SPGH Sociedade Portuguesa de Genética Humana

Practical course on computational tools and bioinformatics for next-generation sequencing

18th – 19th May 2018

Venue: ***Departamento de Engenharia Informática, Instituto Superior de Engenharia do Porto***
Rua Dr. António Bernardino de Almeida, 431, 4200-072 Porto

Programme

18th May

14:00h Welcome and introduction to the course
Rosário Santos & Jorge Oliveira

14:30h-18:30h SESSION I _ From raw data to workable files
Domantas Motiejunas. Bioinformatics, ThermoFisher Scientific, Lithuania (domantas.motiejunas@thermofisher.com)

- Primary NGS bioinformatics - from the raw data signal to DNA sequences
- NGS metrics and quality control (QC) of sequencing runs
- Format and structure of standard NGS files - BAM, BED, and VCF
- Visual inspection of variants in BAM files
- Filter variants according to parameters and annotations.

19th May

09:00h-13:00h SESSION II _ Filtering candidate variants and genes
Jean-Pierre Desvignes & David Salgado. IGE Bioinformatique, Aix-Marseille Université, France (jean-pierre.desvignes@univ-amu.fr; david.salgado@univ-amu.fr)

- Introduction to VarAFT tool
- Filtering candidate variants and applying distinct inheritance disease models
- Practical examples of gene panels and exome cases
- Evaluating coverage analysis of BAM files.

14:00h-18:00h SESSION III _ ENSEMBL Browser: NGS data annotation & data mining
Ben Moore. ENSEMBL, European Bioinformatics Institute (EMBL-EBI), United Kingdom (bmoore@ebi.ac.uk)

- Introduction to ENSEMBL
- ENSEMBL VEP to annotate genetic variants
- Data export with *BioMart* - retrieving genomic information using a web interface
- Variation - SNPs and other polymorphisms, haplotypes, linkage disequilibrium, structural variants such as CNVs.