center
Website	https://www.bsc.es/discover-bsc/organisation/scientific-structure/transcriptomics-and-functional-genomics-lab-tfgl
Group	Transcriptomics and Functional Genomics Lab

Project

Computational systems biology

Project Title:

Uncovering ribosomal RNA expression variation in health and disease

Keywords:

non-coding RNAs, GWAS, complex diseases, ribosomal RNA, gene expression, variant calling,

Summary:

The candidate will join Marta Melé's Transcriptomics and Functional Genomics lab in the Life Sciences Department at the Barcelona Supercomputing Center. The lab is interested in understanding how individual variation in gene expression and splicing profiles can explain phenotypic differences between individuals both in the context of health and disease. To address this question, we use large-scale transcriptomic analysis and the latest single-cell sequencing technologies combined with methods development to study gene expression, splicing and cell type composition variation across human tissues and phenotypes. The ribosomes are the cellular machinery responsible for protein synthesis. Ribosomes are composed of 60% ribosomal proteins and 40% ribosomal RNA (rRNA), which is primarily responsible for protein synthesis. Although ribosomal RNAs account for a vast majority (~90%) of the transcriptional output of a cell, little is known about rRNA expression variation. This is due to the highly complex and repetitive nature of rRNA genes. The release of the first complete human genome sequence offers the unprecedented opportunity to study one of the most complex regions of the human genome. In this study, we will characterize patterns of rRNA expression variation between tissues and individuals. We will develop a computational framework to identify rDNA variants from whole-genome sequencing data and identify novel genomic regions associated with complex diseases using the UK Biobank database. In addition, we will analyze whole-genome sequence and transcriptome data from the GTEx project (838 individuals and 17,382 transcriptomes) to validate our findings. What you will learn: Development of computational pipelines to analyse and interpret large datasets, especially from Whole Genome Sequencing and RNA-seq and single-cell RNA-seq. Working in a High Performance Computing environment. Scientific collaboration in the context of international consortia, effective communication of research findings in internal and external meetings, scientific writing, and critical thinking. Also, the master student will join the Melé lab journal clubs, lab meetings and lab lunches to talk about science but also have fun and discuss non-science related topics with the group.

References:

References 1. Garcia-Perez R, Ramirez JM, Ripoll-Cladellas A, Chazarra-Gil R, Oliveros W, Soldatkina O, Bosio M, Rognon PJ, Capella S, Calvo M, Reverter F, Guigó R, Aguet F, Ferreira PG, Ardlie KG, & Melé M§. The landscape of expression and alternative splicing variation across human traits. § corresponding author. Cell Genomics 0: 100244.

Expected skills:

Strong programming skills in bash, python, R, perl, or similar Some experience working in HPC clusters Some experience with Next Generation Sequencing data analysis Excellent communication skills in spoken and written English Capacity to contribute to research projects with novel research ideas and analysis Capacity to work as a team in a highly collaborative and diverse environment Availability to start in July 2024 is preferred

Possibility of funding:

Yes

Possible continuity with PhD:

To be discussed

Comments:

We are located at the 4th floor of the Barcelona Supercomputing center which has great views over Barcelona. We all go to work at least twice a week - most students go every day -but working from home is allowed a few days per week. For a list of selected publications from the lab see: https://www.bsc.es/mele-marta/publications All master students accepted in the lab will be offered a work contract as well as a practice agreement with the university.