

## Alignment and Variant Calling

### Using Galaxy and the Command Line

**Date: 9 am – 5 pm, Wednesday 22<sup>nd</sup> April 2024**

**Location:** Computer Room 3, Polwarth Building, University of Aberdeen, Foresterhill Campus.

**Format:** Instructor led computer based tutorial with support demonstrators.

**Instructors:** CGEBM Bioinformaticians (Dr Antonio Ribeiro, Dr Louie van de Lagemaat, and Dr Dami Oresegun)

**Description:** The availability of sequenced reference genomes for a large number of species has led to an increase in research that investigates variation in isolates and individuals that could be linked to phenotypic changes. This workshop will work through the key steps involved in the process of taking sequencing data and analysing it to give a list of variants and the genes associated with these changes. We will utilise both Galaxy and the command line to achieve this.

#### **Topics:**

- Aligning Reads to a Reference Genome
- Visualising Alignments
- Variant Calling
- Filtering of Variants

**Who should attend:** This workshop is aimed at researchers, staff and PhD students, interested in resequencing of individuals to identify variants. The aim of the workshop is to demonstrate the keys steps involved in this analysis. Quality control and filtering of raw reads will not be covered. These steps are taught in our Quality Control workshop. Examples of biological interpretations will be shown but detailed steps of these processes will not be covered.

**Pre-requisites:** Introduction to the University of Aberdeen HPC Cluster and Basic Unix Skills workshop or previous experience with the command line and HPC (Maxwell or Macleod).

An account on the teaching HPC, Macleod, is needed. We are providing teaching accounts for all participants to use for the duration of the workshop. If you would like to use the research HPC, Maxwell, after the workshop you will need to obtain an account (Email: [digitalresearch@abdn.ac.uk](mailto:digitalresearch@abdn.ac.uk)). For more details see <http://www.abdn.ac.uk/staffnet/working-here/hpc.php>. Macleod and Maxwell have the same file structure, operating system and scheduler. What you learn on the course is applicable to both University of Aberdeen HPC clusters.

**Closing Date for registrations:** 15 April 2024 or when capacity is reached.

**Closing Date for cancellations:** Cancellations received before 15/04/2024 will receive a full refund minus a £40 admin fee. Full course fees will be charged for cancellations after this date.

**Cost:** For University of Aberdeen staff and PhD students the workshop is £180. Payments must be made before registration. Please enter your order details on the registration form. Details on how to raise an internal order for the course fees are provided on the registration form. Separate course fees apply for external or commercial delegates. Please contact us for further details before registering.

**Registrations:** Please complete the registration form [here](#), or scan the QR code



**Further Information:** Please contact Elaina Collie-Duguid ([e.collie-duguid@abdn.ac.uk](mailto:e.collie-duguid@abdn.ac.uk))

**NOTE** This workshop will only run if a minimum number of attendees register. In circumstances of cancellation, full refunds will be given.