upf. Universitat Pompeu Fabra Barcelona

Master in Bioinformatics for Health Sciences

Master project 2021-2022

Personal Information

Supervisor	Mario Cáceres
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Group	Comparative and Functional Genomics Group

Project

Computational genomics

Project Title:

Functional and evolutionary impact of polymorphic inversions in the human genome

Keywords:

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

Summary:

The master student will integrate in a young, interdisciplinary and highly-dynamic group and the project could vary according to the interest and background of the candidate. In particular, the proposed tasks span a diverse range of themes focused in the functional and evolutionary impact of inversions, which are a little studied class of genomic variants and their effects have been missed in most current genomic studies. 1. Identification and genotyping of inversions from bioinformatic analysis of long-read sequences to study their association with phenotypic traits and disease susceptibility (in collaboration with the CNAG) 2. Comparative study of known human inversion regions in other mammal species genomes to determine if there are inversion recurrence hotspots conserved over long evolutionary distances that might indicate a potential functional role. 3. Development of new functionalities and visualization tools for our human polymorphic inversion data base InvFEST (http://invfestdb.uab.cat/), the world reference of human inversions.

References:

F. A. M. Maggiolini et al. Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. Genome Research 30: 1680-1693 (2020). 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). D. Vicente-Salvador et al. Detailed analysis of inversions predicted between two human genomes: errors, real polymorphisms, and their origin and population distribution. Human Molecular Genetics 26:567-581 (2017). 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 should include scripting/programming skills (python, bash, R and/or perl) and experience in genomic variants and functional analysis. Knowledge of MySQL and PHP would also be helpful for working with the InvFEST database.

Possibility of funding::

To be discussed

Possible continuity with PhD: :

Yes

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.