



UNIVERSITÀ  
di **VERONA**

Scuola di Dottorato  
di **SCIENZE NATURALI  
ED INGEGNERISTICHE**

# PhD Course on "Theory and Computational Practice for Genome Variant analysis using high throughput sequencing data"

**Luciano Cascione, PhD**

Università della Svizzera italiana (USI)

Swiss Institute of Bioinformatics (SIB)

Lymphoma & Genomics Research Program | Bioinformatics Core

Unit at Institute of Oncology Research (IOR)

## Agenda

- Introduction to NGS technologies and experimental design (WGS vs Exome vs Targeted Sequencing (Gene Panels))
- Data Pre-processing, from Fastq to BAM
  - Indexing Reference Genome
  - Mapping reads to a reference genome
  - Sorting and indexing alignment
  - Map quality control
- Variant Discovery, from BAM to VCF
  - Difference in workflow between DNA-Seq and RNA-Seq
  - Marking duplicates, Realignment around SNPs and indels, base quality score recalibration
  - Calling Variants: Somatic vs Germline SNPs + indels + CNVs
- Callset Refinement
  - Variants Annotation and Filtering
  - Genotype Refinement
- Practical Session: Reads-to-variants workflow

## Class meeting

December 11 8.00 – 13.00 Sala Verde

December 12 8.00 – 13.00 Sala Verde

December 13 8.00 – 13.00 Sala Verde



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Local organization and contact:

Prof.ssa Rosalba Giugno

[rosalba.giugno@univr.it](mailto:rosalba.giugno@univr.it)

Students attending this PhD Course are entitled to 4 CFR valid for the specific activities of the Graduate School.

**Registration is mandatory. Please send email to [rosalba.giugno@univr.it](mailto:rosalba.giugno@univr.it)**

**Attendance is restricted to the first 30 registered students.**

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**Scuola di Dottorato di Scienze Naturali ed Ingegneristiche**

Strada Le Grazie, 15 - 37129 Verona | T + 045 802 7026

[laura.marcazzan@univr.it](mailto:laura.marcazzan@univr.it)

P. IVA 01541040232 | C.F. 93009870234