Human Genetics

16:681:535 Fall 2017
Course Information and Policies

Class Time: Monday, Wednesday 3:20-4:40 **Location:** Nelson A237
URL: https://sakai.rutgers.edu

Course Directors:
Dr. Linda Brzustowicz
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The best way to contact the course directors is by email. To help us identify your email as related to class, please be sure to put "**Human Genetics 2017**" in your email subject header. We try to respond to class emails within 24 hours M-F. Office hours are by appointment.

Course Description:
This is a foundational graduate course in human genetics, covering classical and non-classical patterns of inheritance, human genome structure and evolution, normal and abnormal processes of gene expression, molecular genetic pathology, laboratory methods for genetic analysis, and research approaches to the study of human genetic disease using humans and model organisms. There will be homework assignments, a midterm and final.

Course Goals:
The goals of this Human Genetics course are to provide students a foundation in the key biological concepts and experimental methodologies of human genetics. Specific course objectives are to:

1. Comprehend and apply knowledge of human genetics as it relates to a variety of topics including inheritance patterns, population and quantitative genetics, and epigenetics
2. Appreciate a variety of genetic and genomic testing technologies and understand their application and utility, in both research and clinical settings
3. Understand the basis of human variation and disease
4. Understand the utility and limitations of model organism research and how such work leads to advances in the understanding and treatment of human genetic disease

The course will also cover the following Practice Based Competencies for Genetic Counseling:

Domain I: Genetics Expertise and Analysis:

1. **Demonstrate and utilize a depth and breadth of understanding and knowledge of genetics and genomics core concepts and principles**
   a) In specific content areas and
   b) Apply this knowledge and understand how it contributes to etiology, clinical features, natural history, differential diagnoses, genetic testing and test report interpretation, pathophysiology, recurrence risk, management and prevention, and population screening.

5. **Assess individuals' and relatives probability of conditions with a genetic component or carrier status based on pedigree, test results, and other pertinent information.**
   a) Use relevant knowledge and data based on pedigree analysis, inheritance, genetic epidemiology, quantitative genetic principles, and mathematical calculations.

Prerequisites:
16:695:538 or 16:115:512 or enrolled in the Genetic Counseling Master's track.
Undergraduate seniors with 01:477:384 and 01:477:385 may contact the instructor for permission

Recommended Textbook:
Tom Strachan and Andrew P. Read, *Human Molecular Genetics, 4th Edition,* Garland 2010.
Supplemental readings will be posted on Sakai.

Assignments, Due Dates, and Course Announcements: Students are responsible for being aware of all assignment due dates, which are included with each assignment. Changes to due dates or lecture topics are made in class and/or will be posted on the class Sakai website.

Homework:
There will be approximately six homework assignments (problem sets). While students are encouraged to work together on the homework in a general fashion, it is expected that everyone will ultimately complete their own assignments individually. Homework will be due at the start of class. In addition, there may be unannounced quizzes during class time that relate to the content of the homework due that day.

Attendance:
Attendance at each class is expected unless the student is otherwise excused.

Grading:
Final grades will be determined from the following components:

35% Homework and quiz grades
25% Midterm Exam
40% Final Exam (cumulative)

Academic Integrity: ***We expect honesty and integrity of every student in this course.***
Students are encouraged to study together and in some cases may be required to work with one another. However assignments that are turned in for grading must represent each student's individual work – they may not be copied from another person's work.

The Rutgers academic integrity policy can be found here: **http://academicintegrity.rutgers.edu/**

Students who violate the Rutgers Integrity policies will be reported to the Office of Student Conduct. Sanctions will be determined by the Office of Student Conduct according to the procedures described in the University Policy on Academic Integrity.

Class Schedule

Class	Date	Topic	Instructor
1.	Wed Sept 6	Introduction and Classic Mendelian Inheritance Patterns	Brzustowicz
2.	Mon Sept 11	Complex Traits and Non-Mendelian Genetics	Brzustowicz
3.	Wed Sept 13	Hardy-Weinberg and Population Genetics	Brzustowicz
4.	Mon Sept 18	Genetic Epidemiology, Family Aggregation and Segregation Studies	Heiman
5.	Wed Sept 20	Cytogenetics	Aviv
6.	Mon Sept 25	Gene Expression and Regulation	Verzi
7.	Wed Sept 27	Human Genome Structure	Chen
8.	Mon Oct 2	Human Genome Variation and Evolution	Chen
9.	Wed Oct 4	Epigenetics and Complex Disease: Gene-Environment Interactions	Serrano
10.	Mon Oct 9	Molecular Basis of Single Gene Disorders	Tischfield
11.	Wed Oct 11	Cancer Genetics	Axelrod
12.	Mon Oct 16	Pharmacogenetics, Pharmacogenomics, and Personalized Medicine	Cartegni
13.	Wed Oct 18	Midterm	
14.	Mon Oct 23	Fundamentals of Molecular Diagnostics	Tischfield
15.	Wed Oct 25	Non-Coding RNA	Chen
16.	Mon Oct 30	Laboratory Methods for DNA Analysis	Brooks
17.	Wed Nov 1	Laboratory Methods for RNA and Protein Analysis	Brooks
18.	Mon Nov 6	Linkage and Association Analysis	Brzustowicz
19.	Wed Nov 8	Human Genetic Database Resources	Brzustowicz
20.	Mon Nov 13	Next Generation Sequencing in Research and Practice	Xing
21.	Wed Nov 15	Sequencing, Sequence Analysis, and Function Prediction	Xing
22.	Mon Nov 20	Human Reproduction and Reproductive Technologies	Treff
23.	Mon Nov 27	Ethical Issues	Joines
24.	Wed Nov 29	Model Systems for Human Disease: Comparative Genomics & Invertebrate Models	Barr
25.	Mon Dec 4	Non-Mammalian Models for Human Genetic Disease II	Barr
26.	Wed Dec 6	Model Systems for Human Disease: Mammalian Cell Lines	Moore
27.	Mon Dec 11	Model Systems for Human Disease: Mouse Models	Millonig
28.	Wed Dec 13	Human Genetic Research – Practical Considerations	Brzustowicz
29.	TBA	FINAL	