

**Medical Genetics - BIOL 4820
Spring 2018**

Instructor: Dr. Janey Youngblom
Office Hours: Tues. & Thurs. 2:30 – 4:00
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Room: N-265

Required Text: Medical Genetics, by Jorde, Carey, and Bamshad 2016 (5th edition)

<u>DATE (week of)</u>	<u>TOPIC</u>	<u>Chapter Readings</u>
January 25	Background and History	1
January 30	Autosomal Dominant and Recessive Inheritance	4
February 6	Sex-Linked and Nontraditional Modes of Inheritance	5
February 13	Bayes analysis worksheet Clinical Cytogenetics: The Chromosomal Basis of Human Disease	6
February 20	Clinical Cytogenetics: (continued) (February 21 – Last day to drop)	6
February 27	Biochemical Genetics2: Disorders of Metabolism Genetics and Personal Medicine: Pharmacogenomics	7 14
March 6	EXAM #1 (Tuesday) – 50 pts. Multifactorial Inheritance and Common Diseases	12
March 13	Genetic Testing and Gene Therapy Gene Mapping: Linkage Analysis (exclude LOD scores)	13 8 (up to p.164-5 th ed)
March 20	Cancer Genetics Video – BRC genes and Gleevac	11
March 27	Clinical Genetics and Genetic Counseling March 29 - Family history and pedigree due (10 pts)	15
April 3	Spring Break – NO CLASS ALL WEEK	
April 10	EXAM #2 (Tuesday) – 50 pts. April 12 - Digital Stories presentations	
April 17	Digital Stories presentations continued	
April 24	Whole genome/whole exome sequencing	
May 1	Variant Interpretation (tentative)	Separate worksheet
May 8	work in groups on variants (tentative)	Separate worksheet
May 15	Immunogenetics (May 16 = Last day of classes)	9
May 22	FINAL EXAM (Tuesday 8:30-10:30) – 50 pts.	
NOTES:	1-2 guest presentations may be scheduled during the semester, either via zoom or in-person	

REQUIREMENTS FOR THE COURSE AND GRADING INFORMATION

1. Exams – Total = 150 exam points.

Three exams, 50 pts. each. The final exam is NOT cumulative. **BRING SCANTRON FORM NO. 882-E for each exam, including the final.** There will be no makeups for the exam, except if you contact the instructor BEFORE the day of the exam with a valid excuse (e.g. letter from your doctor)

2. Family History intake and pedigree. – 10 pts. Due March 29

Go to URL <http://www.hhs.gov/familyhistory/> for the US Surgeon General Family Health Portrait Initiative and Tool
On the left side menu list, click “My Family Health Portrait 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. Digital Story- 40 pts. (Rubric for grading to be posted on Blackboard) – Presentations start April 12

- About 5 minutes in length.
- Tell a story (either you, have a personal someone else, groups of people), or serve as an educational video for others. Personal stories are generally more compelling
- Focus has to be related to medical genetics, for example:
 - interview an individual or family with a medical disease (e.g. cystic fibrosis, Huntington, Down syndrome),
 - use of genetic testing for personal training enhancement
 - nutrigenomics (need to tease out fake news vs peer reviewed literature)
 - genetic disease support groups
 - etc.
- Example:
 - a) Description: A digital story about a CSU Stanislaus student with mosaic Turner syndrome. Created by Fatima Feroze and Harpreet Mann

4. Activities (In class or homework)– 40 pts total

During the course of the semester, there will be two 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.

a) Bayes risk assessment worksheet (10 pts)

b) variant interpretation- students will be assigned gene variants that they need to curate and interpret – (25 pts)

c) third activity- (5 pts) – writing assignment for “Patients in Waiting” article on Blackboard – Due date TBA

5. Total points and Grading

Total points = 240 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