# CENTRE FOR GENOME ENABLED BIOLOGY AND MEDICINE

University of Aberdeen



# **Alignment and Variant Calling**

Using Galaxy and the Command Line

## Date: not running 2024/5

**NOTE** To express interest, please contact Louie van de Lagemaat (louie.vandelagemaat@abdn.ac.uk).

**Location:** <u>Normally</u> 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)

**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 in the process, including 1) <u>aligning next-generation sequencing reads</u> to a reference genome, 2) <u>detecting a list of variants</u> and 3) identifying the genes associated with these changes. We will utilise both Galaxy and the command line to achieve this. Please note, this course <u>does not cover</u> analysis of multiple sequence alignments using Clustal, JalView, or related programs.

#### **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 provide 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: <u>digitalresearch@abdn.ac.uk</u>). For more details see <u>http://www.abdn.ac.uk/staffnet/working-here/hpc.php</u>. 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: N/A

## Closing Date for cancellations: N/A

**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.

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**Registrations:** N/A

Further Information: Please contact Elaina Collie-Duguid (<u>e.collie-duguid@abdn.ac.uk</u>)