SPECIAL TOPICS IN MODERN GENETICS

1. Genomics and bioinformatics

OUTLINE

1.1: Genome sequencing
1.2: DNA sequence analysis: Bioinformatics and genomic databases
1.3: Gene and protein functional analysis with genomic tools
1.4: The human genome project
1.5: The "-omics" revolution has created a new era in biological research
1.6: Comparative genomics analyzes and compares the genomes of different organisms

LEARNING OUTCOMES

After completing this chapter, the students will have gained knowledge on the following:

• Genomics combines DNA sequencing methods and recombinant DNA technology with bioinformatics, to determine a genome's DNA sequence and study its organization and function.

• Bioinformatics, combining computer science with biology and mathematics, develops software programs and databases that store, compare, analyze and make the sequences of proteins and nucleic acids easily accessible.

• Genome databases, in addition to the sequence of a genome, contain annotations about the location, the function and the regulation of genes.

• The human genome project has significantly increased our knowledge about the organization and function of the human genome, which held many surprises. For example, the number of human genes was lower than expected, while the similarity of the DNA sequence between different individuals and between humans and other species was higher than expected. Innovative emerging technologies now enable a futher in-depth understanding of the characteristics of the human genome.

• Genomics enables high-throughput studies in fields such as structural genomics, functional genomics, transcriptomics, proteomics and metagenomics, which have led to the "-omic" revolution in modern biology.

• Comparative genomics reveals the similarities and differences between the genomes of different organisms.

2. Applications and ethical issues of genetic engineering and biotechnology

OUTLINE

2.1 Production of pharmaceutical proteins in genetically modified organisms
2.2 Production of vaccines
2.3 Applications of transgenic animals in Biotechnology
2.4 Genetic engineering and genomic technologies in clinical diagnosis
2.5 Genetic analysis with DNA microarrays and expression microarrays
2.6 Genetic tests and ethical aspects
2.7 Patents and bioethics
2.8 Whole genome sequencing and bioethics

LEARNING OUTCOMES

After completing this chapter, the students will be able to understand:

- Applications of genetic engineering and biotechnology in the production of pharmaceutical proteins, vaccine development and in the diagnosis of genetic diseases and pathogens.
- Ethical issues arising from the applications of genetic engineering and biotechnology.

3. Forensic genetics and forensic science

OUTLINE

3.1 DNA Standardization methods

- DNA profiling based on VNTR
- DNA profiling based on autosomal STR
- DNA profiling based on Y chromosome STR
- DNA profiling based on mitochondrial profile
- DNA profiling based on SNP
- DNA barcode profiling applied in wild life conservation

3.2. Interpretation of DNA profiles

- Uniqueness of DNA profiles
- Prosecutor's fallacy
- DNA profiles databases
- Technical and ethical issues concerning DNA profiling

LEARNING OUTCOMES

After completing this chapter the students will be able to:

- Understand the process of defining a DNA fingerprint which is based on autosomal and X-linked STRS , and mitochondrial profile.
- Interpret the outcome of the DNA fingerprinting.
- They will also learn how DNA can be used in order to identify plants and animals endangered species

4. Genetically modified organisms/food

OUTLINE

4.1 Definition of genetically modified organisms

- Varieties resistant to herbicides
- Genetically modified plants resistant to insects
- Genetically modified plants directly consumed as food
- Genetically modified foods: safety, risks and public concerns

4.2 How to make a genetically modified organism

- Soya Roundup- Ready
- Golden Rice 2

4.3 Genetically modified food benefits and hazards

- Resistance to herbicides and insecticidal toxins
- Spread and potential risks of genetically modified organisms

LEARNING OUTCOMES

After completing this chapter the students will be able to understand :

- What exactly are genetically modified organisms (GMOs), why and how they are developed
- Gain an understanding of how biotechnology intersects with globalization, trade, poverty, food security, and environmental sustainability

5. Homologous DNA recombination and gene conversion

OUTLINE

5.1 Homologous DNA recombination

- Homologous recombination and Holliday structure
- Mitotic recombination and genetic mosaics
- Genetic conversion
- Genetic linkage based on the LOD score method

LEARNING OUTCOME

After completing this chapter the students will be able to understand :

- The details of homologous DNA recombination
- The formation of Holliday structure and heteroduplex DNA molecules
- The concepts of gene conversion and mitotic recombination
- Mapping human genes based on LOD score calculation

6. Genomics and personalized medicine

OUTLINE

6.1 Personalized medicine and pharmacogenomics
• Optimization of pharmaceutical interventions
• Reduction of adverse drug reactions - Examples
• Causes of adverse drug reactions
• Pharmacogenomics databases
6.2 Personalized medical and diagnostic tests of diseases
• Personalized medicine and cancer
• Personalized medicine and disease diagnosis
• Individual "legal" disease treatment profiles
6.3 Technical, social and ethical problems issues and challenges

LEARNING OUTCOMES

In the section on personalized medicine and pharmacogenomics, the student will understand the importance of studying genetically determined factors that influence the metabolism and action of drugs in humans, and the purpose of their improvement.

7. Epigenetics

OUTLINE

7.1 Epigenetic modifications of the genome
• DNA methylation
• Histone modification and chromatin remodeling
• Small RNA (miRNA) and Long Non-coding RNA (lncRNA)
7.2 Epigenetics and genome imprinting
• Genome imprinting disorders
• Assisted Reproductive Technologies & genome imprinting Disorders
7.3 Epigenetics and cancer

• Gene changes in cancer
7.4 Epigenetics and the environment
7.5 Analysis of the Epigenome

LEARNING OUTCOMES

- The course aims to understand the principles and concepts of epigenetics by the students and, more importantly, to differentiate new concepts from classical Genetics.
- Also, understanding the nature and ambiguity between genetic and epigenetic interactions (gene x environment) and their possible pharmacological regulation, is a further goal.
- The student will understand the cellular processes that lead to the appearance of altered or variable phenotype beyond the action of DNA and mutation.

8. Laboratory practicals

Laboratory exercises aim to deepen the understanding of the concepts and applications presented in the theory.

Specifically, the exercises focus on the following skills:
• DNA sequence assembly. Understand the concept that sequencing large sections of DNA, or even whole genomes, requires the assembly of smaller sequencing reads with overlapping ends. Understand the concept of genome coverage during sequencing. Calculate the probability of finding identical sequences in two DNA segments.
• OMIM database in NCBI. Browse entries related to sickle cell anemia and beta-thalassemia, with the aim of understanding the structure of the OMIM database, to find information about phenotypes, alleles, heredity, and relevant publications. Practice translating a DNA coding sequence into an amino acid sequence.
• Gene Search in Large Databases (NCBI / Ensembl) Developing experience of targeted processes that lead to the search, collection, identification and processing of information about identity, classification, structure, expression and functional characteristics of the genes under study. In addition, the comparison, the intersection of the information between the two databases (NCBI / Ensembl) and their connectivity, is also a learning objective for the development of experience of the above processes.
• Search for Proteins in Large Databases (NCBI / UniProtKB). Developing experience of targeted processes that lead to the search, collection, identification and processing of information about the identity, classification, structure, expression and functional characteristics of the translated sequences and proteins. More importantly, the links between various auxiliary large databases (SMART, PFAM, STRING, etc.) through the aforementioned, is also a learning goal for the development of experience of the above processes.
• Statistical interpretation of STR profiles, Evaluation and presentation of DNA evidence, Databases of DNA profiles, Kinship testing, Parentage testing, Forensic applications of mitochondria and Y chromosome, polymorphisms, Non - human DNA typing, Species identification.
• *Linkage* analysis between two loci using a family pedigree and the LOD score method.
• *Bioethical issues*