Universitat Pompeu Fabra Barcelona

Master in Bioinformatics for Health Sciences

Master project 2021-2022

Personal Information

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Group	Human Genome Diversity

Project

Computational genomics

Project Title:

Analysis of the human genome diversity: unravelling demographic and genomic processes

Keywords:

Genome diversity, human populations, demography, adaptation

Summary:

The interests of our research are focused on the human genome diversity analysis in order to infer the (genomic and population) processes responsible for this diversity and try to establish the (population and epidemiological) consequences of the human genetic variability. Thus, our main research lines are focused on aspects of human genome diversity, population genetics, genome variation and disease susceptibility, and genome evolution and disease. 1. Population processes: Concerning population processes that have modeled the human genetic diversity, we have focused our research on the use of molecular tools to reconstruct the human population history through the phylogeny of genetic markers. Our interest has been focused on the genetic consequences at population level of human migrations and admixtures. The use of well-established phylogenies in the mitochondrial and Y-chromosome human genomes allowed us to unravel the population history of several populations. Nonetheless, we have recently used whole genome variation in the autosomes in order to establish the structure of human populations. 2. Genomic processes: Concerning genomic processes that have modeled the human genetic diversity, our research has have been focused on the relationship between human diversity and complex traits, including complex diseases. The genetic analysis in human populations of genes of biomedical interest might shed light on the evolution of these genes. In this context, we have focused our research in the analysis of genes that have been previously associated to complex diseases, such as psychiatric and immunological diseases. The analysis of these genes has allowed us to conclude that some of the failures in replicating genetic associations are due to extreme genetic differences between populations. In addition, we are also interested in other complex traits, such as height, not directly related to disease.

References:

1. Lorente-Galdos B, Lao O, Serra-Vidal G, Santpere G, Kuderna LFK, Arauna LR, Fadhlaoui-Zid K, Pimenoff VN, Soodyall H, Zalloua P, Marques-Bonet T, Comas D (2019) Whole-genome sequence analysis of a Pan African set of samples reveals archaic gene flow from an extinct basal population of modern humans into sub-Saharan populations. Genome Biology 20:77. 2. Font-Porterias, Arauna LR, Poveda A, Bianco E, Rebato E, Prata MJ, Calafell F, Comas D (2019) European Roma groups show complex West Eurasian admixture footprints and a common South Asian genetic origin. PLoS Genetics 15(9): e1008417. 3. Serra-Vidal G, Lucas-Sanchez M, Fadhlaoui-Zid K, Bekada A, Zalloua P, Comas D (2019) Heterogeneity in Palaeolithic population continuity and Neolithic expansion in North Africa. Current Biology 29:3953-3959. 4. Castro e Silva MA, Nunes K, Lemes RB, Mas-Sandoval A, Amorim CEG, Krieger JE, Mill JG, Salzano MS, Bortolini MC, da Costa Pereira A, Comas D, Hünemeier T (2020) Genomic insight into the origins and dispersal of the Brazilian coastal natives. Proceedings of the National Academy of Sciences USA 117 (5) 2372-2377. 5. Bianco E, Laval G, Font-Porterias N, García-Fernández C, Dobon B, Sabido-Vera R, Sukarova Stefanovska E, Kučinskas V, Makukh H, Pamjav H, Quintana-Murci L, Netea MG, Bertranpetit J, Calafell F, Comas D (2020) Recent Common Origin, Reduced Population Size, and Marked Admixture Have Shaped European Roma Genomes. Molecular Biology and Evolution 37(11):3175-3187.

Expected skills::

Computational skills to manage and analyze genotype and DNA sequence data from whole genomes

Possibility of funding::

To be discussed

Possible continuity with PhD: :

To be discussed