



PM 532: Genetics in Public Health and Preventive Medicine

**UNIVERSITY OF SOUTHERN CALIFORNIA
DEPARTMENT OF PREVENTIVE MEDICINE**

SUMMER 2014

4 Units

Thursdays 10:00-2:00, SSB 115

COURSE COORDINATOR:

Mariana Stern, PhD

Associate Professor of Preventive Medicine

Virtual office hours 24/7

e-mail: marianas@usc.edu

phone: 323-865-0811

office: NOR 5421A

For office visits, please call or email in advance to make an appointment. I will be available to meet after class every week at Soto building.

COURSE DESCRIPTION:

This course will focus on basic concepts in areas such as classic Mendelian genetics, population genetics, molecular genetics, and their applications to public health and preventive medicine. The final goal is to prepare students with necessary knowledge in human genetics before they take more advanced courses in topics such as molecular and genetic epidemiology and statistical genetics, and to expose students to emerging and complex issues related to the integration of genetics in public health policy and practice.

COURSE OBJECTIVES:

By the end of the course students should be able to:

1. Describe the normal structure and function of nucleic acids and chromosomes;
2. Have knowledge on methods used in the study of genetic variability in human population;
3. Discuss and apply basic concepts in population genetics,
4. Describe the genetics and epigenetics of cancer and non-cancer diseases,
5. Communicate effectively using the correct terminology commonly used in human genetic and genetic epidemiology literature,
6. Discuss the relevant issues regarding the application of genetics in public health policy, practice, and preventive medicine.

PREREQUISITIES:

Graduate student status

TEXTBOOKS:

Highly Recommended Medical Genetics

L.B. Jorde, J.C. Carey, M.J. Bamshad
Fourth Edition, 2010. Mosby, Elsevier. ISBN 13: 978-0-323-05373

Recommended:

An Introduction to Human Molecular Genetics

J.J. Pasternak
Second Edition, 2005. Wiley-Liss. ISBN: 0-471-47426-6

Genetics and Public Health in the 21st Century

M.J. Khoury, W. Burke, E.J. Thompson (Eds). Oxford University Press, 2000. ISBN-13: 978-0195128307

COURSE STRUCTURE:

This course will use USC Blackboard (BB) as the main portal for communication between the instructor and students. BB is available at <https://blackboard.usc.edu/>

The syllabus, announcements, lecture slides in pdf format, online homework assignments, and grades will be posted on BB. Please make sure you know how to log into your BB account, and navigate the BB site. ***If you have problems with BB, please contact the BB help line ASAP.***

LECTURES: will cover the assigned textbook chapters, and *will include additional material not covered in the textbook*, such as additional examples and topics for in class discussion. Students are responsible for all topics covered in the textbook and for all the topics discussed during the lectures, even if they are not covered in the textbook. All lectures will be posted in BB after each class. At the discretion of the instructor, some lecture materials might be posted on BB right before the class starts. Therefore, you should come prepared to class to take notes, with or without lecture notes.

READINGS: You are required to study the assigned chapter readings as stated in the class calendar and as indicated by the instructor on the previous lecture, BEFORE class so you can participate in a meaningful discussion about the topic covered that day, and to answer the questions posed in the slides and during the lecture.

CLASS PARTICIPATION: 10%

- Class attendance and participation is expected from all students. This requires that you go over all the suggested materials, as indicated in the course schedule, AHEAD of time so that you can be prepared to participate in discussions.
- Hypothetical scenarios and clinical cases will be proposed in some of the lectures, you are expected to engage in discussions and participate in in-class group discussions. There will be no right or wrong answers. The focus will be on active participation.

HOMEWORK ASSIGNMENTS: 40%

- There are ten (10) online homework assignments. Each homework is worth 10 points.
- Homework assignments will be visible/accessible on Blackboard, under the "Assignments" tab each Friday after class, and will cover the materials covered during the previous lecture.

Homework assignments will be due by 10 AM the following Thursday. All homeworks will be online quizzes to be completed in BB. Therefore, they will remain visible/accessible in BB until 10 AM on the due date. Once the homework is taken off the site, it **CANNOT** be accessed again, therefore, make sure you do the homework in the allotted time. **MANUAL SUBMISSION OF HOMEWORK ASSIGNMENTS WILL NOT BE ACCEPTED.**

- The purpose of the online homework assignments is to provide you with a mechanism to review class materials and identify areas that need reviewing or further discussions with the Instructor. They should be completed individually.

FINAL EXAM: 30%

- There will be one final exam that will cover the entire course. **ATTENDANCE ON ALL EXAMS IS MANDATORY.** Absence from any of the exams will be considered ONLY if there is a documented emergency, such as a doctor's note for serious illness or a death in the immediate family. Any conflict with the final exam should be notified to the instructor at least two weeks before the final exam.

FINAL PROJECT: 20%

- The final project will consist of doing research on a genetic disease/disorder of the student's choice. The Instructor will provide a list of genetic diseases/disorders from which the student can choose. A template with guided questions will be provided by the Instructor to complete the assignment, which should be done individually.

GRADING SCALE:

Class participation	10%
Homework	30%
Final project	20%
Final Exam	40%

A total of 250 points can be earned in the course. Final grades will be determined on the percentage of points earned. The standard grading scale to be used will be (in % of total points):

A	92-100%	B-	80-82	D+	64-67
A-	89-91	C+	76-79	D	60-63
B+	87-88	C	72-75	D-	56-59
B	83-86	C-	68-71	F	<55

The instructor reserve the right to slide the scale downward as appropriate for class performance, but the scale will not be moved upward to increase the rigor. For example, a student receiving a score of 89% will be guaranteed an A- or better, if receiving 72% a C or better, and if receiving a 60% a D or better.

STUDENTS WITH DISABILITIES

Any student requesting academic accommodations based on a disability is required to register with Disability Services and Programs (DSP) each semester. A letter of verification for approved accommodations can be obtained from DSP. Please be certain the letter is delivered to the professor as early in the semester as possible. DSP is located on the University Park campus in STU 301 and is open 8:30 a.m. – 5:00 p.m., Monday through Friday. The phone number is (213) 740-0776.

COURSE CALENDAR:

Week	Date	Lecture Topics	Readings	Homework Assigned	Homework Due
1	5/22	Lecture 1. Introduction to public health genetics. Introduction to chromosome structure and functions Relevance and implications of genetics in public health studies and policy. DNA organization, structure and function, cell division cycle, mitosis and meiosis, chromosomes, recombination, clinical cytogenetics, chromosomal diseases.	Jorde: 2&6 Khoury: 1&2 (Pasternak: 2)		
2	5/29	Lecture 2. Gene structure and function Chemical structure of nucleic acids, DNA replication, DNA transcription, RNA translation, post-translational processing, genetic code, gene structure. Human Genome Project. Discussion of cystic fibrosis as an example of a monogenic disease due to a single mutation. Screening for cystic fibrosis as a paradigm for public health policy development. Documentary: "Expanding the Horizon of Hope: 50 years of Progress". Cystic Fibrosis Foundation	Jorde: 2 (Pasternak: 4)	HW #1	
3	6/5	Lecture 3. Mutations and genetic variability Types of mutations, causes of mutations, detection and measurement of genetic variation, mutation databases. OMIM database. Polymorphisms, techniques for polymorphisms analyses, databases for polymorphism analyses. Genetic ancestry.	Jorde: 3	HW#2	HW# 1
4	6/12	Lecture 4. Basic concepts in classic and population genetics Mendel's Laws, modes of inheritance, gene and genotype frequencies, Hardy-Weinberg equilibrium.	Jorde: 4 (Pasternak: 3)	HW#3	HW#2

Week	Date	Lecture Topics	Readings	Homework Assigned	Homework Due
5	6/19	Lecture 5. Identification of disease genes in the population Gene mapping, linkage analyses, linkage disequilibrium, association studies, physical mapping, positional cloning, molecular diagnosis. <i>Readings: Chapter 8 Jorde, Chapter 3 Pasternak</i>	Jorde: 8 (Pasternak: 3)	HW#4	HW#3
6	6/26	Lecture 6. Biochemical genetics Metabolic diseases. PKU as an example of metabolic disease. PKU newborn screening as paradigm for public health interventions in genetic diseases. Xenobiotic metabolism and its implications for cancer risk and pharmacogenetics. Film: "Extraordinary measures"	Jorde: 7 Khoury: 20	HW#5	HW#4
7	7/3	Lecture 7. Cancer genetics & Epigenetics Basic concepts of cancer biology. Cancer genes (oncogenes and tumor suppressor genes). Cancer predisposition genes. Basic concepts of epigenetics. Epigenetic alterations and cancer development and cancer therapy	Jorde: 11 Khoury: 9 (Pasternak: 16)	HW#6	HW#5
8	7/10	Lecture 8. Genetic Testing I: prenatal and newborn screening Prenatal diagnosis, clinical applications of genetic tests. Newborn screening, public health surveillance.	Jorde: 13-14 Khoury: 7 (Pasternak: 16)	HW#7	HW#6
9	7/17	Lecture 9. Genetic Testing II: carrier screening Diagnosis of carrier mutations, Thalassemia, Tay-Sachs. Documentary: "Confronting the killer gene"	Jorde: 13-14 Khoury: 7 (Pasternak: 16)	HW#8	HW#7

Week	Date	Lecture Topics	Readings	Homework Assigned	Homework Due
10	7/24	Lecture 10. Genetic Testing III: Pre-symptomatic genetic diagnosis. Gene Therapy. Diagnosis of cancer genes. Breast and ovarian cancer susceptibility. Previvors. Gene replacement therapies, gene blocking therapies Documentary: "In the family"	Jorde: 13-14 Khoury: 7 (Pasternak: 16)	HW#9	HW#8
11	7/31	Lecture 11. Genetic counseling for pre-symptomatic diseases in adults. Guest lecture by Charite Ricker, Genetic Counselor. Introduction to genetic counseling. Application of genetic counseling for the diagnosis of individuals at higher risk of developing cancer. Ethical and legal Issues in public health genetics.	Khoury: 25-27	HW#10	HW#9
12	8/7	FINAL EXAM.			