Human Genetics 2090
Genetics of Complex Diseases I
Spring Term 2020; Tuesday and Thursday, 2:30-3:55 PM
2 credits, 10 weeks
A215 Crabtree Hall

Instructor: M. Ilyas Kamboh, PhD
3117 Public Health
412-624-3021
kamboh@pitt.edu
Office Hours: by appointment

Course Description:
This course provides students with an overview of the molecular and biochemical genetic approaches to determine the underlying genetic architecture of common diseases that account for a large portion of the public health burden of disease. The genetic, environmental and epigenetic factors that influence susceptibility to common disease will be illustrated using selected examples, such as cardiovascular disease, neurodegenerative diseases, and mental health diseases.

This course provides students with an understanding of the biochemical and molecular genetic approaches to understand genetically determined susceptibility to common diseases. This will be presented using selected examples of complex human diseases (cardiovascular disease, neurodegenerative diseases, and mental health diseases). Risk of common, complex diseases is determined by genotypes at multiple genetic loci and a complex interaction of genetic variation and environmental exposures. Risk of almost every common disease is influenced by genes, but the relationship between genotype and disease phenotype is weak compared to that observed with rare Mendelian traits. However, understanding the contribution of genes to common disease susceptibility is important to public health.

Course Objectives:
At the end of this course the student will be able to:

• Describe the epidemiology and biochemical and molecular bases of selected common diseases

• Describe the underlying genetic architecture and environmental risk factors that influence genetic susceptibility to a variety of common, complex diseases ranging from cardiovascular disease to neurological disorders

• Describe and discuss the public health impact of a variety of common, complex diseases, including potential pharmacogenomics applications

Purpose of Course:
The goal of this course is to provide students with an understanding of the molecular and biochemical genetic approaches to understanding genetically determined susceptibility to common diseases. This goal will be achieved by using selected examples of complex human diseases (cardiovascular disease, neurodegenerative diseases, and mental health diseases).

Risk of common and complex diseases is determined by the genotypes at multiple genetic loci and their complex interaction with environmental exposures. Understanding the contribution of genetic factors to common disease susceptibility is important to public health because these diseases account for a large fraction of morbidity and mortality in the general population.

This course will also cover the importance of pharmacogenomics and epigenetics to show how genetic differences can affect individual's response to drugs (pharmacogenetics) and how changes in gene expression can occur without changes in the DNA sequence due to DNA methylation, histone modification or RNA interference (epigenetics) as these factors can also play a role in the development of common diseases.

Text Book:
There is no text book for this course. Handouts are used extensively.
All lecture slides and required readings are available on Courseweb.

Grading:
Grades will be assigned based on mid-term exam (50%), final exam (50%). The following will be the grade scale:

90-100%	A
80-89%	B
70-79%	C
60-69%	D
<60%	F

Behavior/Ground Rules
All students are expected to behave professionally in the class (i.e. listen to the lectures attentively and participate in discussion; no use of cell phones; laptops are allowed only related to the classroom teaching activities).

Academic Integrity:
All students are expected to adhere to the school's standards of academic honesty. Cheating/plagiarism will not be tolerated. The Graduate School of Public Health's policy on academic integrity, which is based on the University policy, is available online in the Pitt Public Health Academic Handbook www.publichealth.pitt.edu/home/academics/academic-requirements. The policy includes obligations for faculty and students, procedures for adjudicating violations, and other critical information. Please take the time to read this policy.

Disabilities:
If you have a disability for which you are or may be requesting an accommodation, you are encouraged to contact both your instructor and Disability Resources and Services, 140 William Pitt Union, 412-648-7890 or 412-383-7355 (TTY) as early as possible in the term.

Diversity Statement:
The University of Pittsburgh Graduate School of Public Health considers the diversity of its students, faculty, and staff to be a strength and critical to its educational mission. Pitt Public Health is committed to creating and fostering inclusive learning environments that value human dignity and equity. Every member of our community is expected to be respectful of the individual perspectives, experiences,

behaviors, worldviews, and backgrounds of others. While intellectual disagreement may be constructive, no derogatory statements, or demeaning or discriminatory behavior will be permitted.
If you feel uncomfortable or would like to discuss a situation, please contact any of the following:

- the course instructor;
- the Pitt Public Health Associate Dean for Diversity at 412-624-3506 or nam137@pitt.edu;
- the University's Office of Diversity and Inclusion at 412-648-7860 or

https://www.diversity.pitt.edu/make-report/report-form (anonymous reporting form).

Sexual Misconduct, Required Reporting and Title IX Statement
The University is committed to combatting sexual misconduct. As a result, you should know that University faculty and staff members are required to report any instances of sexual misconduct, including harassment and sexual violence, to the University's Title IX office so that the victim may be provided appropriate resources and support options. What this means is that as your professor, I am required to report any incidents of sexual misconduct that are directly reported to me, or of which I am somehow made aware.

There are two important exceptions to this requirement about which you should be aware:
A list of the designated University employees who, as counselors and medical professionals, do not have this reporting responsibility and can maintain confidentiality, can be found here:
www.titleix.pitt.edu/report/confidentiality

An important exception to the reporting requirement exists for academic work. Disclosures about sexual misconduct that are shared as part of an academic project, classroom discussion, or course assignment, are not required to be disclosed to the University's Title IX office.

If you are the victim of sexual misconduct, Pitt encourages you to reach out to these resources:
• Title IX Office: 412-648-7860
• SHARE @ the University Counseling Center: 412-648-7930 (8:30 A.M. TO 5 P.M. M-F) and 412-648-7856 (AFTER BUSINESS HOURS)

If you have a safety concern, please contact the University of Pittsburgh Police, 412-624-2121.
Other reporting information is available here: www.titleix.pitt.edu/report-0

Statement from the Department of Gender, Sexuality, and Women's Studies
[This statement was developed by Katie Pope, Title IX Coordinator, in conjunction with GSWS instructors.]

Class Schedule:

Date	**Topic**
1/7	Introduction – Genetic basis of common diseases
1/9	Genetic basis of common diseases
1/14	Coronary artery disease (CAD) – Epidemiology, genetic evidence, heritability of risk factors, atherosclerosis process

1/16	Role of lipid metabolism and lipid genes in CAD. Genome-wide association studies (GWAS) of CAD
1/21	Familial Hypercholesterolemia (FH) - Clinical features of heterozygous and homozygous FH, population distribution of heterozygous FH, cholesterol homeostasis
1/23	Role of the LDL-receptor (LDL-R) gene in FH, structure of the LDL-R protein, classification of LDL-R mutations. A brief description of other monogenic disorders that affect plasma LDL cholesterol and the risk of CAD
1/28	Apolipoprotein E (APOE) in lipid metabolism and risk of CAD
1/30	Genetics of APOE in affecting plasma cholesterol levels and the risk of CAD, APOE and Type III hyperlipoproteinemia
2/4	**Mid-Term Exam**
2/6	Alzheimer's disease – types of dementia, Epidemiology and risk factors
2/11	Alzheimer's disease – clinical and pathological features, Diagnostic biomarkers,
2/13	Alzheimer's disease – Genetics and Possible treatments
2/18	Parkinson's Disease
2/20	Introduction to Epigenetics: Etymology, definitions and molecular basis of epigenetics (Dr. Koldamova)
2/25	Mechanisms of Epigenetics: DNA methylation and chromatin remodeling, RNA interference and microRNA (Dr. Lefterov)
2/27	Pharmacogenetics and Pharmacogenomics
3/3	Schizophrenia– Epidemiology, risk factors and biological mechanisms, Genetics (Dr. Nimgoankar)
3/5	Autism spectrum disorders (Dr. Padiath)
3/10 & 3/12	**Spring Break – No Classes**
3/17	Multiple Sclerosis (Dr. Padiath)
3/19	**Final Exam**