



Master project 2024-2025

Personal Information

Supervisor Mario Cáceres
Email mcaceres@icrea.cat
Institution Hospital del Mar Research Institute
Website https://www.imim.es/programesrecerca/informaticabiomedica/en_genomica-comparada-funcional.html
Group Comparative and Functional Genomics

Project

Computational genomics

Project Title:

Functional and evolutionary impact of inversions and complex variants in the human genome

Keywords:

Structural variants, Human Genetics, Functional effects, Phenotypic traits, Evolution

Summary:

The master student will integrate in a highly-dynamic research group focused on the study of genome variation associated with individual and species differences. To address these questions, we use humans as a model and take a multidisciplinary approach that combines experimental and bioinformatic analysis of the great wealth of data available, generating results of interest to many diverse fields. Our main line of research is the global analysis of inversions and other complex regions, which aims to investigate the biological significance of the less known types of variants in the human genome. Thus, the proposed tasks span a diverse range of themes focused in the functional and evolutionary impact of these unique variants, whose effects have been missed in most current genomic studies, and the project could vary according to the interest and background of the candidate. 1. Bioinformatic characterization of complex genomic regions with Oxford Nanopore ultra long reads and analysis of their functional consequences and association with phenotypic traits and disease susceptibility. 2. Evolutionary analysis of the role of natural selection on human inversions and their effect on nucleotide variation patterns. 3. Development of new functionalities and visualization tools for our human polymorphic inversion data base InvFEST (<http://invfestdb.uab.cat/>), as a model for new reference catalogues of structural variants.

References:

F. Degenhardt et al. Detailed stratified GWAS analysis for severe COVID-19 in four European populations. *Human Molecular Genetics*, 31: 3945-3966 (2022). E. Campoy et al. Genomic architecture and functional effects of human inversion supergenes. *Philosophical Transactions of the Royal Society B* 377: 20210209 (2022). M. Puig et al. Determining the impact of uncharacterized inversions in the human genome by droplet digital PCR. *Genome Research* 30: 724-735 (2020). C. Giner-Delgado et al. Evolutionary and functional impact of common polymorphic inversions in the human genome. *Nature Communications* 10: 4222 (2019). A. Martínez-Fundichely et al. InvFEST, a database integrating information of polymorphic inversions in the human genome. *Nucleic Acids Research* 42 (D1): D1027-D1032 (2014).

Expected skills:

Expected skills depend on the actual line of research chosen, but ideally should include scripting/programming skills (bash, R, etc.) and experience in genomic variants and functional or evolutionary analysis. Knowledge of MySQL and PHP would also be helpful for working with the InvFEST database.

Possibility of funding:

Yes

Possible continuity with PhD:

To be discussed

Comments:

Depending on the degree of experience of the candidate and the task performed it is possible to obtain financial support for the master practice. Also, at the end of the master there is the possibility to apply for a PhD fellowship.