Universitat Pompeu Fabra Barcelona | Master in Bioinformatics for Health Sciences

Master project 2020-2021

Personal Information

Supervisor	Mario Cáceres
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Website	https://invfest.uab.cat/
Group	Comparative and Functional Genomics Group

Project

Computational genomics

Project Title:

Functional and evolutionary impact of polymorphic inversions in the human genome

Keywords:

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.se.

Summary:

The master student will integrate in a young, interdisciplinary and highly-dynamic group. 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 the project could vary according to the interest and background of the candidate. 1. Bioinformatic analysis of the functional consequences of inversions and their association with phenotypic traits and disease susceptibility through imputation of inversion genotypes in large-scale datasets, in which the effect of these changes has been typically missed. 2. 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. 3. 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.

References:

M. Puig et al. Determining the impact of uncharacterized inversions in the human genome by droplet digital PCR. Genome Research (in press) (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). M. Puig et al. Functional impact and evolution of a novel human polymorphic inversion that disrupts a gene and creates a fusion transcript. PLoS Genetics 11(10): e1005495. doi:10.1371/journal.pgen.1005495 (2015). 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 perl, python and bash programming and experience in working with DNA sequence data and functional analysis. Knowledge of MySQL and PHP would also be helpful for working with the InvFEST database.

Possibility of funding::

Yes

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.