

**Medical Genetics - BIOL 4820  
Spring 2010**

**Instructor:** Dr. Janey Youngblom  
**Office Hours:** Tues. & Thurs. 2:30-4:00  
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**Required Text:** Thompson and Thompson Genetics in Medicine, by Robert Nussbaum, Roderick McInnes, Huntington Willard (2007)

**Problems at the end of the chapters:** It is highly recommended that you do the problems at the end of each assigned chapter. At least one question from the problem set for each of the assigned chapters will be included in the exams. All the answers to the problems are provided in the back of the book.

**Optional book:** Genome: The Autobiography of a species in 23 chapters (2000), by Matt Ridley (order through Amazon.com). 5 point extra credit quiz given on the last day of class.

<b>DATE</b>	<b>TOPIC</b>	<b>T &amp; T Chapter Readings</b>
2/16 -2/18	Introduction: Genetics and Genomics in Medicine The Human Genome and the Chromosomal basis of Heredity The Human Genome: Gene Structure and Function	1 (review) 2 (review) 3
	Tools of Human Molecular Genetics	4
2/23	<b>FURLOUGH DAY – TUESDAY ONLY</b>	
2/25	Principles of Clinical Cytogenetics Epigenetics	5
3/2- 3/4	Clinical Cytogenetics	6
3/9- 3/11	Patterns of Single-Gene Inheritance (includes consanguinity and coefficient of inbreeding) (includes gain of function, haploinsufficiency, dominant negative,)	7 11 (323-5)
3/16	<b>MIDTERM – 50 pts. (last day to drop is March 15)</b>	
3/18	Genetic Counseling and Risk Assessment (includes Bayes analysis)	19
3/23- 3/25	Genetics of Common Disorders with Complex Inheritance Genetic Variation in Individuals and Populations	8, 19 (519-520) 9
3/30 4/1	Genetic Variation in Individuals and Populations cont'd <b>FURLOUGH DAY- THURSDAY ONLY</b>	9
4/5- 4/9	<b>SPRING BREAK</b>	
4/13- 4/15	Human Gene Mapping and Disease Gene Identification (including LOD scores)	10, 19 (518-519)
4/20	<b>Midterm #2 – 50 pts.</b>	
4/22	The Molecular, Biochemical, and Cellular Basis of Genetic Diseases	12
4/27- 4/29	Treatment of Genetic Diseases Prenatal Diagnosis	13 15

5/4- 5/6	Ethical Issues in Medical Genetics Cancer Genetics and Genomics	20 16
5/11	Personalized Genetic Medicine Pharmacogenetics and Pharmacogenomics	17 18
5/13	Student presentations	
5/18- 5/20	Student presentations	
6/1	<b>Final Exam (8:30- 10:30) – 50 pts.</b>	

## REQUIREMENTS FOR THE COURSE AND GRADING INFORMATION

1. **Exams** – Two midterms and one final. 50 pts each. Final is NOT cumulative.
2. **Term Paper** (Written and Oral presentation) – 50 pts.  
**(TOPIC MUST BE APPROVED BY THE INSTRUCTOR BEFORE SPRING BREAK)**

### Written Term Paper – 40 pts (DUE – day of the final exam)

Students will work in pairs to select a medically and genetically related topic. Selection of topics may include interesting diseases, public policy issues, interesting case studies, bioethical issues, new diagnostic technologies, screening programs, and a number of other possibilities. Think creatively!

The paper should be 6 pages in length, typed, 12 point font, one inch margins on all sides, and double spaced. One paper will be jointly prepared and submitted with your partner. A bibliography must be provided with a minimum of 8 different references. At least 3 of these references must be current, with publication dates within the last 2 years or less. No more than half of the references can be internet citations, unless they are peer-reviewed journal articles. Use the interlibrary loan to receive hard copies of journal articles not available online or at our campus library. Since some articles may take over a week to arrive, you need to start your paper as soon as possible.

The term paper will be graded on the following criteria:

- Thoroughness of literature review and content coverage of the topic
- Clarity of information conveyed
- Complexity and originality of the topic
- Organization of the content
- Grammar and spelling
- Adherence to term paper guidelines

### Oral Powerpoint presentation - 10 pts.

Each student must prepare and present a powerpoint presentation on their term paper topic. The presentation should be about 8 minutes long, with a couple of additional minutes allowed afterwards for answering questions.

The presentation will be graded on the following criteria:

- Thoroughness of content coverage for the topic
- Preparedness of the presenters (reading the entire presentation is not allowed)
- Clarity of information conveyed
- Organization of the content
- Creativity of the presentation content
- How well the presenters answer questions from the audience

3. **Clickers** – interspersed throughout the semester will be lectures that include clicker participation. There will be a total of 5 days when they are used. For each of these days, you will get a maximum of 5 points for your participation. Total = 25 points for the whole semester.

**4. Extra Credit – 5 point quiz on the Ridley book given on the last day of class.**

#### 4. **Total points and Grading**

Total points = 225 pts.

This class can only be taken for a letter grade. Credit/no credit grading is NOT an option. The plus/minus grading system will be implemented as follows:

93.5-100%	= A
90-93%	=A-
87-89.5%	=B+
83.5-86.5%	=B
80-83%	=B-
77-79.5%	=C+
73.5-76.5	=C
70-73%	=C-
67-69.5%	=D+
63.5-66.5	=D
60-63%	=D-
<60%	=F