
**Entity:** Vall d'Hebron Institut de Recerca (VHIR)

**Unit or Department:** Translational Bioinformatics Unit, Neurosciences

**Student's tutor:** Xavier de la Cruz

**Positions available:** 1

## Project description/ Research lines.

### Identification and prediction of pathological mutations in the clinical setting: applications to exome and gene panel sequencing in diagnosis

The student joining our group will integrate in an ongoing project aimed at the identification of pathological mutations with diagnostic purposes. Our goal in this area is to develop an original methodology for the identification of pathological mutations and to bring it all the way down to bedside applications. To this end, we have divided our work as follows: (i) tool design and development, (ii) validation in specific diseases, and (iii) clinical application. The first part is entirely addressed within our group and incorporates a significant methodological novelty, its gene-specific nature, able to overcome the performance limits of general prediction methods. It is the continuation of a research line in which we have a substantial experience, and which plays a growing role in the use of exome data for diagnosis and causative gene identification. For the second part, we have sought groups from the VHIR community (Immunology; Neurovascular disease; and Mitochondrial and Neuromuscular disease) and from outside (Translational Research; Oncogenetics; both from VHIO; Cancer and Iron group, from IMPPC) to test our results in specific diseases. In the third part, we have contributed to the actual diagnosis of several, hard cases provided by the Vall d'Hebron Hospital.

This project constitutes the core of our research and has grown in accordance, particularly since we joined the Vall d'Hebron Research Institute (VHIR). Apart from developing our methodology, we have established a network of collaborations with our environment and beyond, with the goal of solving specific problems and testing our tools. To the collaborations mentioned above, I would like to add the work we are doing with the group of Dr. Cedric Notredame, for testing the effect of improved multiple sequence alignments on variant annotations. Within a more applied context, it is of note our work with Prof. Joan Seoane in the conception of novel technology to address the problem of identifying variants from exome experiments in cancer samples. Finally, I will add that we have been trusted by Illumina, Co., the main company in genome/exome sequencing products, to use our expertise for the benchmarking of their TruSight Tumor Sequencing Panel in Spain.

**Period for the internship:** *preferably first or second semesters*

**Requirements:**

- Genetics and molecular biology knowledges
- Wellcomed but not required: familiarity with the computer environment in linux, and/or programming language (e.g. Perl or Python)
- Very important: dynamic attitude towards learning novel technologies

**Where to apply:** Interested candidates please send *CV and academic records* to Prof. Xavier de la Cruz (xavier.delacruz@vhir.org)