

DATA ANALYSIS IN GENETICS

01 Data Analysis in Genetics

Chapter Outline

1. Omics
2. Biology and Big Data
3. Genome Features
4. Genome Study

Learning Objectives

After completing this chapter, the student will be able to:

- explain the paradigm shift in Biology and the need for Big Data Analysis
- describe the main features of genomes
- describe different perspectives on genomics research

02 DNA Sequencing Technology

Chapter Outline

1. First Generation DNA Sequencing
2. Second Generation DNA Sequencing
3. Third Generation DNA Sequencing
4. Genome Sequencing

Learning Objectives

After completing this chapter, the student will be able to:

- understand sequencing technologies: chain termination method, sequencing by synthesis, Single Molecule Real Time sequencing
- explain Paired-End Sequencing
- explain depth and breadth of sequencing coverage
- describe Hierarchical and Whole Genome Shotgun Sequencing

03 Genome Assembly

Chapter Outline

1. FASTQ Files
2. De novo Assembly
3. Reference-based Assembly
4. SAM Files

Learning Objectives

After completing this chapter, the student will be able to:

- describe the structure of FASTQ and SAM files
- explain de novo and reference based assembly
- assess assemblers performance

04 Variant Calling

Chapter Outline

1. Single Nucleotide Polymorphisms and Structural Variations
2. Variant Calling Workflow

3. VCF File Format

Learning Objectives

After completing this chapter, the student will be able to:

- describe types of genomic variants and how they are determined
- explain types of error associated with alignment, assembly, and variant calling
- describe VCF file data format

05 Genome Annotation

Chapter Outline

1. Structural Annotation
2. Functional Annotation
3. Submit Genome Annotation to Databases

Learning Objectives

After completing this chapter, the student will be able to:

- understand the need for repeat identification
- explain gene prediction methods
- describe functional annotation results
- describe GFF3 file data format

06 RNA Sequencing

Chapter Outline

1. Gene Expression
2. Transcriptomics
3. RNA Sequencing Steps
4. RNA Sequencing Data Analysis

Learning Objectives

After completing this chapter, the student will be able to:

- describe the major categories of coding and noncoding RNA
- compare the use of microarrays and RNA-seq for measuring mRNA levels
- understand experimental and sequencing design
- explain different types of expression quantification
- understand differential gene expression

07 Metagenomics

Chapter Outline

1. Microbiome
2. Metataxonomics
3. Functional Metagenomics
4. Structural Metagenomics
5. Taxonomic Resources
6. Applications

Learning Objectives

After completing this chapter, the student will be able to:

- compare meta-omics methods
- explain metagenomic sequencing, assembly and classification

- understand the issues related to taxonomy resources
- describe metagenomics applications

08 Genome Data Visualization

Chapter Outline

1. BED File Format
2. UCSC Genome Browser
3. Ensembl Genome Browser

Learning Objectives

After completing this chapter, the student will be able to:

- describe BED file format

understand how to visualize, compare and analyze genomics data