

Medical Genetics - BIOL 4820
Spring 2016

Instructor: Dr. Janey Youngblom
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Required Text: *Medical Genetics*, by Jorde, Carey, and Bamshad 2016 (5th edition)

DATE	<u>TOPIC</u>	<u>Chapter Readings</u>
January 28	Background and History	1
February 2	Autosomal Dominant and Recessive Inheritance	4
February 9	Sex-Linked and Nontraditional Modes of Inheritance	5
February 16	Clinical Cytogenetics: The Chromosomal Basis of Human Disease	6
February 23	Family history and pedigree due (3/15) using US Surgeon General Tool Clinical Cytogenetics: (continued) February 24 – LAST DAY TO DROP	- 10 pts 6
March 1	Biochemical Genetics: Disorders of Metabolism Video on William syndrome (~15 minutes)	7
March 8	EXAM #1 (Tuesday) – 50 pts. Genetic Testing and Gene Therapy	13
March 15	Activity #1 Multifactorial Inheritance and Common Diseases Family history and pedigree assignment due	12
March 22	Cancer Genetics Video – BRC genes and Gleevac (~ one hour)	11
March 29	Spring Break – NO CLASS	
April 5	Activity #2 Clinical Genetics and Genetic Counseling Must Notify instructor of your term paper topic	15
April 12	EXAM #2 (Tuesday) – 50 pts.	
April 19	Variant Interpretation	Separate worksheet
April 26	Immunogenetics	9
May 3	Developmental Genetics	10
May 10	Oral Presentations	
May 17	Oral Presentations	
May 24	FINAL EXAM (Tuesday 11:15 - 1:15) – 50 pts.	

REQUIREMENTS FOR THE COURSE AND GRADING INFORMATION

1. **Exams** – Three exams total, 50 pts. each. The final exam is NOT cumulative. **BRING SCANTRON FORM NO. 882-E for each exam, including the final.**
2. **Family History intake and pedigree.** – 10 pts.
Go to URL <http://www.hhs.gov/familyhistory/> for the US Surgeon General Family Health Portrait Initiative and Tool. Use the Tool to answer family health history information and print out a copy of your pedigree. Turn it in to the instructor by the due date provided in your syllabus.
3. **Case Presentation and accompanying Term Paper** (Written and Oral presentation) – 50 pts.
(Case studies provided by the instructor)

Written Term Paper – 25 pts

Each group of 4 students will be given a case scenario. The information that should be covered for each case scenario is listed in the table below for grading criteria. **The group should turn in ONE paper** that is 5 pages in length, typed, 12 point font, one inch margins on all sides, and double spaced. A bibliography must be provided with a minimum of 5 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.

Grading Criteria for Case Studies
Quality of background information provided about the specific genetic condition/topic.
Briefly but concisely describe the laboratory technique(s) used in generating the lab results, if applicable
Thoroughness in explaining and interpreting the results of the genetic test(s), and discussing <ul style="list-style-type: none"> - Their Clinical implications for the patient - Potential implications for other family members (if appropriate)
Appropriateness and completeness of recommendations on follow-up studies or next steps for the patient and/or other family members
Addressing any potential psychosocial or ethical considerations involved in the case, if any
Discussion of resources (e.g. support groups) or services available for the patient and/or family members
Grammar, spelling, and adherence to written paper guidelines

Oral Powerpoint presentation - 25 pts. (Each member of the group must contribute equally to the group effort. If one or more members do not contribute their fair share to the group, they will be scored accordingly. Thus not all members of the group will necessarily receive the same score. More about this will be discussed by the instructor.)

Each group of students must provide a powerpoint presentation on their case study. The presentation should be about 10 minutes long, with a couple of additional minutes allowed afterwards for answering questions.

The powerpoint presentation will be graded on the following criteria:

- Thoroughness in covering all the criteria listed above for the written paper, except the last criteria
- Preparedness of the presenters (reading the entire presentation is not allowed)
- Clarity of information conveyed
- Organization of the content
- How well the presenters answer questions from the audience

4. Activities – 15 pts. for each of two activities turned in during the semester (Total = 30 points)

During the course of the semester, there will be 2 class meetings in which you will break up into groups and conduct activities in smaller groups. These dates will be announced approximately 2 class periods in advance and posted on blackboard.

5. Miscellaneous - 10 points total. It will include turning in additional assignments announcement throughout the term, e.g. writing a commentary to an assigned reading article, watching a video and writing a reflection paper.

- 1) First assignment for 5 pts- Reflection paper : Listen to NPR This American Life episode titled “The DIY Scientist, the Olympian, and the Mutated Gene”. You can access the episode by going to <http://www.thisamericanlife.org/radio-archives/episode/577/something-only-i-can-see> and downloading it as a podcast. There is one story before this main story. Write your thoughts about the episode in a 2-page reflection paper: double spaced, 12 pt font, 1” margins all around.

Due date: February 16th. Submit your paper in class (5 pts.)

6. Total points and Grading

Total points = 250 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