

Introduction to Bioinformatics

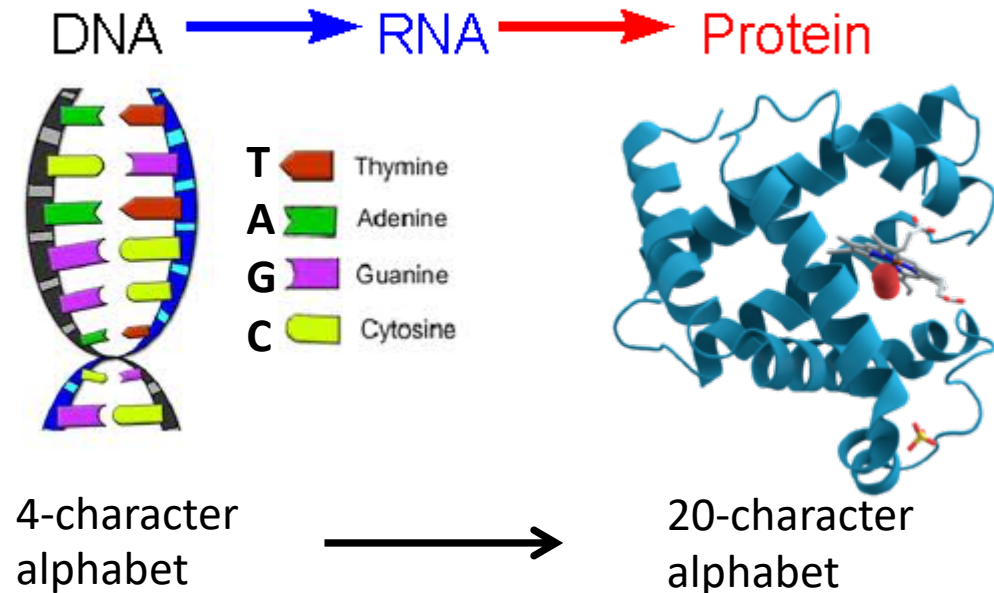
CSC 314

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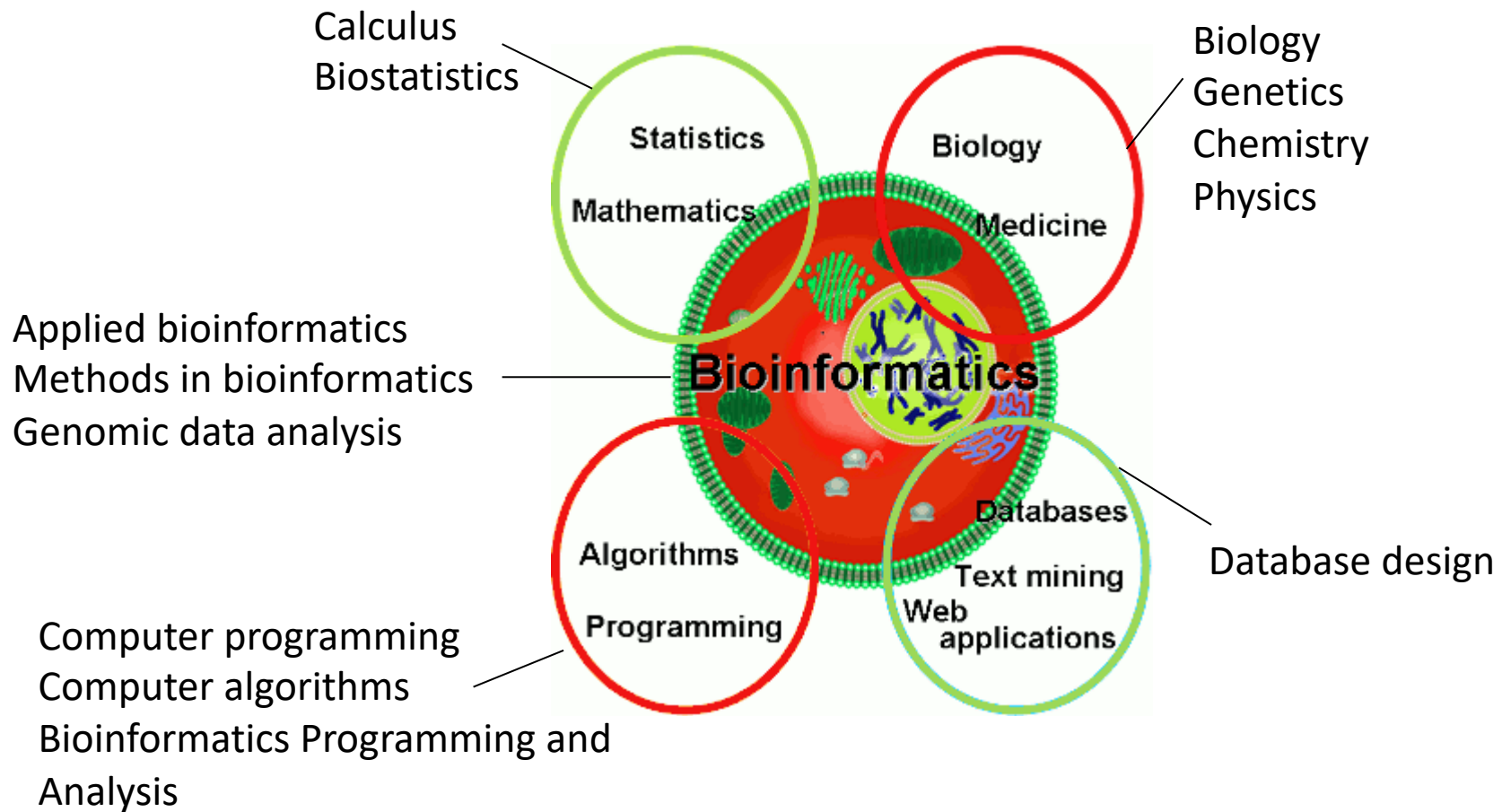
What is bioinformatics

- Bioinformatics:
 - Biology + information
 - the study and utilization of methods for storing, retrieving and analyzing biological data
 - Falls under the category of Big Data



- How much information:
 - Human genome: 3 billion nucleotides
 - ~20,000 genes
 - many more when considering “junk DNA” and alternative splicing
 - >10 million sites of DNA variation
 - Countless possible interactions between DNA, RNA, and proteins

Bioinformatics is interdisciplinary



What is Big Data?

- Datasets that are "large" or at least complex
- Generated by scientific studies, technology, and commerce
- From technology
 - <https://web-assets.domo.com/blog/wp-content/uploads/2020/08/20-data-never-sleeps-8-final-01-Resize.jpg>
 - <http://www.internetlivestats.com/>

Big Data Examples

- What does FB know about you?
 - <http://www.nbcnews.com/science/gay-conservative-high-iq-your-facebook-likes-can-reveal-traits-1C8805606>
- What does Target know about you?
 - <http://www.nytimes.com/2012/02/19/magazine/shopping-habits.html>
- What information does your genome reveal about you?
 - Sex
 - Paternity and ancestry
 - Eye color
 - Relative Risk of breast cancer
 - Relative Risk of alcoholism

(Note that many genetic traits are complex, not well understood, and genetic predictions can be difficult to interpret; genetic traits are often not black/white, and genetic traits are often influenced by environmental factors)

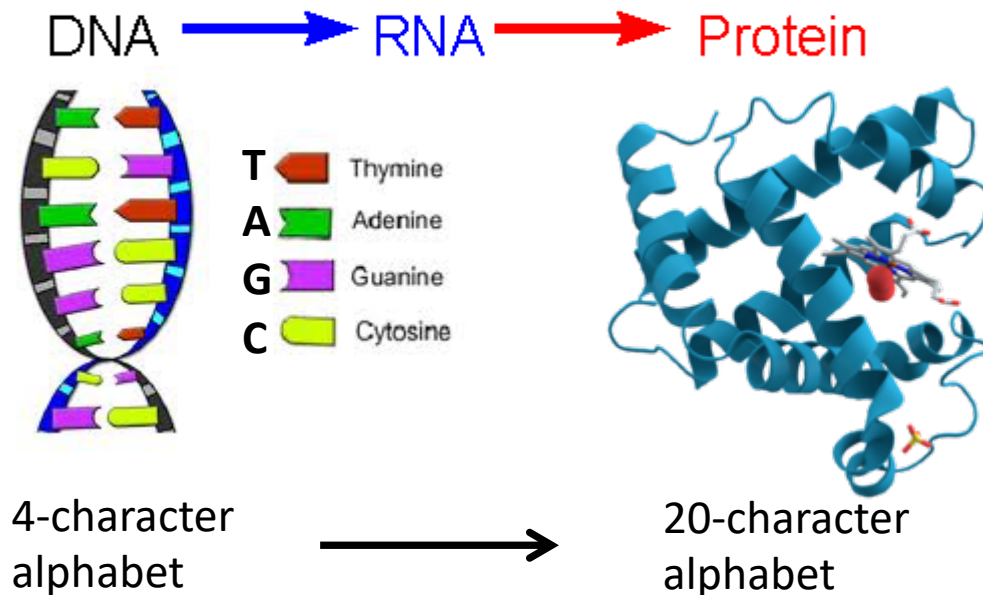
What is this?

```
public class helloWorld {  
    public static void main(String[] args) {  
        System.out.println("Hello World!");  
    }  
}
```

Bioinformatics is an information science

- **Computer code** is a *set of instructions* that tells a computer how to process data and output results
- The **genetic code** is also a set of instructions, that tells a cell how to produce a molecule (RNA/protein) from DNA
 - Information flows from DNA → RNA → protein
 - The DNA information determines the structure/function of RNA and protein

Central Dogma of Molecular Biology



- The function of a protein can be predicted from its DNA or protein sequence
- Just like Java is a language for computers, genetics is the language of life (DNA is the alphabet)
 - This is a fundamental concept in bioinformatics

Bioinformatics example

- Let's look briefly at the gene *BRCA1* (breast cancer type 1 susceptibility protein)
 - <http://www.ncbi.nlm.nih.gov>
 - Search Nucleotide for BRCA2
 - Click on the RefSeqGene entry
- This is a preview and will make much more sense by the end of the semester

Intro to Genetics (Genetics 101)

- What are genes?
 - http://www.youtube.com/watch?v=ubq4eu_TDFc
 - Genes are part of what molecule?
 - How many bases is DNA composed of?
 - How are genes organized?
 - How many *pairs* of chromosomes do humans have?
- What are SNPs?
 - <http://www.youtube.com/watch?v=tJjXpiWKMyA>
 - What is the human genome?
 - What is a SNP?

Intro to Genetics (Genetics 101)

- Where do your genes come from?
 - <http://www.youtube.com/watch?v=-Yg89GY61DE>
 - Where do your genes come from?
 - What are homologous chromosomes?
 - What determines your sex?