PULSE

Postdoc Upskilling in Laboratory & Specialized Expertise

*A Professional Development Initiative for Postdoctoral Scholars at UNC Core Facilities*

## Title of Proposed Project

Development of the GENYSIS long-read sequence methylation analysis pipeline

## Name of Core Facility

GENYSIS Clinical Genomic Analysis Core

## Core Director Name *Official contact for the program*

Tam P. Sneddon

## Project Description

GENYSIS uses multiple analysis pipelines to identify germline sequence and structural variants in the research genomes of study participants with rare genetic disease. At this time, methylation analysis of long read sequencing is a component not properly evaluated or included within our workflow. Methylation is a process where additions to the DNA can affect gene regulation and have been shown to be involved in several human diseases. It is hoped that by adding this component into our automated pipeline we will discover the underlying cause of the disease of some of our study participants.

The postdoc would evaluate available programs that analyze methylation from long-read sequence data generated and include components of these into our existing analysis pipeline. The postdoc will present to the group with their findings and then will build that into our existing custom pipeline.

## Expected Skill Development

The postdoc will gain hands on experience with methylation data from Oxford Nanopore Technology long read sequencing projects and develop skills of building pipelines to enable large scale automation of analysis. It is hoped that the postdoc might also be able to use this information to discover the case of a study participant's underlying genetic condition.

## Scheduling Considerations

Project can be performed remotely with internet access. The postdoctoral scholar will have a weekly meeting with the mentor and ideally be able to attend our weekly GENYSIS tech meeting held on Monday afternoons at 3pm. Other optional meetings may be attended.

## Estimated Time Commitment

8 hours/week
Note: Time commitment is negotiable and can be split over the week or consolidated into one day a week. There are desks available in the ClinGen/GENYSIS suite on the 4th floor of the Medical Biomolecular Research Building for the postdoc to work, although most meetings can be attended via Zoom.

## Expected Project Duration

12 weeks
Note: Expected project duration is negotiable, but it is thought that 12 weeks would provide the maximum benefit

## Designated Mentor(s)

Scott Melville, Research Associate – Bioinformatician, will be the main contact point for this role (95%)

Additional mentors may include

- Tam Sneddon, Director
- Bradford Powell, Faculty Director

## How often will the mentor meet with the participating postdoctoral scholar?

Weekly check-ins
As needed / flexible
Note: Regularly scheduled one-to-one weekly meetings. Additional meetings can be held when required to promptly solve issues as they arise.

## Required Background or Skills

We are looking for a postdoc who has a medium to high understanding of bioinformatics and some understanding of genetics. No knowledge of methylation processes is required. Familiarity with UNC's computing clusters Longleaf or Sycamore and experience with BASH/Linux commands is a must. Part of this role will involve building pipelines to process multiple samples so knowledge of tools such as snakemake or nextflow would be handy but not required.

## Onboarding Plan

Onboarding during the first week will include required online HIPAA and CITI Human Subjects Research training, if not already certified (2-3 hours), and this would be in addition to the overall project timeline. Mentor and mentee will meet in person or remotely to go over necessary information.

## Training Plan

Given the applicant will be familiar with a computing cluster, it is not anticipated there will be much technical training required. Any additional genetic training that is required will be delivered in person, via remote meetings, or directions to self-learning online modules. Training for pipeline tools (eg snakemake, nextflow) will occur through online training and with the mentor.

## Potential Deliverables/Outcomes

☒ Method development
☒ Poster presentation
☒ Contribution to manuscript
☒ Skill Development