# Topics of lectures & Assigned Staff Members for

# Biochemistry & Genetics

## in the Foundation Block

## Academic Year 1430-1431 H (2009-2010 G)

## Biochemistry

| Lecture Title | Lecturer |
|---|---|
| Amino acids & protein structure | Dr. Usman Ghani |
| Structure & function of carbohydrate | Dr. M.Azhar |
| Lipid compounds of physiological importance | Dr. Amr Moustafa |
| Enzymes & coenzymes-1 | Dr. Usman Ghani |
| Enzymes & coenzymes-2 | Dr. Usman Ghani |
| Metabolism: anabolism & catabolism-1 | Dr. Amr Moustafa |
| Metabolism: anabolism & catabolism-2 | Dr. Amr Moustafa |
| Cell signaling & regulation of metabolism | Dr. M. Azhar |
| Introduction to Mol. Biology-1 | Dr. Sherif Saleh |
| Introduction to Mol. Biology-2 | Dr. Sherif Saleh |
| Molecular techniques | Dr. M.Azhar |
| Biochemical markers for diagnosis & follow up of diseases | Dr. Usman Ghani |

## Genetics

| Lecture Title | Lecturer |
|---|---|
| Human chromosomes: genotypes/phenotypes | Prof. M.Hazmi |
| Mode of inheritance | Prof. M.Hazmi |
| Mutations vs. polymorphism | Prof. M.Hazmi |
| Diagnosis of genetic diseases | Prof. M.Hazmi |
| Genetic Counseling | Prof. M.Hazmi |

# Amino acids & Protein Structure

## Objectives:

1. To introduce proteins as the most abundant functionally diverse molecules and their structure /function relationship.
2. To learn about the structure and properties of amino acids, the basic building units of proteins.
3. To achieve a comprehensive understanding of the complexity of protein molecules in terms of four levels of structural organization.
4. To create an understanding of the forces that stabilize a protein structure & understand protein denaturation.
5. To know about variation or mutations in amino acid sequence or protein misfolding that may lead to diseases.

## Outlines of the lecture:

1. Introduction to proteins and their structure-function relationship
2. Structure of amino acids
3. Classification of amino acids
   - neutral , basic & acidic amino acids
   - amino acids with non polar and polar side chains
   - define: pka , pI and buffers
4. **Optical properties of amino acids**
5. **Peptides and the peptide bond**
   - formation and naming,
   - characteristics of the peptide bond
6. **Structure of proteins and levels of organization:**
   - Primary structure
   - Secondary structure
     - α-helix: properties and causes of disruption
     - β-pleated sheets: types
     - Supersecondary structure and motifs
     - Collagen as an example of secondary structure
   - Tertiary structure and domains
   - Quaternary structure: subunits
7. **Forces stabilizing the protein structure**
8. **Protein denaturation:** definition and factors that cause denaturation
9. **Diseases of protein misfolding:** a brief list

# Structure and Function of Carbohydrates

## Objectives:

1. To understand the role of carbohydrates in providing and storing of energy for the body function.
2. The role of GAGs complex in various body functions

## Outline of the lecture:

1. Classification and structure of carbohydrates

   - Classification of carbohydrates
   - Isomers and epimers
   - Enantiomers
   - Cyclization of monosaccharides
   - Joining of monosaccharides
   - Complex carbohydrates

2. Glycosaminoglycans

   - Overview of glycosaminoglycans
   - Structure of glycosaminoglycans
   - Classification of glycosaminoglycans
     Examples like- chondroitin sulfate, keratan sulfate, hyaluronic acid and heparin

# Lipid Compounds of Physiological Importance

## Objectives:

1. To understand the biochemical and physiological properties of different lipid compounds
2. To understand the complex formation of phospholipids, glycolipids & lipoproteins
3. To understand the functional role of lipid compounds in physiological state and in relation to clinical problems.

## Outlines of the lecture:

1. **Fatty Acids**
   - Saturated Vs. Unsaturated (sis Vs. trans double bond)
   - Unsaturated: mono Vs. polyunsaturated
   - Polyunsaturated: ω3 Vs. ω6.
   - Eicosanoids: Prostanoids, Leukotrienes
   - Effect of Chain length and degree on saturation on physical & physiological properties of FA.
2. **Triacylglycerol**
   - Transfer in the circulation: VLDL & chylomicrons
   - Storage form in Adipose Tissue
3. **Phosholipids**
   - Phosphatidic acid & derived compounds
   - Sphingomyelins
4. **Glycolipids**
   - Cell surface carbohydrates
   - Ceramide & cerbrocides
   - Gangliosides especially GM1: intestinal receptor for cholera toxin, and their cellular accumulation in lipid storage disorders
5. **Steroids**
   - Cholesterol
   - Other cholesterol-derived steroids: Bile acids & hormones
6. **The structure & function of lipoproteins.**

# Enzymes & Coenzymes

## Objectives:

1. To introduce enzymes as biological protein catalyst which affects the rate of chemical reactions & to learn about their structure , properties & localization of the enzymes within the cell
2. To achieve a comprehensive understanding of mode of action of enzymes & factors affecting reaction velocity
3. To know Michelis-Menten kinetics
4. To study different types of enzyme activity inhibitors
5. To learn about regulation of enzyme activity
6. To be introduced to enzyme as tools for clinical diagnosis

# Enzymes & Coenzymes-1

## Outlines of the lecture

- **Introduction to enzymes:**
  - i. definition
  - ii. structure of enzymes ( coenzymes, cofactors)
  - iii. active site, induced fit model(enzyme –substrate interaction) & specificity
  - iv. definition of ribozymes , isozymes, zymogens
- **Classification of enzymes** (table)
- **Mode of action of enzyme** (referring to free energy of activation)
- **Factors affecting reaction velocity** ( curves )
- **Michelis -Menten equation** ( Km definition, conclusions)

# Enzymes & coenzymes-2

## Outlines of the lecture:

**1. Inhibition of enzyme activity:**

i. competitive inhibition
ii. non-competitive inhibition
iii uncompetitive inhibition
iv. application ( statins , insectisides, drugs as penicillin)

**2- Regulation of enzyme activity:**

i. allosteric enzymes
ii. covalent modification
iii. induction & repression

**3- Enzymes in clinical use:**

i. functional and non-functional enzyme
ii. Brief notes on enzymes used as biomarkers for diagnosis of diseases

# Metabolism: Anabolism and Catabolism

## Objectives:

1. To understand the concept of metabolism
2. To understand the concept of anabolism
3. To understand the concept of catabolism
4. To understand the meaning of a metabolic pathway
5. To understand the general outlines for regulation of a metabolic pathway
6. To understand the integration between anabolic and catabolic pathways
7. To identify the catabolic and anabolic pathways for glucose

## Outlines of lectures 1 & 2:

1. Metabolic pathway
2. Catabolic pathways
3. Anabolic pathways
4. Metabolic map
5. **Regulation of metabolism** (e.g., Allosteric, covalent modification, induction/repression)
6. **General outlines of metabolic pathways of glucose** (Glycolysis, Krebs cycle, HMP, Glycogenolysis, glycogenesis, and gluconeogenesis, interconversion of hexoses)
7. **Catabolic pathways of glucose** (e.g., Glycolysis, Krebs cycle)
8. **Anabolic pathways of glucose** (e.g., Glycogenesis)

# Cell Signaling & Regulation of Metabolism

## Objectives:

1. To understand the pathway of metabolism to coordinate the function of body.
2. To study the communication system between cells by chemical signaling to perform function.

## Outline of the lecture:

1. **Regulation of metabolism:**
   - Signal from within the cells
   - Communication between the cells

2. **Second messenger system:**
   - Role of adenylyl cyclase in intracellular signaling
   - Role of IP3 and diacylglycerol (DAG) in intercellular signaling.

3. **Mechanism of insulin action:**
   - Insulin receptor
   - Signal transduction

# Introduction to Molecular Biology
# (Nucleotides, DNA, RNA & DNA Language)

## Objectives of lectures 1 & 2:

1- To learn the language of molecular biology through being oriented about the structure of nucleotides which is the basic unit for DNA & RNA
2- To learn the structure & function of DNA & different types of RNA
3- To achieve a comprehensive understanding of the concepts of information transformation (DNA replication & gene expression)
4- To achieve appropriate preparation for more understanding of topics of Genetics especially concerned with mutations, polymorphism & diagnosis of genetic diseases.
5- To construct a good foundation for understanding of molecular techniques that are widely applied in clinical diagnosis & medical research such as PCR

# Introduction to Molecular Biology-1

## Outlines of the lecture:

1- Nucleotides:

- What are nucleotides
- Importance of nucleotides
- Structure of nucleotides
- Metabolism of nucleotides
  i. synthesis: sources of atoms of purine & pyrimidine rings (with reference to folic acid importance)
  ii. degradation: end products of purine & pyrimidine rings (with reference to hyperuricemia & gout)

2- DNA

- Importance of DNA
  i. Storage of genetic information
  ii. Transformation of genetic information to new cells (replication)
  iii. Transformation of information for protein biosynthesis
- Location of DNA in prokaryotes & eukaryotes
- Structure of DNA
  i. Structure of single strand of DNA
  ii. Structure of double stranded DNA (hydrogen bonds & denaturation)
  iii. Sequence of DNA (importance )
  iv. Linear & circular DNA (with reference to importance of plasmids in biotechnology)

3-RNA

- Structure of RNA
- Types of RNA (mRNA, tRNA & rRNA)
- Importance (functions) of each the three types of RNA
- What is meant by gene expression i.e. transcription + translation

# Introduction to Molecular Biology-2

## Outlines of the lecture:

### 1-Replication:

- Concept of semiconservative nature of replication
- Origins of replication in prokaryotes & eukaryotes
- Steps of replication in prokayotes (*E.coli*) with reference to main enzymes involved
- Eukaryotic replications (main differences from prokaryotic replication)

### 2-Gene expression:

- **Concepts of gene expression**
- **Transcription**
    - i. Definition
    - ii. Steps of transcription in prokaryotes
        - Initiation
        - Elongation
        - Termination-
    - iii. Eukaryotic transcription
        - Eukaryotic RNA polymerases
        - Promoters of RNA polymerase II
        - Role of transcription factors & enhancers in eukaryotic transcription
        - Posttranscriptional modification of mRNA (capping. polyA-tail & splicing)
- **Concepts of protein biosynthesis (translation)**
    - i. Genetic code
    - ii. Summary of steps of translation (initiation, elongation & termination)

# Molecular Techniques

## Objectives:

1. To identify molecular techniques that are currently in clinical practice
2. To understand the principle, types and medical applications of polymerase reaction (PCR).
3. To understand the use of restriction endonucleases for the production of recombinant DNA and recombinant proteins
4. To understand the principle of blotting techniques and their medical applications

## Outlines of the lectures:

1. Molecular techniques: An overview
2. PCR
3. Restriction endonucleases, recombinant DNA and recombinant proteins
4. Blotting techniques: Southern, northern, western techniques
5. The use of Southern blotting for the detection of mutations

# Biochemical Markers for Diagnosis and Follow Up of Diseases

## Objectives:

1. To develop an understanding of the importance of different biochemical markers in the diagnosis and follow up of a disease.
2. To learn about enzymatic markers used in the diagnosis of diseases such as pancreatic and liver disease.
3. To learn about plasma proteins and tumor markers used in the diagnosis of various diseases including cancer

## Outlines of the lecture:

1. Introduction to markers, enzymatic and non-enzymatic diagnosis and prognosis of diseases
2. Enzymes as markers of diseases: amylase, alanine aminotransferase (ALT), aspartate aminotransferase (AST)
3. Plasma proteins as markers of diseases: albumin
4. Tumor markers: α-fetoprotein, prostate specific antigen (PSA)

# GENETCS: Lecture 1
# Human Chromosomes, Genotypes/Phenotypes

## Objectives:

1. To know the normal structure and number of human chromosomes (Cytogenetics)
2. To be introduced briefly to various methods of human chromosomes staining.
3. To understand the Condensation/decondensation, and Packing of chromosome.
4. To know the definitions of genotype and phenotype.
5. To understand the relation between genotype & phenotype
6. To know various patterns of inheritance
7. To understand the methods of regulation of gene expression.
8. To differentiate between gene expressivity, and gene penetrance.
9. To be familiar with genetic heterogeneity with clinical examples.
10. To know how new mutation can explain the new appearance of AD disease.
11. To know how the genes with codominant are alleles expressed.
12. To be introduced to Pseudodominance inheritance

## Outlines of the lecture:

1- **Genetic material in living cells**
2- **The human genome project**
3- **Chromosomes**
   - i. The human chromosomes
   - ii. Chromosome structure
   - iii. Classification of chromosomes
   - iv. Staining methods for the genetic analysis of chromosomes
   - v. Condensation-decondensation of chromosomes
   - vi. DNA packaging in chromosomes

4- **Genotype-phenotype correletation**
   - i. Genotype
   - ii. Phenotype
   - iii. Genotype-phenotype relation

5- Gene expressivity
6- Gene penetrance
7- Gene heterogeneity
8- New mutations
9- Codominance
10- ABO blood group system as an example of codominance
11- Pseudodominance

# GENETCS: Lecture 2
# Mode of Inheritance

## Objectives

1. To know various patterns of inheritance
2. To know the classification of autosomal inheritance, with clinical examples
3. To be able to use the Punnet square
4. To understand the sex-linked mode of inheritance, with clinical examples
5. To understand the nature of inheritance of mitochondrial genetic disorders

## Outlines of the lecture:

1- Single gene disorders

2- Mode of inheritance of single gene disorders

3- Autosomal inheritance

4- **Autosomal dominant inheritance (with examples)**
   - i. Marfan syndrome
   - ii. Acute intermittent porphyria
   - iii. Punnet square for autosomal dominant inheritance

5- **Autosomal recessive inheritance (with example)**
   - i. cystic fibrosis

6- **Sex-linked inheritance**
   - A- Y-linked inheritance
   - B- X-linked inheritance
     - 1- X-linked recessive disorders (with examples)
       - ii. Lesch-Nyhan syndrome
       - iii. Hemophilias
       - iv. G 6-P D deficiency
     - 2- X-linked dominant disorders (with examples)
       - i. X-linked hypophosphatemic rickets (vitamin D resistant rickets)
       - ii. Incontinentia pigmenti (IP)

7- **Mitochondrial disorders**

8- **Basic decision tree for determining the mode of inheritance in a pedigree**

# GENETCS: Lecture 3
# Mutations Vs. polymorphism

## Objectives:

1. To know the definition of Mutations & their consequences.
2. To understand the 3 Classes/Levels of mutations (at the genome level, at the chromosome level, at the gene level)
3. To understand the Types of mutations
4. To know the classification of genetic diseases (with examples on each category).
5. To know the definition of Polymorphisms & their consequences
6. To be able to differentiate between various types of polymorphisms based upon the method of detection
7. To understand the uses of polymorphisms

## Outlines of the lecture:

A- Mutation
- Definition
- Types (spontaneous vs. mutagen-induced, deleterious vs. non deleterious, nucleotide substitution vs. deletions/insertions)
- Levels of mutations (Genome, chromosome, or gene)
- Genetic diseases:

1- Single gene disorders (with examples)
2- Chromosomal disorders (numerical vs. structural, with examples)
3- Multifactorial disorders (with examples)
4- Mitochondrial disorders
5- Acquired somatic genetic diseases (with example)

**B- Polymorphism:**
- Definition
- Types of polymorphisms (based on the method of detection)
    1- DNA polymorphism
    2- Protein polymorphism
    3- Altered physical features
    4- Chromosome heteromorphisms
- Uses of polymorphism
- Areas of significance of polymorphism
- Clinical importance of polymorphism:
    1- in forensic medicine
    2- as genetic markers

# Genetics: Lecture 4
# Diagnosis of Genetic Diseases

## Objectives

1- To present a brief description of methods to diagnose genetic diseases.
2- To introduce the main tools of Molecular Biology and Recombinant DNA technology used in diagnosis & follow up genetic diseases.

## Outlines of the lecture:

1- Overview for diagnosis of genetic diseases

2- Family history

3- Clinical presentation

4- Recombinant DNA technology

i. Applications of DNA technology
ii. Genetic Engineering
iii. Requirements for DNA technology
    - Restriction endonuclease
    - Sources of DNA & cDNA synthesis
    - Vectors
    - Probes
iv. Principles molecular cloning

5- **Polymerase chain reaction (PCR) & its applications**

# Genetics: lecture5
# Genetic Counseling

## Objectives:

1. To understand the principle steps of genetic counseling.
2. To know the ethical principles in performing genetic counseling.
3. To understand unique features of genetic counseling in Arabic/Islamic communities.
4. To know the sequence of steps for pre-marital screening.

## Outlines of the lecture:

1- Genetic counseling for Mendelian disorders

2- Essential components of genetic counseling

3- Ethical principles

4- Genetic counseling in achondroplasia (as an example)

5- Establishment of Mendelian inheritance

6- Premarital screening