VELSERA

SevenBridges

Applied Bioinformatics

reveals

Agenda

- Introductions
- Course overview
- Bioinformatics intro
- Platform registration

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Applied Bioinformatics

Introductions: Lecturers

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Different backgrounds - bioinformatics engineers

- Bojana Smiljanić, BF

- Boris Majić, ETF

- Marko Zečević, ETF

- Milan Kovačević, PMF

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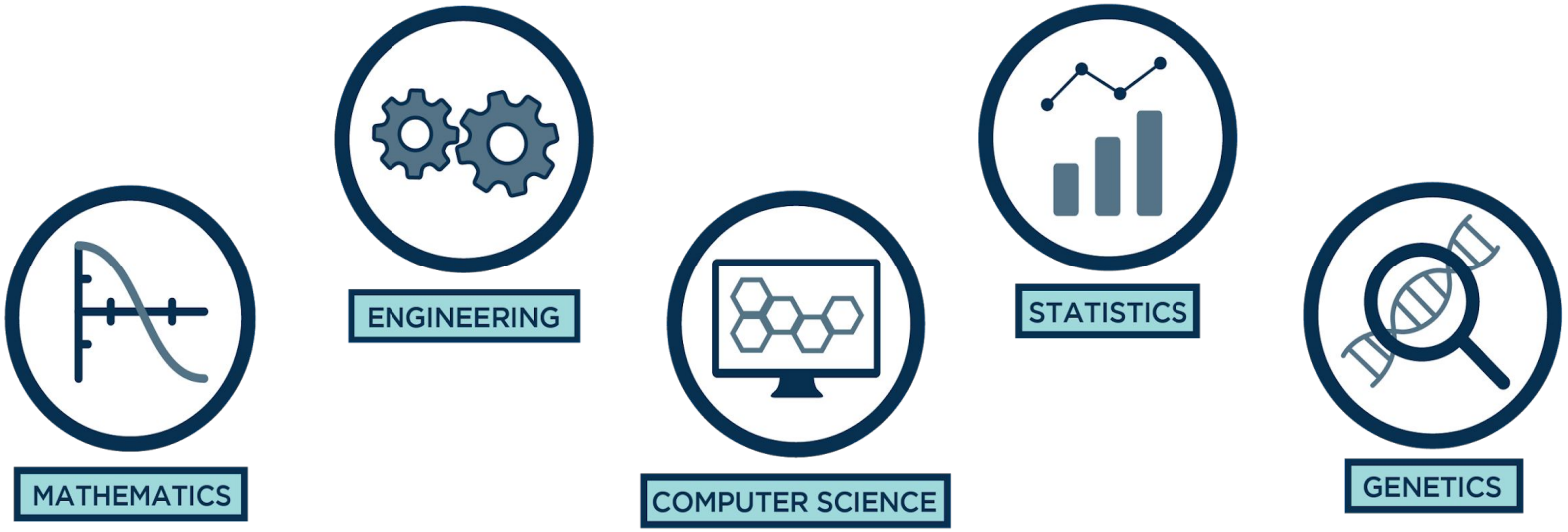
Bioinformatics analyst



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Applied Bioinformatics

Course overview

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Course logistics (1/2)

- 2 classes each week
- A mixture of lectures and hands-on exercises
- Exercises will be done in IPython notebooks on the CGC platform
    - We will provide help with the Python syntax if needed
- The course is not covered by a single textbook
- Lessons are mostly linked

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Course logistics (2/2)

- Attending classes is mandatory and highly recommended
- Practice test in the end of semester
- For all course related questions contact
  - milan.kovacevic@velsera.com

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Course topics

- Introduction to biological background and sequencing (2 weeks)
- DNA analysis (4 weeks)
- RNA analysis (2 weeks)
- Structural variation detection (1 week)
- Cancer genomics (1 week)
- Methylation/epigenetics analysis (1 week)
- Exam practise (1 week)

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Bioinformatics intro

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Bioinformatics intro

During this course you will learn/hear:

- More about
  - Human cells
  - DNA
  - RNA
- Why is the DNA so important?
- What can we find out from the DNA?
- More about certain diseases, like cancer
- Genome digitalisation
- How to analyse genomic data
- Interesting algorithms
- Optimisations for huge data

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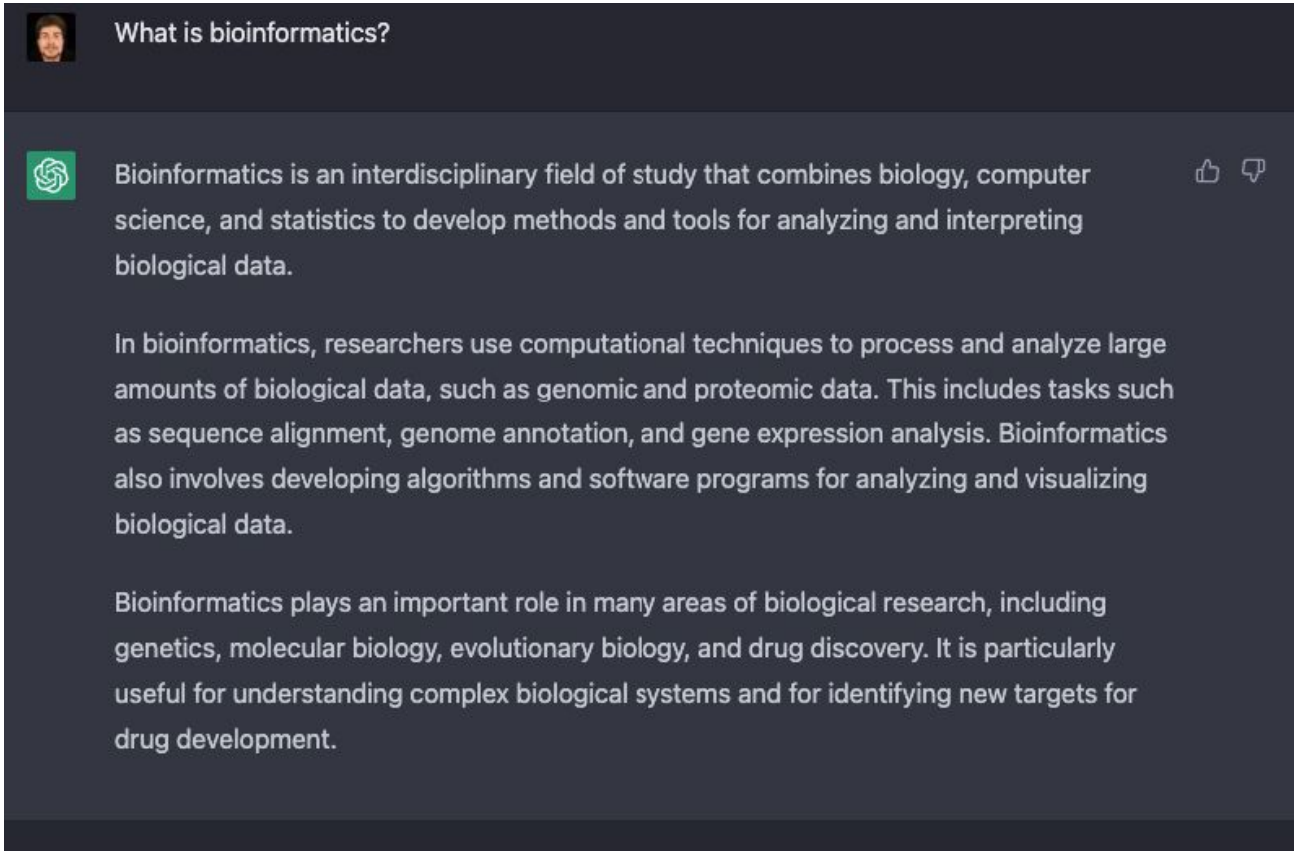
What is bioinformatics?



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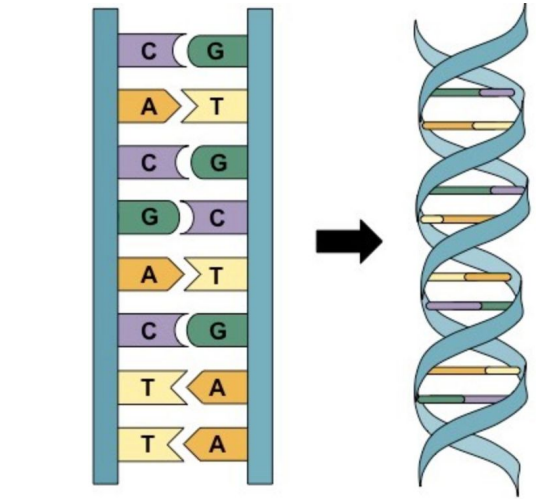
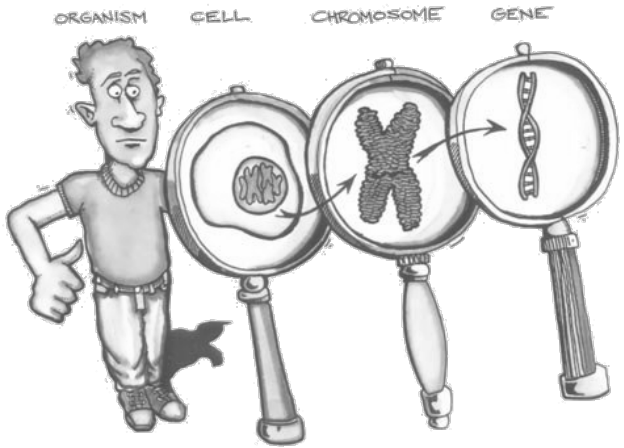
Bioinformatics intro

- A, T, C, G
- 3.000.000.000 letters in a DNA molecule
- 46 chromosomes
- Almost everything in the body is predefined by the order of those letters





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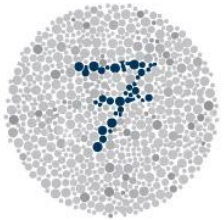
Bioinformatics intro

Some interesting characteristics:

LONGER EYELASHES



DALTONISM

LESS SLEEPING

SUPER STRENGTH

And some not that interesting things:

- Cancer
- Rare diseases
- Autoimmune diseases
- Neurological diseases

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DNA digital representation

- DNA is stored as a string
- Human genome project and reference genome
- 3 billion As, Ts, Cs and Gs translates into ~ 3 GB of data
  - font 4

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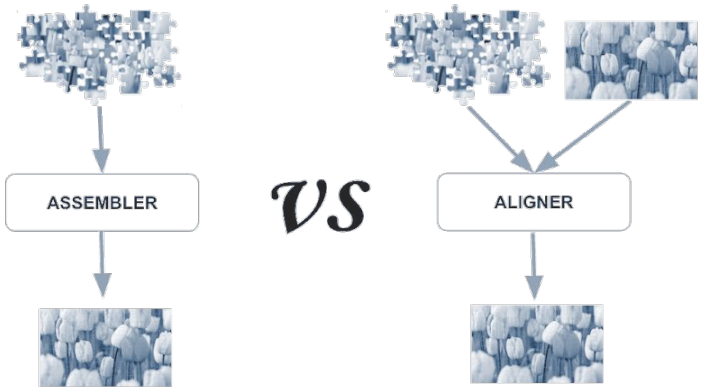
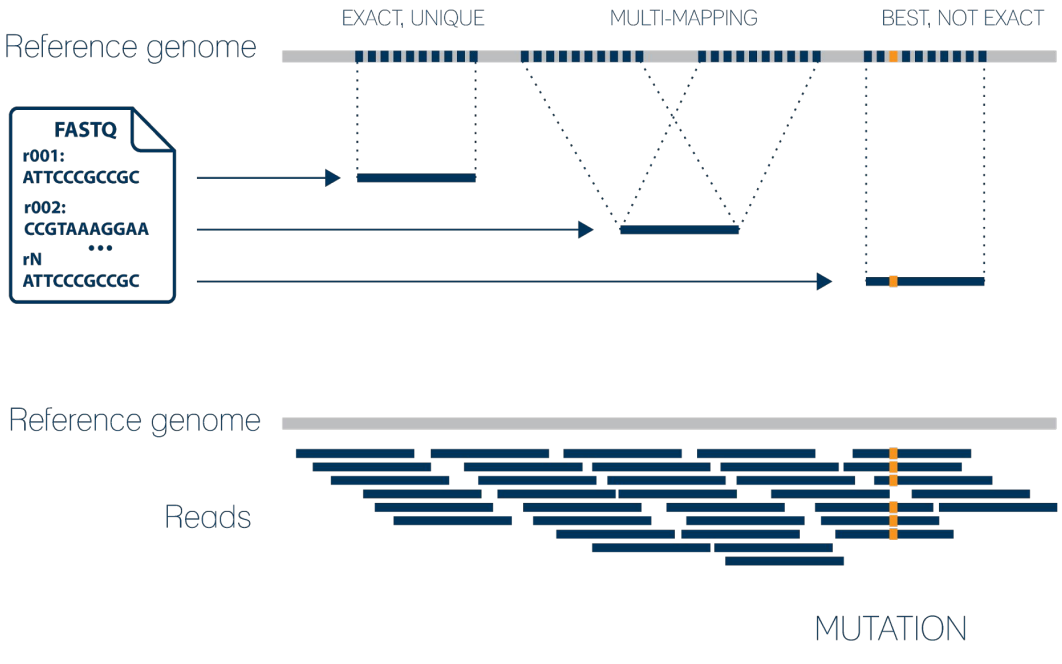
Bioinformatics intro

Common 1st step:
Reconstruct personal genome



$$\Theta(n^2) \text{ vs } \Theta(nm)$$



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Bioinformatics intro

Single nucleotide variant

Deletion

Insertion

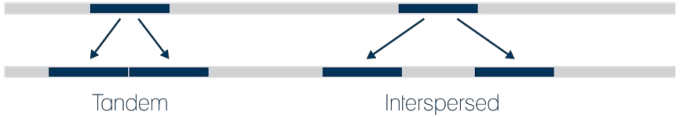
Inversion

Copy number variant

Translocation

Whole genome duplication

Duplication



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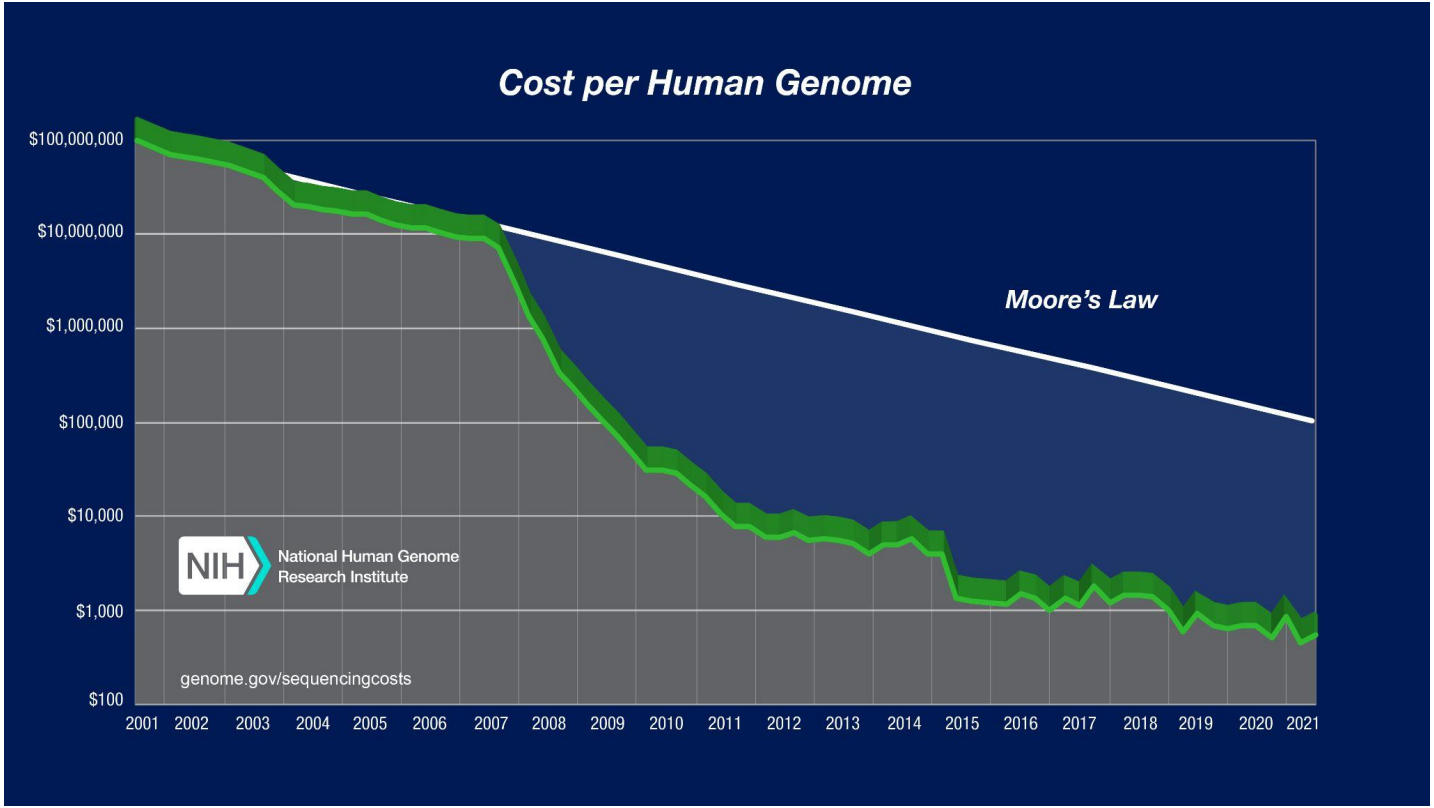
Why bioinformatics



source: https://www.genome.gov/about-genomics/fact-sheets/DNA-Sequencing-Costs-Data

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Why bioinformatics

My Full DNA: Whole Genome Sequencing with mtDNA

€259.00 EUR ~~€850.00 EUR~~ You save €591.00 EUR

Type of report: My Full DNA... 1

ADD TO CART

DESCRIPTION

My Full DNA by Dante Labs gives you unparalleled access to your health, your family traits, and predispositions. Having your full genome sequenced means:

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Platform registration

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CGC registration

- Exercises are going to be done on CGC platform
- **cgc.sbgenomics.com**
- CGC = Cancer Genomics Cloud
  - Funded by NCI - National Cancer Institute (NIH)
  - Powered by Seven Bridges
  - For academic use
  - Many researchers/ institutes / labs are using it for their analysis
- More information about the project available here.
- Registration

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Course materials

https://github.com/milankovacevicsbg/MATF-2023-Primenjena-Bioinformatika

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Thank you!

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