Busto Arsizio, July, 1 - 2024

Basic course on NGS

Part 1 (4h). Molini Marzoli Busto Arsizio, July, 1 - 2024

General concepts on Nucleic Acid sequencing
Monday, July, 1 2024. 14-18 – Busto Arsizio
Lecturers: Prof. Bonapace, Prof. Acquati, Dr. Valli

14-14.20
1) DNA sequencing: historical overview and Human Genome Project. 20 min (Acquati)

14.20-14.40
2) Complexity of the functional genome: from the raw DNA sequence to the mature RNA (Anatomy of genes and chromatin organization. The (epi)genome: container of biological information for directing functions of organisms, transmit biological information along generations, and for evolution (Genome organization, DNA sequence and DNA modifications. RNA and RNA modifications. Chromatin structure and organization)). 20 min (Bonapace)

14.40-15.25
3) Sequencing approaches to analyse the functional genome. 45 min (Bonapace)
> Entire genome: whole genome sequencing
> Transcriptional units: Whole-exome sequencing
> Specific genes: Gene targeted sequencing
> DNA modifications: Whole DNA methylation sequencing, RRBSeq, methylation microarray profiling Infinium Methylation-EPIC.
> Transcriptome: RNA sequencing and modifications
> Chromatin occupancy: Chromatin immunoprecipitation (ChIP) and ChIP-seq
> Chromatin accessibility: ATAC-seq (Assay for Transposase-Accessible Chromatin using sequencing)
> Chromatin structure: Hi-C
> What about evolution ?

15.25-15.40
- Break

15.40-16.25
- Sequencing in human diseases. 45 min (Acquati)
> Applications of DNA sequencing in human health and disease
> single point mutations: SNP genotyping
> deletions, insertions, inversions ecc.: whole genome sequencing

16.25-16.55
What biological material can be sequenced ? (Valli)
- Tissues of organisms
- Cell cultures
- Single cells
- Microorganisms

16.55-17.45
Introduction to sequencing methods (Valli)

- From Sanger sequencing to NGS, an overview
- Alternative sequencing methods: Oxford nanopore, PacificBio.
- Future perspectives and what can and can't be learned from genome sequencing ?
- How to select the optimal NGS method

17.45-18.00

- Discussion

Part 2 (4h). Varese, July 3, 2024

NGS platform: from the theory to the practice …
Wednesday, July, 3 2024. 15-18.30
Laboratory of Medical Genetics: Prof. Pallotti and Francesco Orsini
Laboratory of Microbiology: Prof. Nicasio Mancini and Dr. Genoni Angelo

Overview of the platform. Advantages and disadvantages of NGS
1) The NGS workflows. Exon sequencing and microbial genome sequencing as examples
- Next-generation sequencing and a clinical case: searching for the gene
- from the diagnosis to the
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- Prepare the sample and library
- Create the template
- Sequence
2) The Illumina machines - guided tour of the sequencers:
- Illumina MiniSeq: potentials and limits
- Illumina 550DX: potentials and limits

Part 3 (4h). Molini Marzoli – Busto Arsizio, July 3 and 4, 2024

Analyse data and interpret results – Bioinformatics
Friday, July, 5 2024. 14-18 Molini Marzoli – Busto Arsizio
Lecturers: Dr. Paolo Kunderfranco, Bioinformatic manager - Humanitas research
Prof. Fasano

14-14.45 Dr. Paolo Kunderfranco
1) The principles of informational genomics. Basic pipelines for DNA sequencing analysis and Principles of Single-cell transcriptome sequencing

14.45.15.00 Break

15.00-16 Dr. Paolo Kunderfranco
2) Seminar: High-Resolution Analysis of Mononuclear Phagocytes Reveals GPNMB as a Prognostic Marker in Human Colorectal Liver Metastasis

16.16.20 Break

16.20-17.45 Prof. Mauro Fasano
3) Results analysis: Dimensionality reduction in datasets. Feature extraction and subset selection. Cross-validation. Over-representation analysis. Gene/protein databases. Ontology and pathway databases. Gene-set enrichment analysis

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