生物信息学：导论与方法
Bioinformatics: Introduction and Methods

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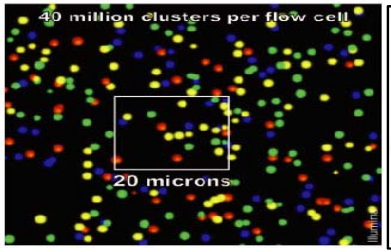

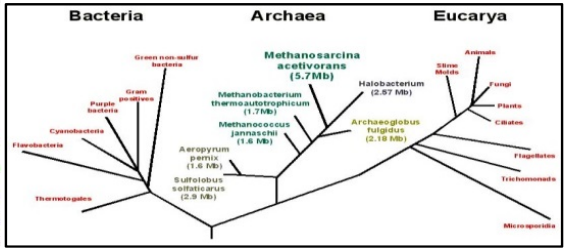

Introduction and History

北京大学生物信息学中心 魏丽萍

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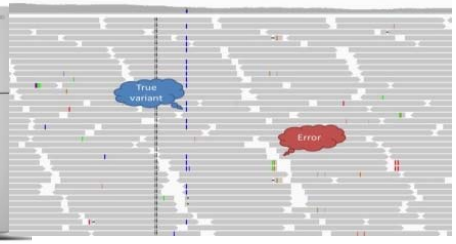

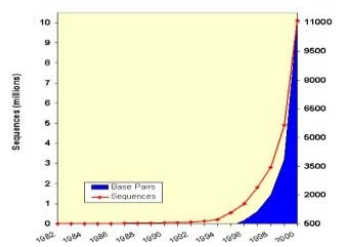

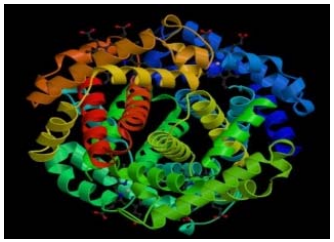

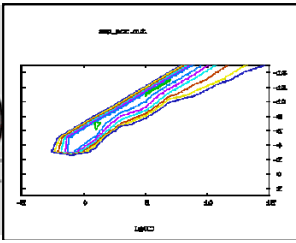

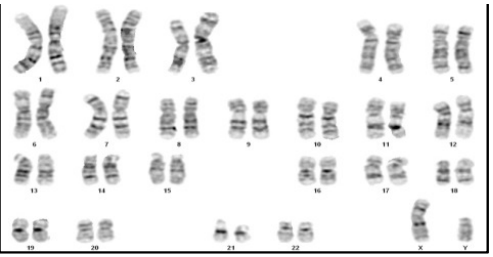

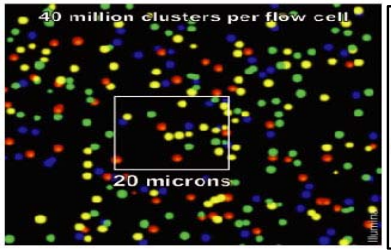

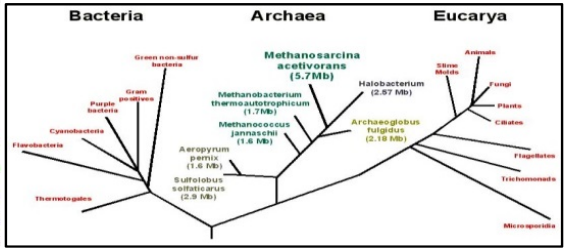

Unit 1:
What is bioinformatics?

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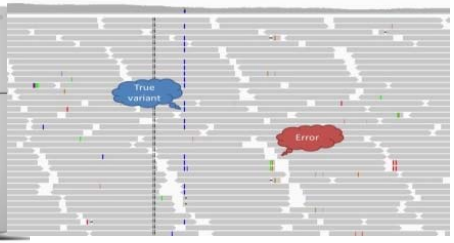
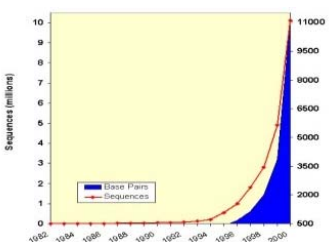
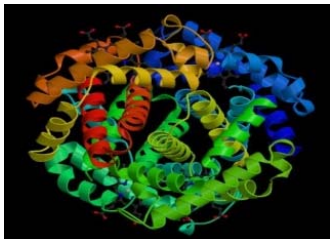
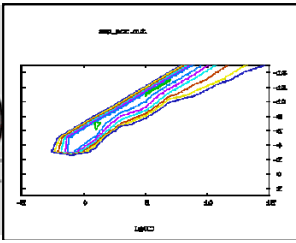
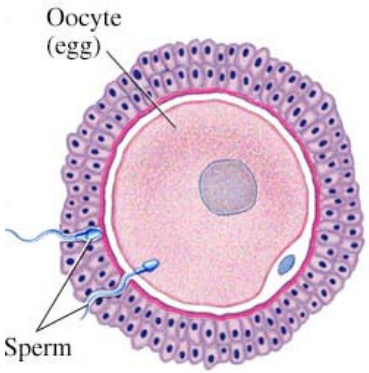








The miracle of life



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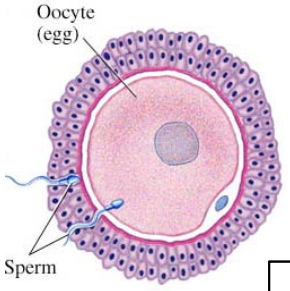
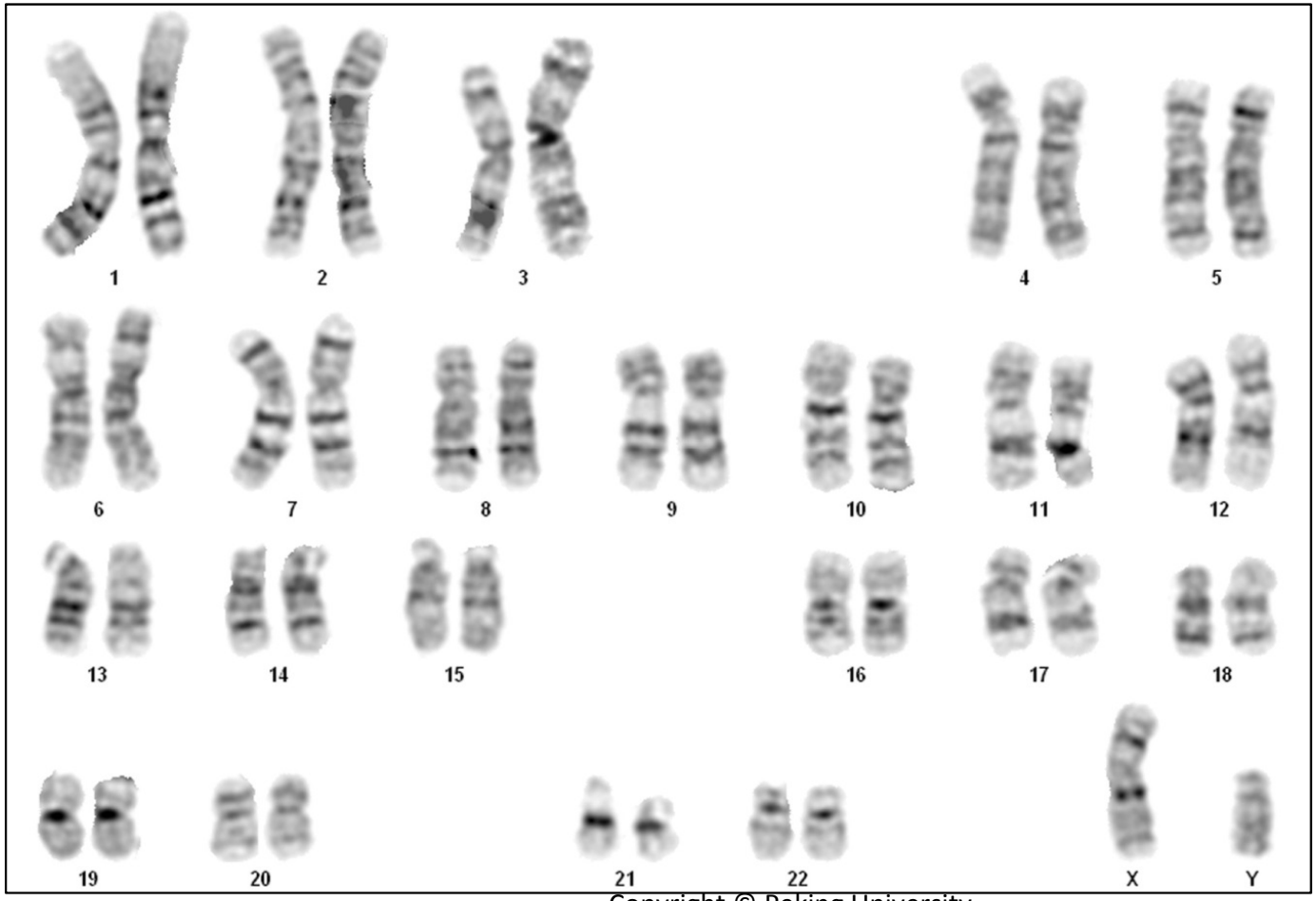


Genome: the “manual of life” -- ***Almost***

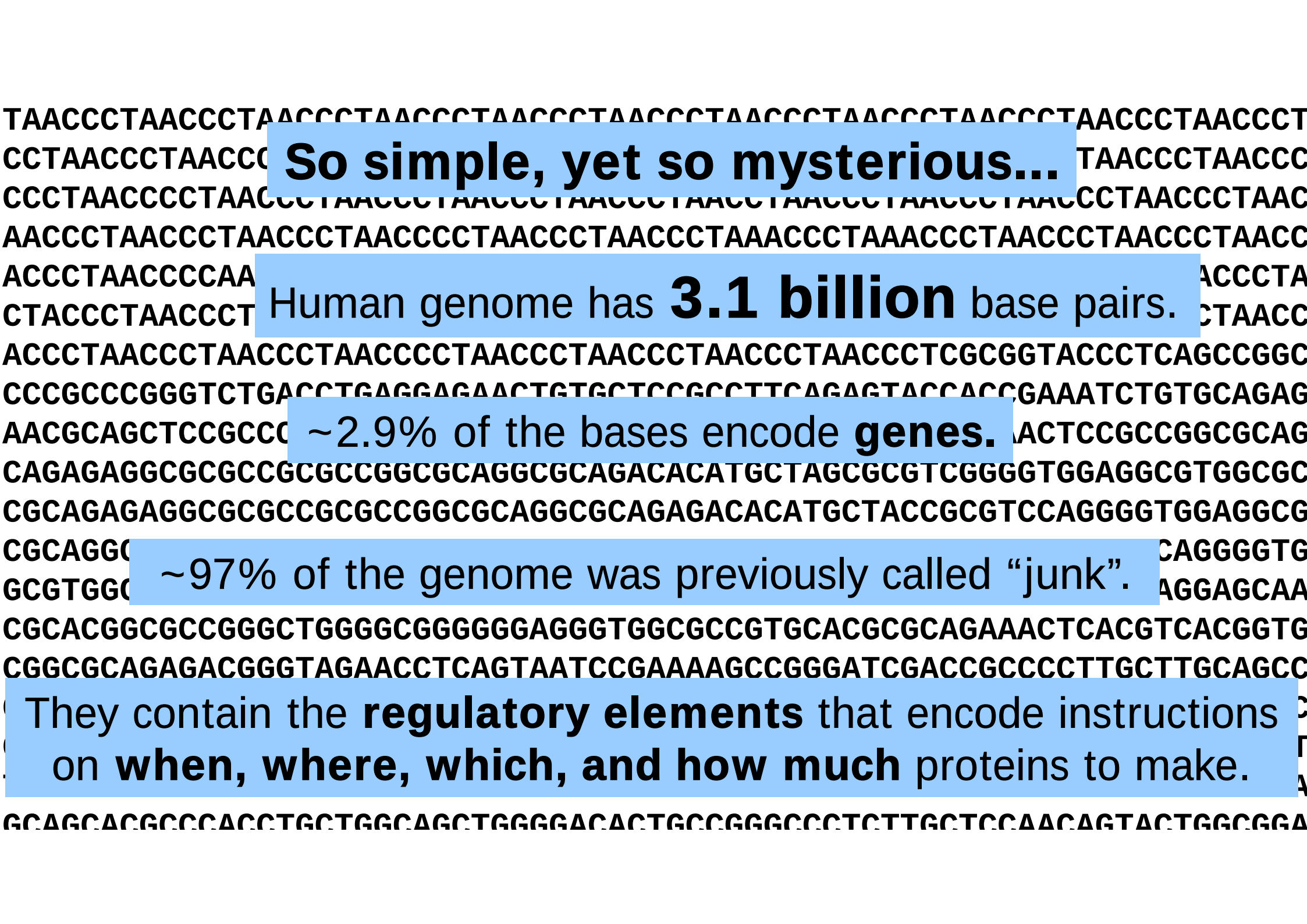


mitochondrial DNA

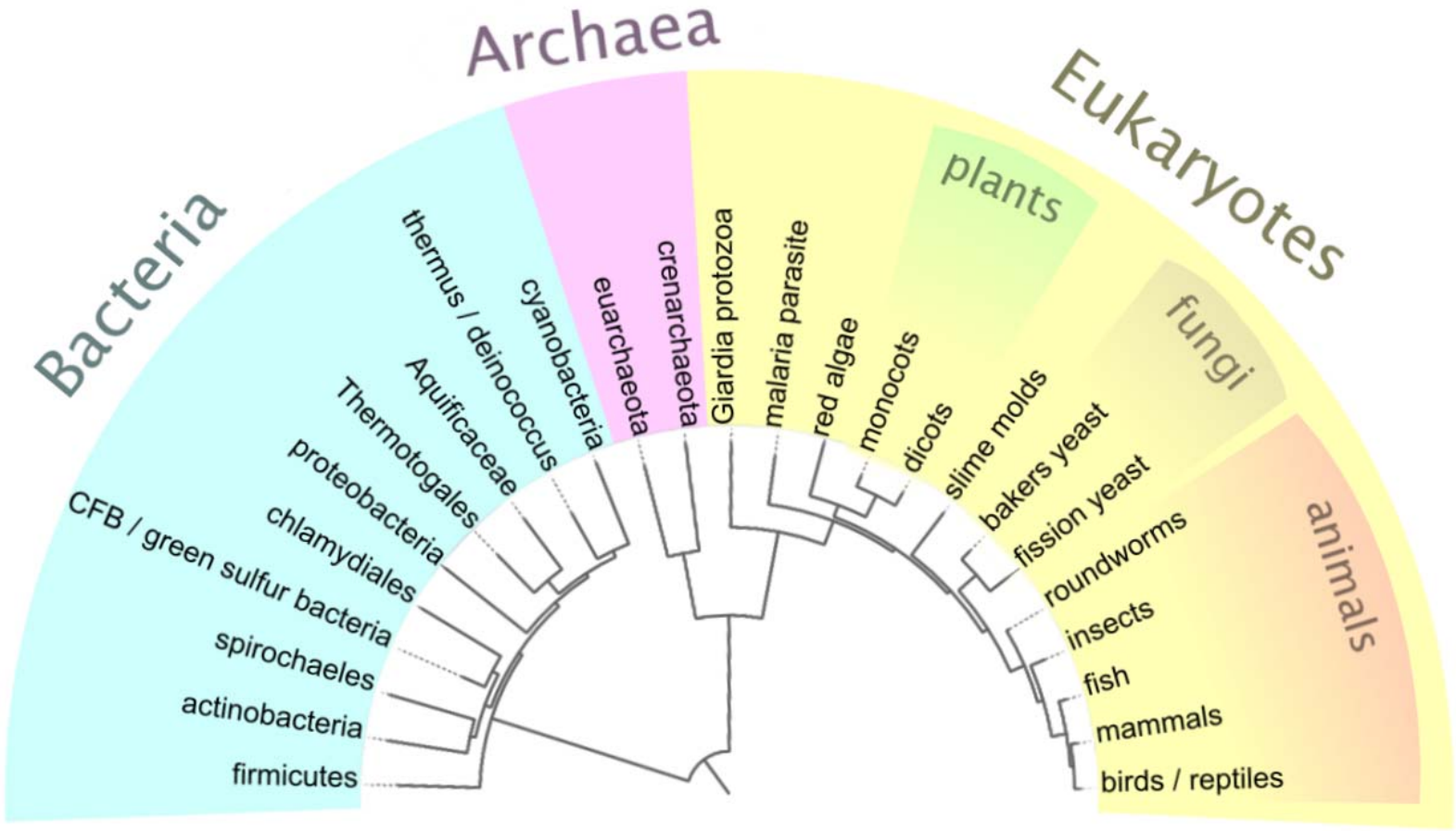
epigenetics

environments/nurture

chance

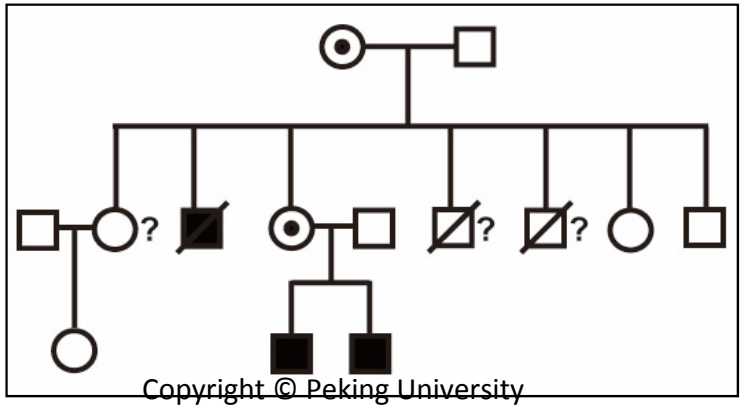

So simple, yet so mysterious...
Human genome has 3.1 billion base pairs.
~2.9% of the bases encode genes.
~97% of the genome was previously called “junk”.
They contain the regulatory elements that encode instructions on when, where, which, and how much proteins to make.

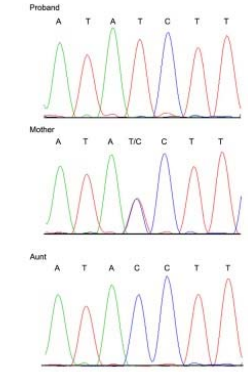
The universal code: Other species' genomes
The Tree of Life

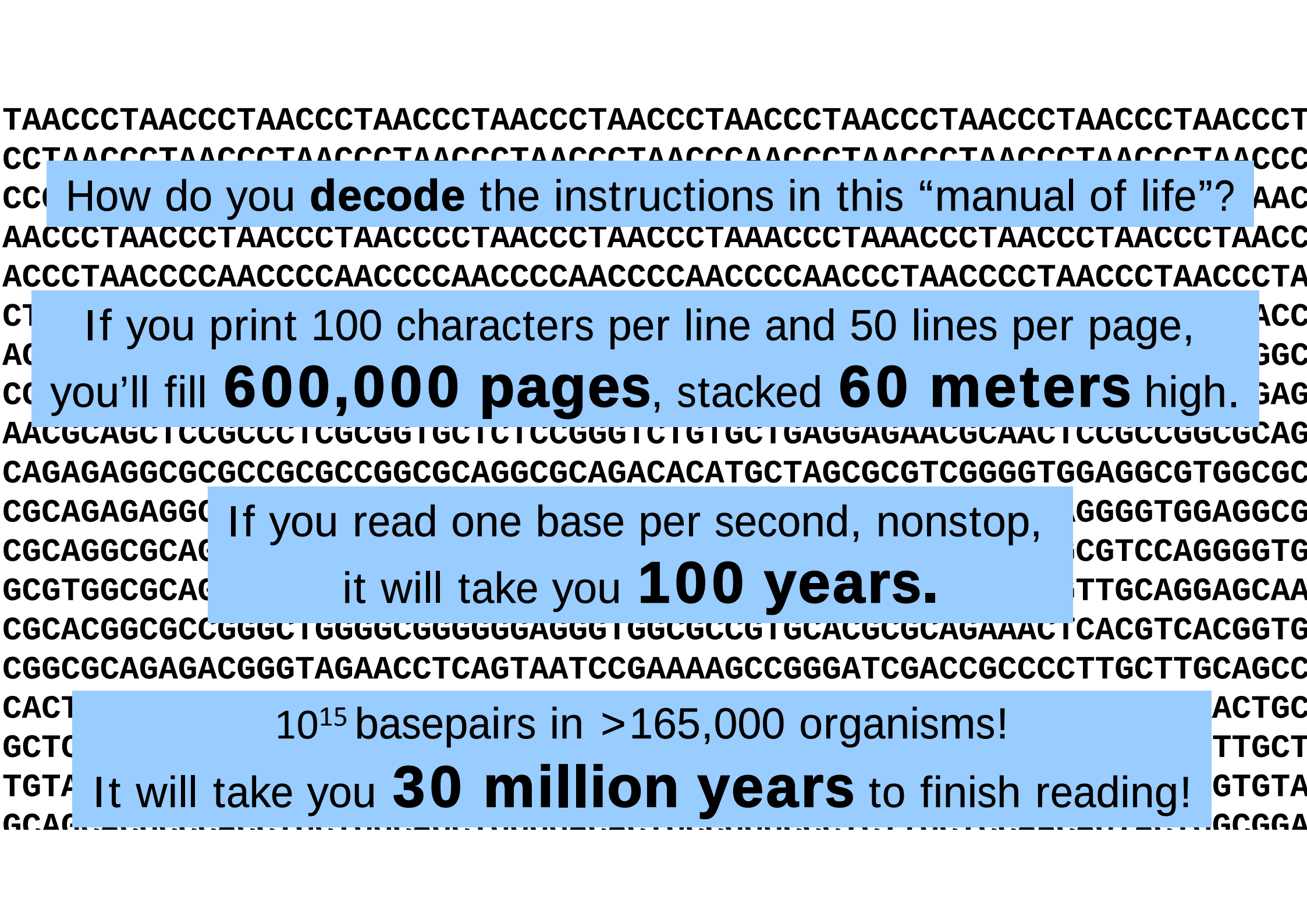


http://commons.wikimedia.org/wiki/File:Simplified_tree.png

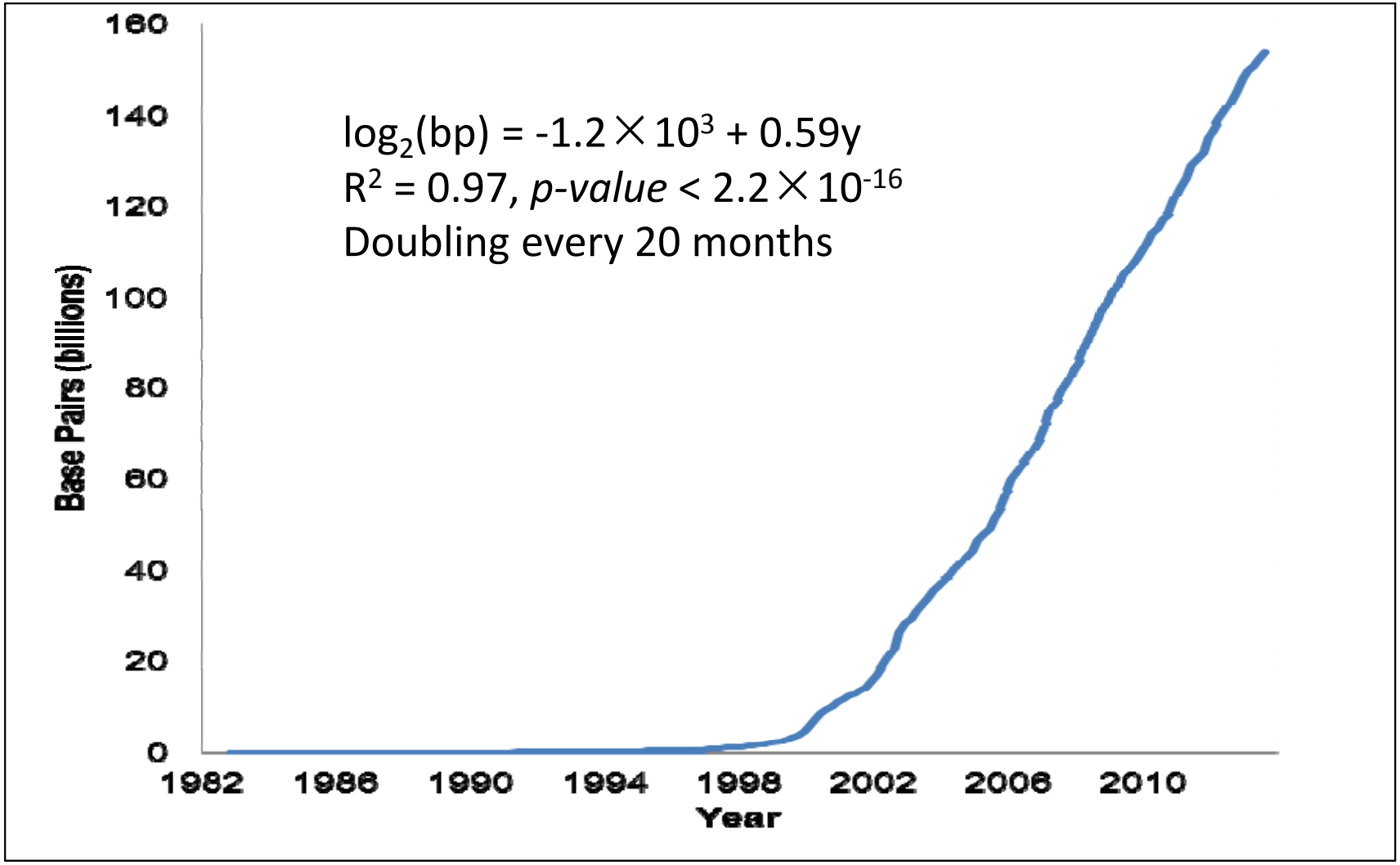
Human Genetic Variations





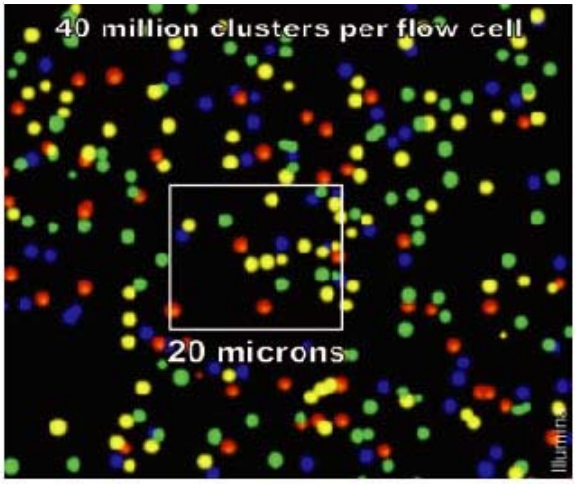

How do you **decode** the instructions in this “manual of life”?
If you print 100 characters per line and 50 lines per page,
you’ll fill **600,000 pages**, stacked **60 meters** high.
If you read one base per second, nonstop,
it will take you **100 years.**
10^{15} basepairs in >165,000 organisms!
It will take you **30 million years** to finish reading!

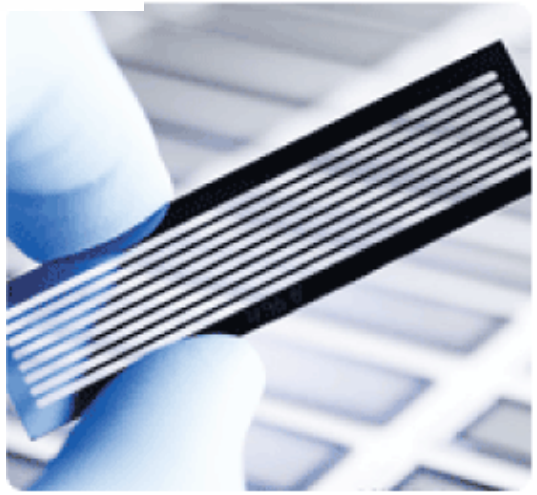
Genbank growth

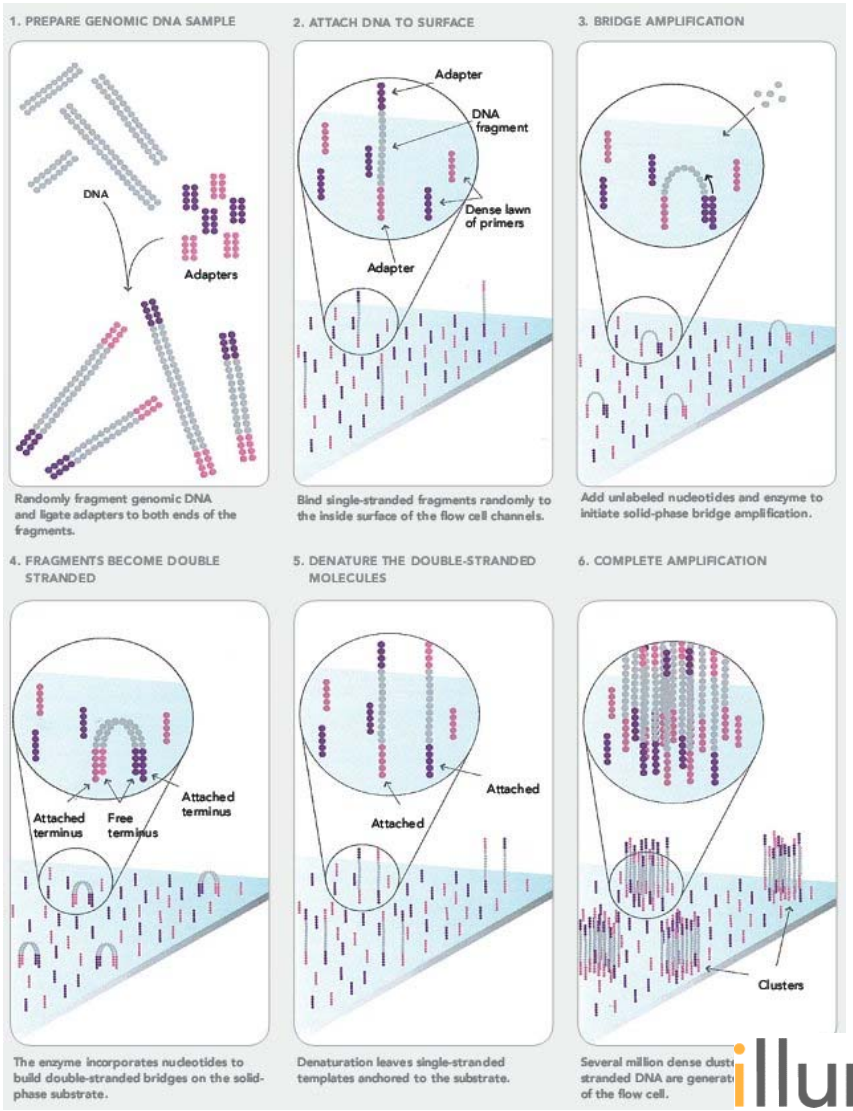


Data Source: ftp://ftp.ncbi.nih.gov/genbank/gbrel.txt

Next-Generation Sequencing: Your genome, one day, $3000!

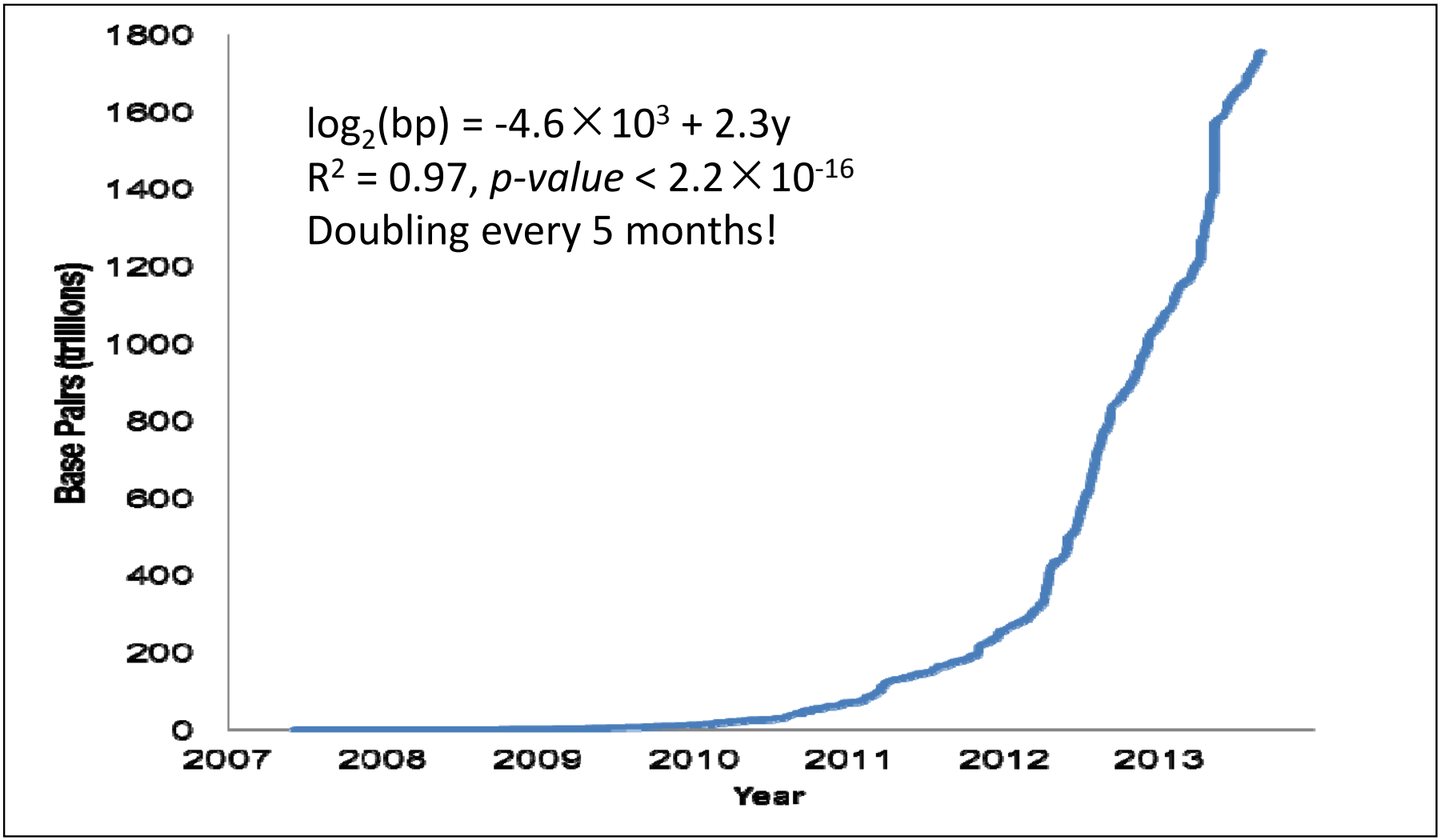






illumina®

SRA Growth



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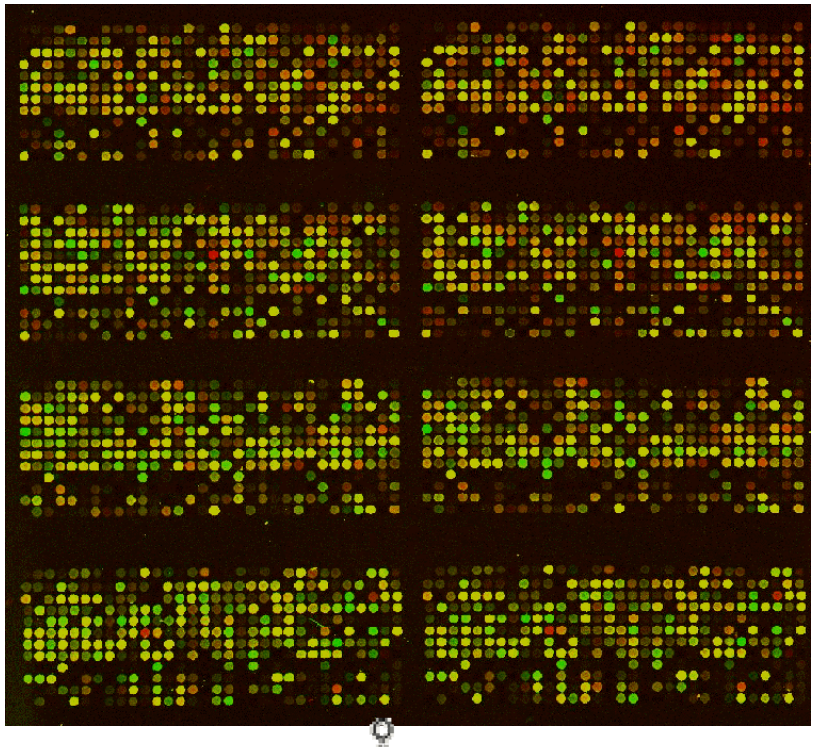

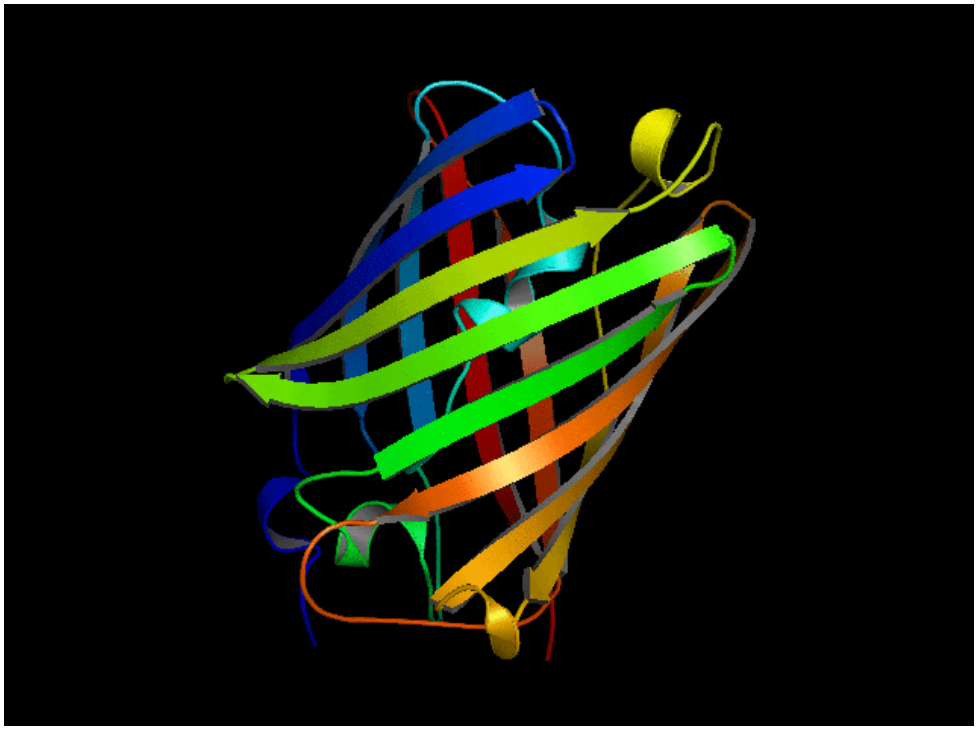

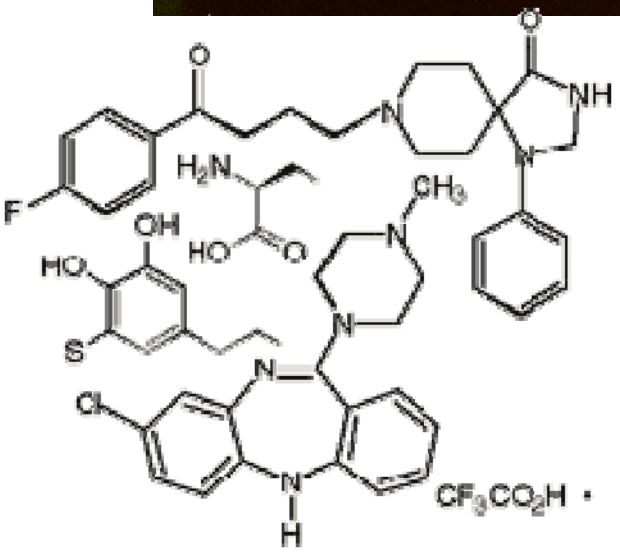
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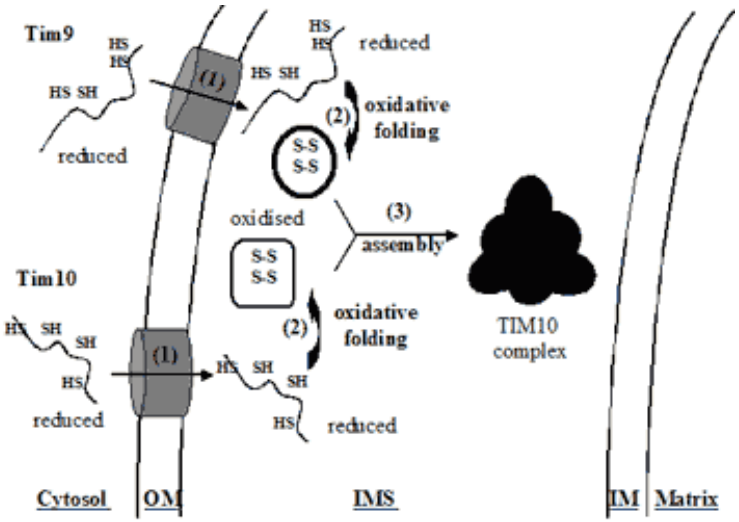
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TIM10
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OM
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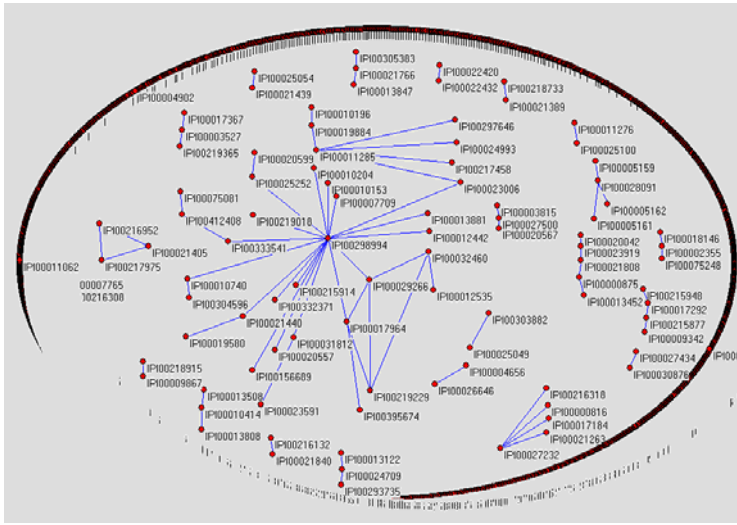
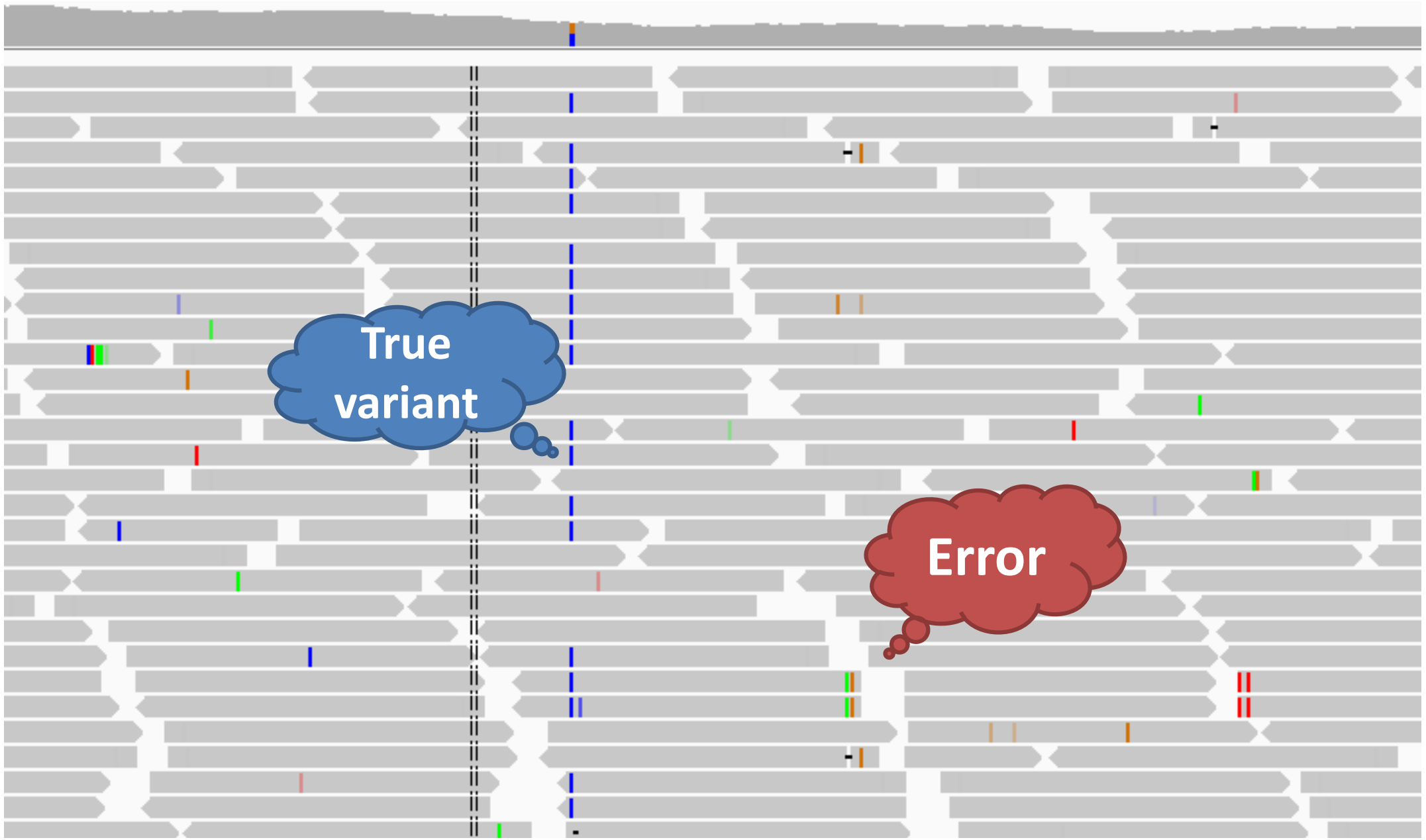
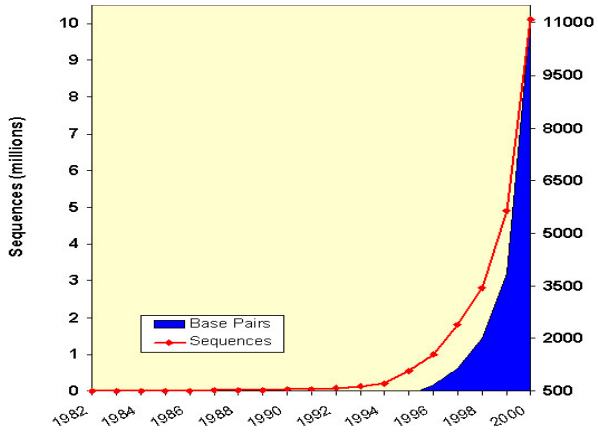



True variant
Error

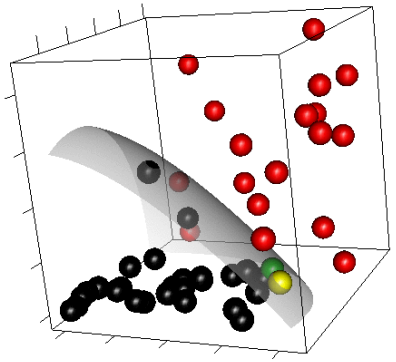
Opportunities and challenges hand-in-hand: the driving forces of bioinformatics

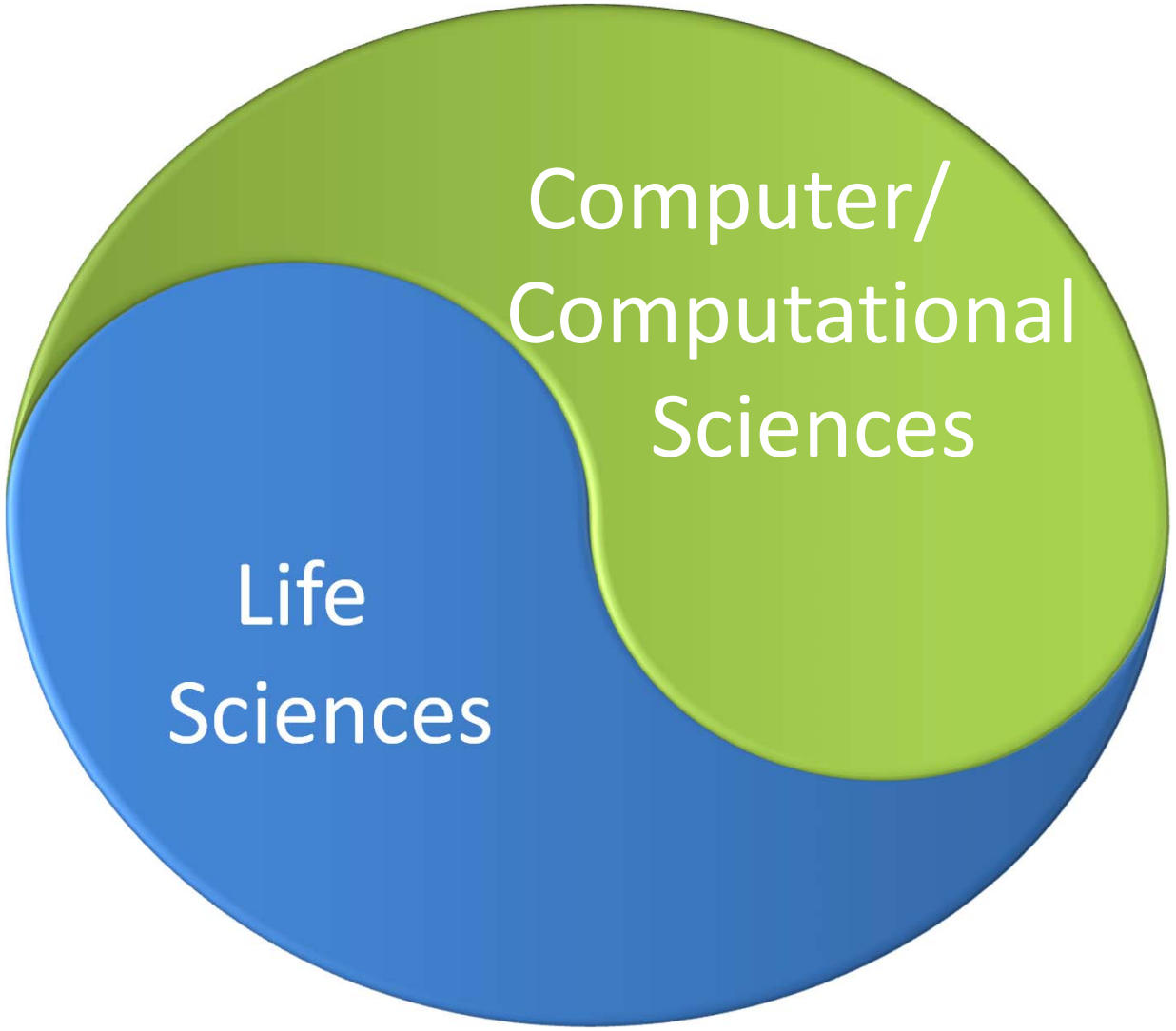
- High-throughput data
  - Huge amount
  - Explosive growth
  - Low signal-to-noise ratio
  - Multiple types



ftp://ftp.ncbi.nih.gov/genbank/gbrel.txt

- Requirements for the methods
  - Data needs to be stored in efficient ontology-based database systems
  - The huge amount of data requires efficient algorithms
  - Exponential growth requires scalable methods
  - The low signal-to-noise ratio requires accurate methods
  - Multiple types of data require data integrative methods





Bioinformatics

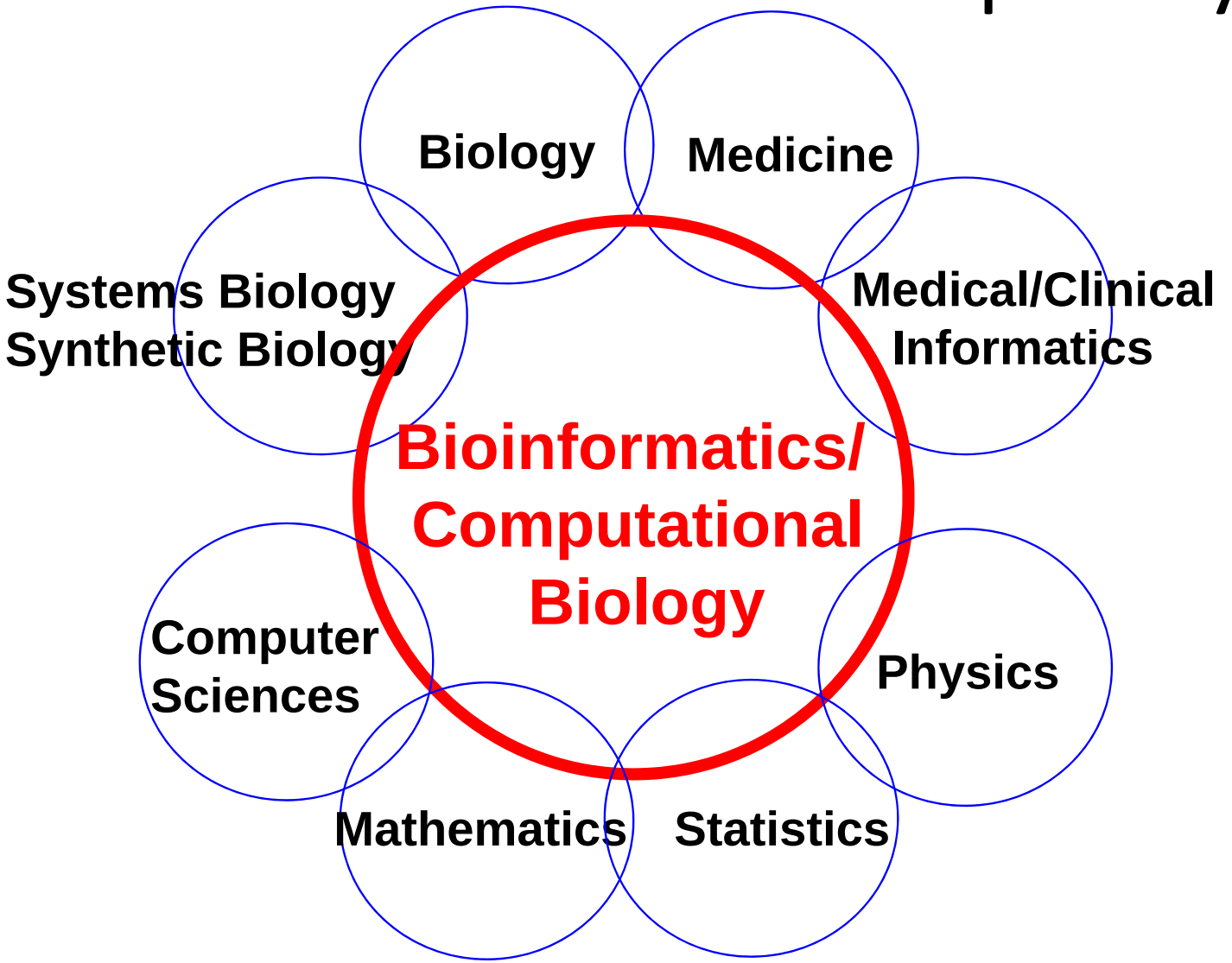
What is bioinformatics?

Bioinformatics: an interdisciplinary field that develops and applies computer and computational technologies to study biomedical questions

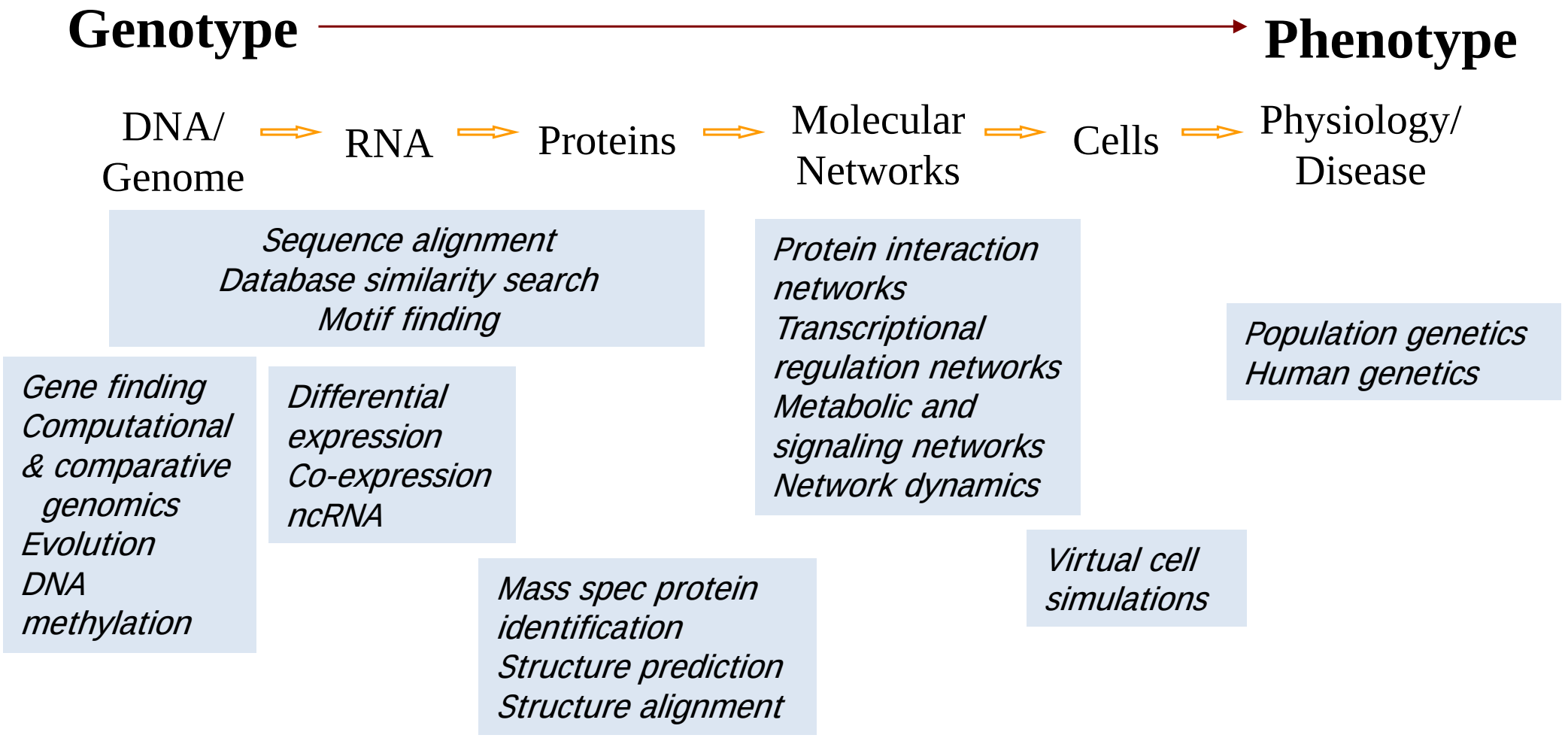
- As a technology, bioinformatics is a powerful technology to manage, query, and analyze big data in life sciences.

- As a methodology, bioinformatics is a top-down, holistic, data-driven, genome-wide, and systems approach that generates new hypotheses, finds new patterns, and discovers new functional elements.

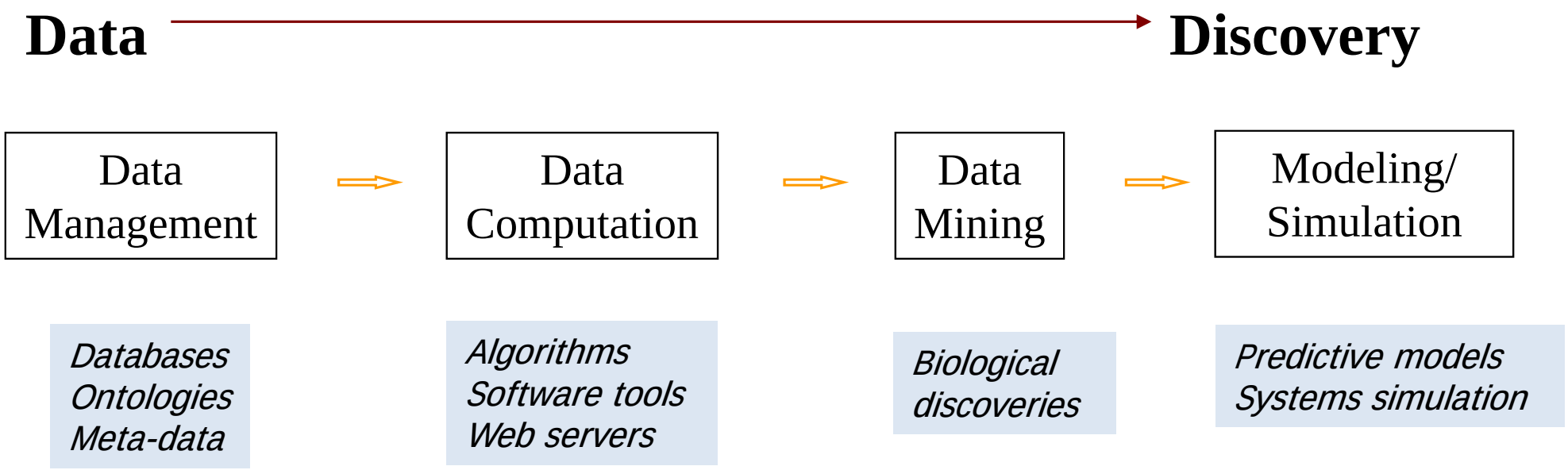
Bioinformatics is an interdisciplinary field



The Bio- in Bioinformatics



The –informatics in Bioinformatics



Summary Questions

- What is bioinformatics?
- Can you name some of the biomedical questions studied in bioinformatics?
- Can you name some of the technical questions studied in bioinformatics?
- Why are you interested in bioinformatics?

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