Genomics Education Programme

NHS Health Education England

Jamie Ellingford

Jamie has had an interesting academic journey to date, he has completed a BSc in Biology at the University of Sheffield, an MRes in Translational Medicine ('bench-to-bedside') at the University of Manchester, a PhD in Genomics at the University of Manchester and he has also worked as a Research Bioinformatician at the Central Manchester NHS Foundation Trust.

Jamie's PhD thesis was based on research focused on computational strategies to identify novel disease-causing variation underpinning rare diseases through WGS, WES and custom gene panel approaches. Although this project was largely computational, he also spent a considerable amount of time in the wet laboratory. Going forward, Jamie has plans to develop his skills in both of these areas; although a major focus will be computational.

The 'big' questions that drive Jamie's research interests are: (1) evolutionary genomics, and (2) applied 'omics in healthcare. Jamie'a research fellowship will bring both these interests together and will be based at the Manchester Centre for Genomic Medicine (MCGM) in the Central Manchester Teaching Hospitals.

Aside from genomics, Jamie's interests are sports, music and travel – so for him Manchester is an ideal place to live. A highlight from his PhD studies was a 3-month trip to India to work in a Bioinformatics institute.

Outline of Research (Full Time, Postdoctoral)

EIGER: a bioinformatics approach to Evaluate and Integrate Genomic variation impacting gene expression and Regulation.

A major objective of this research is to identify pathogenic variants in the non-coding genome which underpin rare diseases. Initially, the project will focus on disorders within the hearing & sight and the cardiovascular GeCIP domains, with the expectation that establishing an analysis framework for these disorders will enable integration and expansion to other clinical specialties. The project involves both bioinformatics and laboratory-based analyses, including the interrogation of publically-available resources from the ENCODE project, the use of phenotype-driven variant analysis tools and the creation of patient-specific RNA-seq and chromatin conformation datasets. As a result of this fellowship, I aim to release computational tools and strategies that will assist with the identification of pathogenic variants in the non-coding genome.