GMS Bioinformatics for Genomic Medicine course

22rd – 26th October 2018

Organised and delivered by the WHG Bioinformatics Core group

Location: Room B, Wellcome Centre for Human Genetics

Tutors: Silvia Salatino, Raquel dos Santos Silva

Course Overview

This course will give you an overview of variant analysis in NGS data (whole-genome, exome, targeted gene panels). Topics will include include large genomic projects (e.g. 1000 Genomes, UK10K, etc.), mutation types and their functional effects, variant calling, filtering and visualisation. The course will also include a practical session and a presentation session by the attendants (on a peer-reviewed article that will be distributed during the course).

Course Material

Course material can be found at the following link:
http://www.well.ox.ac.uk/bioinformatics/training/BGM_2018/

Course Preparation

Attendees will need to bring their own laptop for the practical session, either MacOS, Linux, or Windows (7 or newer). Android OS is not compatible with the software that will be used during the course. Please ensure you have an Eduroam wifi account; if not, a temporary OWL account can be provided. Programming knowledge is not a requirement for this course.

Student presentations

Attendees will be asked to prepare a short presentation (~10-15 min each) about one of the following topics:

1) Variant detection from RNA-seq data: advantages, challenges, algorithms

2) Variant detection from ancient DNA: challenges and possible solutions

3) Variant detection from long-read technologies (e.g. PacBio or ONT): differences from short-read technologies and current strategies

4) Somatic variant calling in cancer: challenges and current approaches

5) Comparison of state-of-the-art variant calling software

6) Variant calling from single-cell data and its application in cancer

7) Identification of structural variants (e.g. CNVs, translocations, etc.)

8) A large genomic project of your choice (e.g. 1000 Genomes, UK Biobank, etc.): study design, patient consent, data analysis, impact, etc.

9) Risk of patient identification from sequencing data (despite anonimization!)

10) Assessing the accuracy of variant calling software: benchmark datasets construction and quality metrics

11) Case-control studies: the transition from GWAS to high-throughput sequencing

12) Exomiser and the phenogenomics approach to variant prioritisation

Schedule

Monday 22nd Oct

10:00 – 12:00	Lecture: Variant analysis of NGS data, part I
13:00 – 16:00	Students work on set task: look at papers given in AM session

Tuesday 23rd Oct

10:00 – 12:00	Lecture: Variant analysis of NGS data, part II
13:00 – 16:00	Students work on set task: prepare presentations

Wednesday 24th Oct

13:00 – 16:00	Practical session: Exome sequencing case study

Thursday 25th Oct

(break)

Friday 26th Oct

10:00 – 12:00	Student presentations, 15 mins each (including 5 mins for questions)
12:00 – 13:00	Lunch (provided)
13:00 – 15:00	Student presentations, 15 mins each (including 5 mins for questions)