IX International Course of Massive Data Analysis FOR GENOMICS

Course Presentation

EMBL-EBI

ciberer
CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED
DE ENFERMEDADES RARAS

PRINCIPE FELIPE
CENTRO DE INVESTIGACION

INB

Ignacio Medina
imedina@ebi.ac.uk

Presentation

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Introduction
Who we are

- Teachers:
  - David Montaner: Head of the Biostatistics Unit
  - Marta Bleda: Bioinformatician
  - Ignacio Medina: Project Manager at EBI Variation
- From Joaquin Dopazo group at CIPF:
  - http://bioinfo.cipf.es/
- More than 8 years of experience in microarrays and NGS data analysis and Bioinformatic tool development, and developing methodologies for data analysis
- Many suites and tools developed: GEPAS, Babelomics, Genome Maps, VARIANT, ...
- More than 50 papers in the last 8 years in peer reviewed journals: NAR, Bioinformatics, Nat. Biotech., …
- Many collaborations with experimental and clinic groups
- Many international courses run last years: Massive Data Analysis (MDA)

Introduction
Goals, ambitious

- To be able to conduct a whole NGS data analysis from scratch in a Linux environment
- To know and understand the different analysis pipelines and data formats (fastq, sam/bam, vcf)
- To preprocess and perform QC of data
- To learn how to install and use the ecosystem of tools to perform NGS data analysis
- To tune up data analysis pipelines by simulating data
- To perform some basics functional interpretation of variant and RNA-seq analysis
- To present some cloud based solutions being developed

Ignacio Medina
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Program
First day

- 09:30 Presentation
- 10:00 Introduction to NGS Technologies for Genomic Studies
- 10:30 Introduction to GNU/Linux shell
- 11:00 Coffee Break
- 11:30 Quality Control for NGS Raw Data (FASTQ) and Data Preprocessing
- 12:30 Lunch Break
- 14:00 Mapping NGS Reads for Exome and Transcriptomics Studies I
- 16:00 Tea Break
- 16:15 Mapping NGS Reads for Exome and Transcriptomics Studies II
- 17:30 Finish

Program
Second day

- 09:30 Visualization of NGS data (BAM)
- 11:00 Coffee Break
- 11:30 Variant Calling (SNPs & INDELs) and Variant Visualization (VCF) I
- 12:30 Lunch Break
- 14:00 Variant Calling (SNPs & INDELs) and Variant Visualization (VCF) II
- 15:15 Variant Annotation
- 16:00 Tea Break
- 16:30 Association studies with VARIANT
- 17:30 Finish

Ignacio Medina
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Program
Third day

- 09:30 RNA-seq data preprocessing
- 11:00 Coffee Break
- 11:30 RNA-Seq Quantification and Isoforms Finding
- 12:30 Lunch Break
- 14:00 Functional Analysis
- 15:00 Exercises and questions
- 16:00 Tea Break
- 16:30 Exercises and questions
- 17:30 Finish

Ignacio Medina
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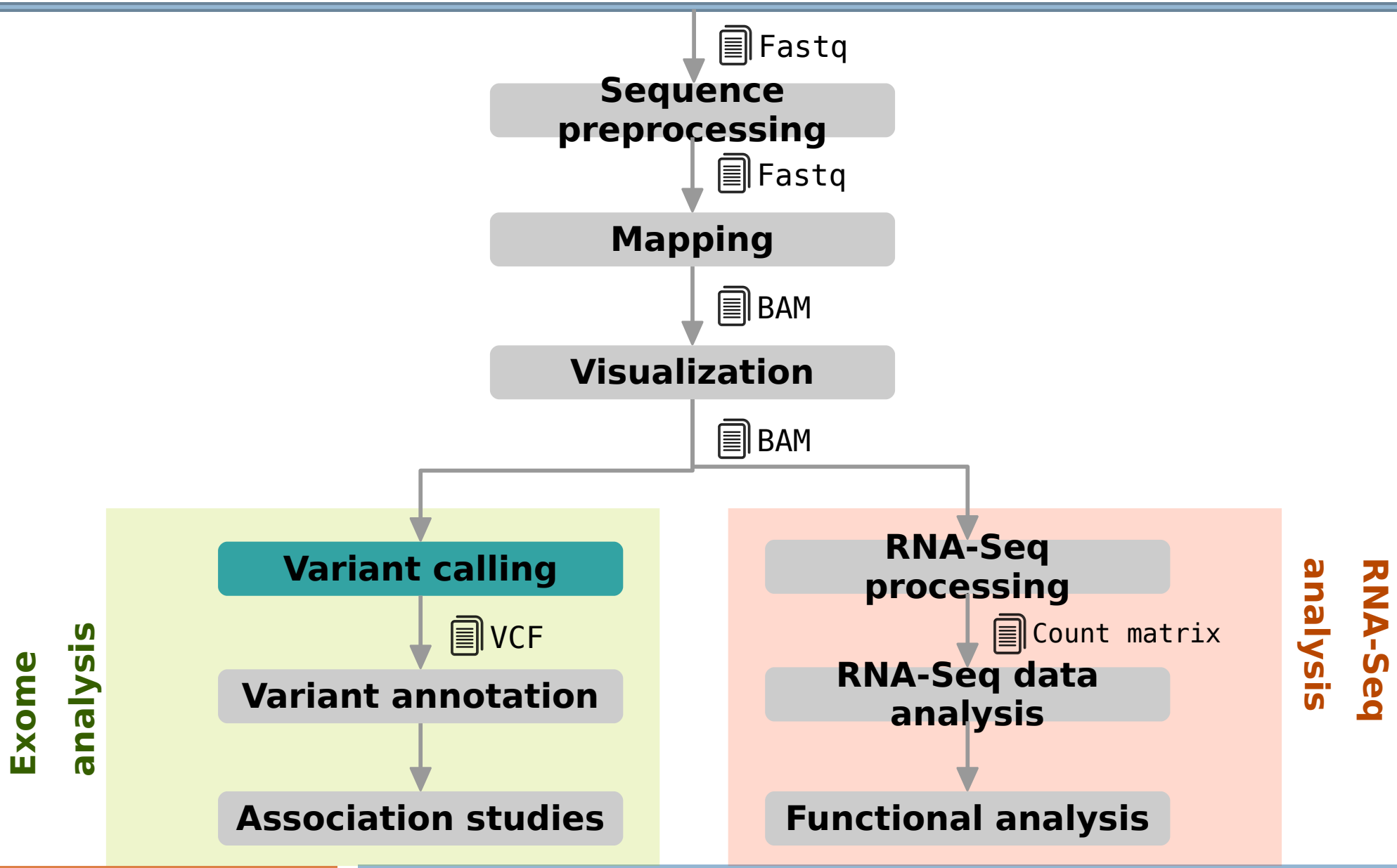
Analysis pipeline



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Presentation

Some considerations

- NGS data is big, very big, huge! Biology is now a Big Data science
  - **No web applications** to perform analysis *yet*, sorry.
  - Most tools developed to work on **Linux**, many command line programs
- How to work in NGS?
  - Small datasets (<1TB): workstations
  - Medium sized datasets (<40-50TB): **clusters**
  - Big datasets (50TB-1PB): distributed and cloud based solutions
- Exercises during the course will be made with **chromosome 21** to speed up analysis and not use to much memory. The whole process is the same
- You are expected to download and prepare the software we are going to use. **Software is not installed intentionally**.

What about you?
Brief presentation

- Who are you?
- Which is your background?
- Which is your interest?
- What do you expect of this course?