Interpretation automation of genetic variations from rare diseases

Record number : OPR-1298

Overview

RESEARCH DIRECTION

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INFORMATION

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ADMINISTRATIVE UNIT(S)

Faculté de médecine et des sciences de la santé

LEVEL(S)

Stage postdoctoral

LOCATION(S)

Campus de la santé

Project Description

We are searching for a postdoctoral student with an expertise in bioinformatics to join our team in a project framework in clinical genomics financed by the Ministère de la Santé et des Services Sociaux. This project, titled «Optimization of genomic data interpretation and clinical report of Quebec's health network through supervised automation», aims to enhance Quebec's clinical genomic bioinformatic platform (CLIN). The second part of the project focused on interpretation automation of genetic variations from rare diseases, particularly through integration and enhancement of our bioinformatic tool PhenoVar for diagnostic predictions. This offer could lead to a bioinformatician job within our research team.

Main responsibilities:
-Tool development and integration :
  -Contribute to the integration of the PhenoVar tool in the CLIN platform.
  -Develop API and other technical solutions to facilitate automation process.
  -Adapt and enhance PhenoVar based on new recommendations from the American College of Medical Genetics and Genomics (ACMG) and integrate research tools from literature.
-Tool validation from existing genomic data:
  -Validate the functionality and performance of PhenoVar once integrated into the CLIN platform, in comparison with other tools from the CLIN platform through standardized genomic data with known diagnostics (benchmark).

-Continuous improvement:
  - Work on improvement of tools and analysis pipelines based on project needs and user comments.
  -Integrate new functionalities and enhance tools prediction capabilities.

-Documentation and communication:
  - Document process, analysis and results in a concise way.
  - Regularly communicate project progress and results to interested parties.

-Collaborate with multidisciplinary teams from CIUSSE, and partners from Ste-Justine and Quebec University Hospital Centers.

Required qualifications:

-Education: Doctorate in bioinformatics, genomics, computational biology or a related field.

-Technical skills:
-Experience with bioinformatics tools and softwares, particularly those used in analyzing genetic variations.
-Programming skills: Python, Jay.
-Software development skills: Django, Javascript, JQuery, CSS, MySQL, Git.

-Interpersonal skills:
-Ability to work as a team and to collaborate with multidisciplinary researchers.
-Excellent oral and written communication skills.
-Organization and multitasking skills.

-Additional assets:
-Knowledge of public genomic databases and ACMG classification principles.
-Knowledge in machine learning and artificial intelligence.

Discipline(s) by sector

Sciences de la santé

Génétique

Funding offered

Yes

Ministry of Health.

Partner(s)

Collaboration avec Vincent Ferretti (UdeM-CHU Ste-Justine)