UIVERSITY OF JORDAN / FACULTY OF MEDICINE
HUMAN GENETIC COURSE FOR 2nd YEAR MEDICAL STUDENTS
COURSE OUTLINE (0561214)
SUMMER COURSE 2013-2014

Sunday - Thursday / Lecture theater: 202 (6) M. El-Khateeb

Introduction *Chapter 1 (Ref 2)*

- Early beginnings
- Mendel and laws of inheritance
- The chromosomal basis of inheritance
- The origin of medical genetics
- Classification of genetic diseases
- Definitions
- The impact of genetic diseases
- Major new developments

GENETIC Variation, POLYMORPHISM, AND MUTATION *Chapter 3 (ref1)*

- Genetic Variations
  - Terminology
- Cause of Genetic Variations
  - Evolution
  - Gene Flow and Drift
  - Gene Frequency
  - Adaptation
  - Natural Selection
- Mutation
  - Genome level
  - Chromosomal Level
  - Gene Level
- Genetic Diversity Among Individuals
- Inherited Variation and Polymorphism at the DNA Level
- The Molecular Basis of Mutations and Their Detection

Chromosomal basis of Hereditary *Chapter 3 and 18 (ref2)*

Chromosomes and Cell division

- Human Chromosome
- Methods of chromosomal analysis
- Molecular cytogenetics
- Chromosomal Nomenclature
- Cell division
- Gametogenesis
- Chromosomal abnormalities

Chromosomal Disorders

- Incidence of Chromosomal abnormalities
- Disorders of the autosomes
- Disorders of the sex chromosomes
- Disorders of sexual differentiation
- Chromosomal Breakage syndromes
- Indications for chromosomal analysis

Patterns of inheritance *Chapter 7 and 19(ref 2)*

- Mendelian laws
- Family studies and pedigree drawing
- Terminology
- Autosomal Dominant inheritance
  - Pleotropy
  - Reduced penetrance
  - Codominant
  - New Mutations
  - Homozygosity for autosomal traits
- Autosomal Recessive inheritance
  - Consanguinity
  - Pseudodominance
  - Locus heterogeneity
  - Mutational heterogeneity
- Sex Linked inheritance
  - X- linked dominant
  - X- linked recessive inheritance
  - Variable expression of heterozygous in females
  - Homozygosity for X-linked disorders
  - Skewed X-inactivation
- Y-Linked Inheritance
- Partial sex linkage
- Establishing the mode of inheritance
  - Autosomal Dominant inheritance
  - Autosomal Recessive inheritance
  - Sex Linked inheritance
- Multiple alleles
- Anticipation
- Mosaicism
- Uniparental Disomy
- Genomic Imprinting
  - Prader-Willi Syndrome
  - Angelman Syndrome
- Mitochonderial Inheritance
- Single gene inheritance
  - Hemoglobimop{athies
  - Cystic fibrosis
  - Huntigton disease
  - Myotonic Dystrophy

- Duchane Muscular Dystrophy
- Neurofibromatosis
- Hemophilia

Biochemical Genetics — *Chapter 11(Ref 2)*

- Inborn errors of metabolism: Garrod and alkaptnuria
- One gene: one protein / one gene: one polypeptide hypothesis
- Disorders of amino acids metabolism
- Urea Cycle Disorders
- Disorders of Carbon hydrate metabolism
- Disorders of Steroid metabolism
- Disorders of lipid metabolism
- Disorders of amino acids metabolism
- Organic Acids disorders
- Pharmacogenetics

Multifactorial and population genetics: — *Chapter 9 (ref 2)*

- Principles of Multifactorial Inheritance
- Polygenic inheritance and normal distribution
- Multifactorial Inheritance, liability and threshold model
- Identifying genes which causes multfactorial disorders
- Disease Model of Multfactorial inheritance
  - Cleft Lip and cleft Palate
  - Diabetes
  - Hypertension
  - Coronary heart Disease
- Human Populations
- Phenotypes, Genotypes, and Gene Frequencies
- The Hardy-Weinberg Law Factors
- Affecting Hardy-Weinberg Equilibrium
- Measurement of Human Mutation Rates

Genetics and Cancer: — *Chapter 14*

- Inheritance of susceptibility to some forms of cancer e.g.
- Retinoblastoma
- Chromosome breakage syndromes
- Chromosome abnormalities in cancer
- Causes of Cancer
- Cancer Genes
- Major Classes of Cancer Genes
- Identification of Inherited Cancer Genes
- Molecular Basis of Cancer

Prevention and Treatment of Genetic Disease 20,21,23 (ref 2)

- Criteria for genetic screening
- Carrier testing for autosomal recessive
- Presympotomatic diagnosis of autosomal Dominant Disorders
- Neonatal screeinig
- Prenatal diagnosis
  - Techniques used in prenatal diagnosis
  - Indications for prenatal diagnosis
  - Penatal treatment
- Preimplantation genetics
- Genetic counseling
- Treatment of genetic diseases
  - Conventional approaches
  - Protein/ Enzyme replacement
  - Drug Treatment
  - Tissue removal
  - Recombinant DNA
- Gene therapy
- Transplantation and Stem cell therapy

REFERNCES

1. **MEDICAL GENETICS**
   Jorde, Carey, Bamshad, White
   Published by Mosby

2. **ELEMENTS OF MEDICAL GENETICS**
   Robert Muller and Ian Young
   Published by Churchill Livingstone

3. **ESSENTIAL MEDICAL GENETICS**
   Connor, Ferguson-Smith
   Published Blackwell Science