## **Bioinformatician**

Pacific Northwest Research Institute (PNRI) is seeking a Bioinformatician for the Carvalho lab who will focus on investigating the molecular basis of structural and copy number variations in rare human genetic diseases. This position requires the ability to analyze complex research problems and will involve the development and implementation of a variety of computational methods and tools using short and long-read next-generation sequencing to study the human genome. We are seeking a candidate who can work as part of a multidisciplinary team of scientists and researchers.

The candidate will be part of collaborative projects aimed at genotyping and molecular characterization of the genome in patients and family members with Mendelian genetic diseases. Projects aim to identify and characterize structural variants, single-nucleotide variants and indels using data from various experimental platforms (e.g., Illumina, PacBio, Nanopore, BioNano, array CGH). The candidate will apply a wide range of existing NGS analysis software or assist in developing new analytical tools as needed to discover novel DNA variants affecting genes that can cause disease. Job responsibilities also include data transfer and database maintenance, analysis pipeline development and deployment, and working knowledge of population databases for use in characterization of variant pathogenicity. This position will have daily interactions with lab members and PI as well as outside collaborators to work collaboratively in project development and progress. Skill in manuscript writing is an essential qualification.

The successful candidate will be able to deploy, integrate, and maintain software and databases, and provide computational support of scientific projects related to research goals of the Principal Investigator.

## **Qualifications:**

- Bachelor's or master's degree in bioinformatics, computer science, genetics, or related field
  plus two or more years of experience working with NGS data and relevant tools for
  bioinformatics analysis, in either a professional or academic setting.
- Working understanding of human genetics.
- Competency with scripting languages (Python, R, bash) and the Linux programming environment.

- Experience working with command line-based software for NGS processing, analytical pipelines, and standard data formats (e.g., BWA, GATK, Samtools, SAM/BAM, VCF).
- Ability to work, collaborate and communicate well within a multidisciplinary team.
- · Must be self-motivated and able to take initiative, work independently, and learn rapidly.
- Strong organizational abilities.
- Strong written and oral communication skills.

## **About PNRI:**

PNRI is an independent, nonprofit, biomedical research institute with a distinguished history of contributing scientific advances to improve health in a variety of disease areas. PNRI is committed to building a culture that encourages originality, risk-taking, and interdisciplinary collaboration. Please see www.pnri.org for more information.

## **How to Apply:**

For immediate consideration, please submit a cover letter and Curriculum vitae at: <a href="mailto:hr@pnri.org">hr@pnri.org</a>. NO PHONE CALLS PLEASE.