



Master project 2024-2025

Personal Information

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Project

Computational genomics

Project Title:

Implementation of a highly-accurate CNV detection strategy

Keywords:

NGS, CNV, genomics, python

Summary:

The project focuses on improving the accuracy of Copy Number Variation (CNV) detection from targeted Next-Generation Sequencing (NGS) data. To enhance accuracy, the project devises several strategies: - Implementing new methods: Novel approaches will be implemented to improve CNV detection sensitivity and specificity. - Fine-tuning existing tools: Currently used CNV detection tools in qGenomics will be optimized to better capture variations in the data. - Performing consensus calling: Multiple CNV calling algorithms might be employed, and a consensus approach will be utilized to increase the reliability of the identified CNVs. - Establishing a comprehensive reference for benchmarking, which will potentially involve combining in-silico variant datasets with publicly available and curated datasets. - Incorporating more layers of annotations to aid in variant interpretation. For example, crossing CNVs with regions of homozygosity. Depending on the progress and outcomes of the project, similar efforts may be extended to include structural variant calling from Whole Genome Sequencing (WGS) NGS data.

Expected skills:

self-management, problem solving, communication, team work

Possibility of funding:

To be discussed

Possible continuity with PhD:

To be discussed

Comments:

The lab is located at Esplugues de Llobregat and there's possibility of hybrid work model.