

## DESCRIPTION

Based on the campus of BC Children's Hospital (BCCH), the Mostafavi and van Karnebeek laboratories partner with a strong genetics community to apply multiple types of omics technologies to unravel the (epi-) genetic pathophysiology of rare neurodegenerative diseases. We are looking for a postdoctoral scientist to lead the research on developing and applying computational and statistical approaches to combine multiple types of genomics data (including whole genome sequencing, metabolomics, and DNA methylation data), with the goal of identifying disease mechanisms and biomarkers, along with treatment targets, to improve patient outcomes. This translational work requires close interaction with BC Children's Hospital clinicians to understand the phenotypes of patients. A background in molecular biology and familiarity with bioinformatics tools and techniques would be a strong asset for the applicant.

This project builds upon the success of the TIDEx project at BCCH, which through implementation of state-of-the-art sequencing and computational approaches in a clinical setting has pioneered the discovery of multiple confirmed rare-human-disease genes and pathways.

The ideal candidate will also pursue research on statistical data integration of multi-omics data in the context of common neurological disease. In this direction, the candidate will work with members of the Mostafavi lab and collaborators at Brigham's Women's Hospital in Boston to develop and apply methods for combining genotyping data, RNA-sequencing gene expression data, DNA methylation data in the context of Multiple Sclerosis and Alzheimer's Disease.

## MAJOR RESPONSIBILITIES

- Conduct research and supervise projects in the area of Applied Genome Analysis.
- Develop and apply methods for the identification of disease causing variations in genome sequences.
- Develop and apply computational methods for statistical data integration of multiple types of genomics data.
- Present findings at local, national and international scientific meetings where the impression can have direct influence on future scientific funding.
- Assist in the development of grant and scholarship applications related to the research.
- Work with principal investigator to ensure that all scientific milestones are achieved.

## QUALIFICATIONS

Education and Experience:

- Ph.D. degree in a life sciences discipline, Bioinformatics, or Computational Biology.
- A minimum of 2 years demonstrated experience years' in bioinformatics with focus on analysis of genomics data.

Skills and Abilities:

- Familiarity with metabolomics data and analysis tools
- Knowledge of bioinformatics, with emphasis on genome sequence analysis and gene annotation
- Knowledge of scientific tools, technologies and online resources for gene disease relationship prediction
- A scripting programming language like Python or Perl.
- A programming language for data manipulation and computational modeling, like R or MATLAB
- Experience with the Linux environment
- Experience with computer clusters
- SQL database use via API interfaces
- Experience with gene network analysis tools
- Experience with project management tools, like Git and GitHub.

## **HOW TO APPLY**

Please email your cover letter and resume to [sara@cmmt.ubc.ca](mailto:sara@cmmt.ubc.ca). Due to the number of resumes we receive, we are unable to confirm receipt of submissions over the phone, or provide the status of competitions except to those who are selected for an interview.