

## Ph.D Biomedical Genetics – Course Syllabus, 2011

*Objectives and Goals:* The purpose of this core course is to introduce students to human genetics, the role of genetics in human diseases, methods to detect disease susceptibility genes, the ethics of genetic testing and gene therapy for genetic diseases.

*Background:* Because of the Human Genome Project, genetics is now playing a significant role in the investigation into the pathogenesis of diseases. Our students need to understand the importance of human genetics in research. This course will give an overview of the subject so they will be aware and knowledgeable about the various aspects involved in human genetic research.

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**Prerequisites:** Biochemistry and biochemistry lab

**Textbook:** *Introduction to Genetic Analysis 9<sup>th</sup> edition.* Anthony Griffiths, Susan Wessler, Richard Lewontin, Sean Carroll, 2008. W.H. Freeman and Company, Inc., New York.

**Other course material:** Some faculty members will post their lecture material on Blackboard and will send an announcement to the students prior to class concerning their expectations for the material.

Students will be assigned to read the various chapters in the text **prior to class**, in order to be familiar enough with the material **to participate in discussions led by the instructors**. The instructors can also give additional reviews, papers or handouts prior to the class to facilitate class participation by the students. Critical thinking about the role of genetics in human disease research will be emphasized and the students will be encouraged to discuss both the advantages and disadvantages of genetic research as it relates to human disease.

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### Course content, Schedule and Assignments:

| DATE | TOPIC  | INSTRUCTOR |
|------|--|------------|
|      | Introduction to course -<br>chromosome structure & function                                    | Newman     |
|      | Genes in pedigrees & populations<br>Monogenic/multifactorial inheritance<br>Mendelian genetics | Lyn        |
|      | DNA Mutation & repair  | Powell     |
|      | Single Nucleotide Polymorphisms<br>Human Genome Project<br>Gene Mapping                        | Song       |

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|--|--------------------------|--------|
|  | Genetic Testing & Ethics | Newman |
|  | Genetic Therapies        | Newman |
|  | <b>Oral Exam</b>         |        |

### Assignments:

- I. Chromosome Structure and Function - Gale Newman
- II. Genes in pedigrees and populations - Deborah Lyn
  - A. Monogenic versus multifactorial inheritance
  - B. Mendelian genetics
  - C. Complications of Mendelian pedigree patterns
  - D. Polygenic-threshold theory
  - E. Gene frequencies
- III. The Human Genome Project and Model Organisms - Gale Newman
  - A. The Human Genome Project
  - B. Gene mapping
  - C. Model organisms genome mapping
  - D. Relationships between the human genome and other genomes
  - E. Organization of the human genome
- IV. DNA mutation and repair
  - A. Mutations – Mike Powell
  - B. Polymorphisms – Qing Song
  - C. DNA repair – M. Powell
- V. Genetic mapping and identification of disease and susceptibility genes - Jonathan Stiles
  - A. Genetic markers - J. Stiles
  - B. Identification of human disease genes - J. Stiles
    1. Genetic mapping
    2. Admixture mapping
  - C. Identification of human susceptibility genes - Leonard Anderson
    1. Linkage analysis of complex characters
    2. Association studies and linkage disequilibrium
    3. Haplotyping
- VI. Admixture (Handouts) - G. Newman
- VII. Epigenetics - G. Newman
- VIII. Cancer genetics - Gary Sanford
- IX. Genetic testing - G. Newman
- X.

## **Student Evaluation Methods, lecture:**

Students will be given a letter grade based on the following:

Written exam - 50% - A short answer exam worth 100 points will be given. Questions from the instructors teaching during this period will be given to the course director. Grading of the questions will be done by the instructor submitting the question.

Oral exam - 40% - oral exams will be given at the end of the course. Each student is given a 30 minute session with two or more instructors. The instructors for all sections will submit 5 questions prior to the exam and submit them to the course director. The instructors and course director will meet and discuss the appropriateness of each of the questions before using them for the exam. Grading of the oral exam will be on the basis of accuracy of the answers as well as the depth and breadth of the student's knowledge of the subject. Immediately after the oral exam, the participating instructors will determine the grades for each student.

### **Instructors:**

|                                     |        |         |
|-------------------------------------|--------|---------|
| Gale Newman, Ph.D., Course director | X 1636 | HG 325  |
| Deborah Lyn, Ph.D.                  | X1521  | MEB 215 |
| Mike Powell, Ph.D.                  | X1582  | RW 336  |
| Qing Song, M.D., Ph.D.              | X1845  | RW 2    |
| Jonathan Stiles, Ph.D.              | X1585  | HG 349D |
| Leonard Anderson, Ph.D.             | X8920  | RW 2    |
| Gary Sanford, Ph.D.                 | X1504  | HG 309C |