Temple University
Department of Biology
College of Science & Technology

HUMAN GENETICS
BIOLOGY 3201
FALL, 2017

Lecture time:	M W F, 12:00-12:50 PM
Lecture location:	332 Biology-Life Sciences
Instructor:	Rhonda H. Nicholson, Ph.D., Assistant Professor for Biology
Office address:	402B Biology-Life Sciences
Office phone:	215-204-9547
Email address:	rnichol@temple.edu
Office hours:	Monday and Wednesday, 2:30-4:00 PM (or by appointment)

Prerequisite: Genetics (Biology 2296) Grade: C- or better

Course Materials: The required textbook is "Thompson & Thompson Genetics in Medicine", 8th edition (2016), by Nussbaum, McInnes, and Willard, available at the Temple University Bookstore or online.

Course Goals: This course examines all primary topics in medical human genetics including the chromosomal basis of disease as well as single-gene and multifactorial genetic diseases that lead to birth defects and cancer. When known, the biochemical bases of human genetic diseases will be emphasized. Genetic variation in populations and why certain disease genes are maintained in specific populations will also be studied. We will then apply our knowledge of human genetic disease to risk assessment, genetic screening, genetic counseling, and personalized medicine.

Academic requirements: Students are expected to attend every lecture and take careful notes. Students are strongly encouraged to re-copy their lecture notes in order to incorporate important points from the required reading assignments. Study guides will be provided to help you focus and learn the lecture and reading material. The carefully written and annotated lecture/reading notes and completed study guides will be used for reference during examinations. *Lecture notes and study guides used for reference during examinations must be hand written with no photocopies from either the textbook or notes from another student. Failure to comply will result in disqualification of your test.*

Grading: The final grade in this course will be determined from four examinations:
Examination I=20%
Examination II= 20%
Examination III=20%
Final examination: Part I – 10%
Part II – Cumulative concepts from examinations I - IV =10%
Grades will be assigned on a *straight scale* according to the following formula:
100-87%: A, 86%-78%: B, 77-67%: C, 66-58%: D, 57% and below: F

Examination format: **The examination format will be "short answer" (2-8 sentences) and calculation of genetic problems.**
There will be no make-up examinations. ***Exams must be taken during their scheduled time.*** **If you are absent from an examination due to a medical emergency, a letter explaining the circumstances, signed and dated by a physician (unrelated to the student), must be submitted to me as soon as it is reasonably possible. If you miss an examination and I am not notified within 36 hours, you will receive a zero for that examination. In the event that an examination is missed due to an *officially documented* medical or legal emergency, the average of the other two regular examinations will be substituted for the score of the missed examination. You must also provide to the instructor *handwritten* completed study guides and lecture notes to prove that you finished the work covered on the missed examination.** ***This does not apply to the final examination.*** **If you miss the final due to serious medical and/or legal circumstances, you must provide proper documentation as described above within 36 hours of the scheduled final examination. In this event you must take a make-up examination by Friday, December 22nd. If you cannot take the final by December 22nd, you will have to take an incomplete. For information on the Incomplete Policy, refer to Temple University Policy #02.10.13 on the "Policies and Procedures" link on the Temple homepage.**

Incomplete: You must be passing the course and only miss the final examination in order to apply for an "Incomplete". If circumstances force you to miss the final and apply for an incomplete, two steps must be taken. First, the professor and the student must sign an "Incomplete Contract" that is held in the Biology Office.

Course withdrawal: The last day to drop a course is Monday, September 11th. The final date for course withdrawal (no tuition refund) is Tuesday, October 24th. The following statement regarding course withdrawal is from the Temple University student bulletin, policy # 02.10.14:

During the first two weeks of the fall or spring semester or summer sessions, students may withdraw from a course with no record of the class appearing on the transcript. In weeks three through nine of the fall or spring semester, or during weeks three and four of summer sessions, the student may withdraw with the advisor's permission. The course will be recorded on the transcript with the instructor's notation of "W," indicating that the student withdrew. After week nine of the fall or spring semester, or week four of summer sessions, students may not withdraw from courses.

- ***No student may withdraw from more than five courses during the duration of his/her studies to earn a bachelor's degree.***
- ***A student may not withdraw from the same course more than once.***
- ***Procedure: Withdrawal from a course is accomplished with a Schedule Revision (Drop/Add) form, processed through a registration office.***

Cell Phones: *Cell phones must be off during class!* A student who engages in text messaging, answers or places a cell phone call during lecture will be penalized for misconduct in accordance with university regulations.

Letters of Recommendation: You must meet with me during the semester and receive a final grade of B or better in order to obtain a positive letter of recommendation. If you are applying to graduate, medical, or dental school, please provide a Professional School Recommendation Form (available from the Office of Pre-Professional Health Studies) along with a personal statement.

For students applying to post-baccalaureate programs, please provide a personal statement and a list of *no more than 5* recipients for the letter of recommendation.

Disability disclosure: ***Any student who has a need for accommodation based on the impact of a documented disability should contact me privately to discuss the specific situation by the second week of classes. To learn about resources available to you, please contact Disability Resources and Services at 215- 204-1280 in 100 Ritter Annex. I will work with DRS to coordinate reasonable accommodations for all students with documented disabilities.***

Student and Faculty Academic Rights and Responsibilities Policy:
Freedom to teach and freedom to learn are inseparable facets of academic freedom. The University has a policy on Student and Faculty and Academic Rights and Responsibilities (Policy #03.70.02) that can be accessed through the following link: http://policies.temple.edu/getdoc.asp?policy_no=03.70.02.

BIOLOGY 3201 LECTURE SCHEDULE (Note: schedule may be subject to change)

LECTURE	DATE	TOPIC	CHAPTER
1	Mon. (8/28)	Human Genetic Disease Introduction The Human Genome I Organization and Transmission	2
2	Wed. (8/30)	The Human Genome II Gene Structure and Function	3
3	Fri. (9/01)	The Human Genome III Epigenetics and Imprinting	3
4	Wed. (9/06)	Mutation and Polymorphism	4
5	Fri. (9/08)	Clinical Genomic Analysis I Cytogenetics and Genomic sequencing	5
6	Mon. (9/11)	Clinical Genomic Analysis II Microarray analysis	5
7	Wed. (9/13)	Chromosomal Basis of Disease I Aneuploidy, Duplications, and Deletions	6
8	Fri. (9/15)	Chromosomal Basis of Disease II Sex Chromosome Disorders	6
9	Mon. (9/18)	Single-Gene Disorder Inheritance Patterns I Autosomal and X-Linked	7
10	Wed. (9/20)	Single-Gene Disorder Inheritance Patterns II Mosaicism, Expansion, Mitochondrial Mutations	7
--	**Fri. (9/22)**	**TEST 1 (Lectures 1-8)**	
11	Mon. (9/25)	Multifactorial Genetic Disorders I Analysis of Quantitative Traits	8
12	Wed. (9/27)	Multifactorial Genetic Disorders II	8
13	Fri. (9/29)	Multifactorial Genetic Disorders III Environmental Contributions	8

LECTURE	DATE	TOPIC	CHAPTER
14	Mon. (10/02)	Identifying the Genetic Basis of Human Disease Linkage Analysis, Association, Disequilibrium	10
15	Wed. (10/04)	Disease Gene Mapping and Identification I *L*ogarithm of the *OD*ds	10
16	Fri. (10/06)	Disease Gene Mapping and Identification II Odds ratio and Relative Risk	10
17	Mon. (10/09)	Molecular Basis of Genetic Disease I Categories of Mutation, Hemoglobinopathies	11
18	Wed. (10/11)	Molecular Basis of Genetic Disease II Mutations and Enzymopathies	12
--	**Fri. (10/13)**	**TEST 2 (Lectures 9-16)**	
19	Mon. (10/16)	Molecular Basis of Genetic Disease III Mutations of Receptors and Transporters	12
20	Wed. (10/18)	Molecular Basis of Genetic Disease IV Mutations of Structural Proteins	12
21	Fri. (10/20)	Molecular Basis of Genetic Disease V Mitochondria, Unstable Repetitive Sequences	12
22	Mon. (10/23)	Treating Genetic Diseases I Metabolic Manipulation	13
23	Wed. (10/25)	Treating Genetic Diseases II Enhancement of Protein Function	13
24	Fri. (10/27)	Treating Genetic Diseases III Modulation of Gene Expression	13
25	Mon. (10/30)	Treating Genetic Diseases IV Gene Therapy	13
26	Wed. (11/01)	Developmental Genetics I Dysmorphology	14
--	**Fri. (11/03)**	**Test III (Lectures 17-25)**	

LECTURE	DATE	TOPIC	CHAPTER
27	Mon. (11/06)	Developmental Genetics II Overview of Human Development	14
28	Wed. (11/08)	Developmental Genetics III Molecular Mechanisms of Development	14
29	Fri. (11/10)	Genetic Basis of Cancer I Tumor Promoter and Tumor Suppressor Genes	15
30	Mon. (11/13)	Genetic Basis of Cancer II Hereditary Cancer Syndromes	15
31	Wed. (11/15)	Genetic Basis of Cancer III Genomics and Cancer Therapy	15
32	Fri. (11/17)	Genetic Counseling I Mendelian Applications and Risk Assessment	16
33	Mon. (11/27)	Genetic Counseling II Complex Disorders	16
34	Wed. (11/29)	Prenatal Diagnosis and Screening I Methods of Prenatal Diagnosis	17
--	**Fri. (12/01)**	**Test IV (Lectures 26-33)**	
35	Mon. (12/04)	Prenatal Diagnosis and Screening II Prenatal Screening	17
36	Wed. (12/06)	Population Genetics I Hardy-Weinberg Law	9
37	Fri. (12/08)	Population Genetics II Mutation, Selection, and Genetic Disease Frequency	9
38	Mon. (12/11)	Genetics and Personalized Medicine	18
--	**Wed. (12/20)**	**FINAL EXAMINATION 10:30-12:30** **(Lectures 34-38; Cumulative Section Lectures 1-33)**	