Medical Genetics - BIOL 4820
Spring 2019

Instructor: Dr. Janey Youngblom
Office Hours: Tues. & Thurs. 2:15 – 3:45
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Room: N-265

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

DATE (week of)	TOPIC	Chapter Readings
January 29	Background and History	1
January 31	Autosomal Dominant and Recessive Inheritance	4
February 5	Sex-Linked and Nontraditional Modes of Inheritance	5
February 12	Clinical Cytogenetics: The Chromosomal Basis of Human Disease	6
February 19	Clinical Cytogenetics: (continued) **(February 21 – Last day to drop)**	6
February 26	Biochemical Genetics2: Disorders of Metabolism Genetics and Personal Medicine: Pharmacogenomics **February 26 - Family history and pedigree due (10 pts)**	7 14
March 5	**EXAM #1 (Tuesday) – 50 pts.** Multifactorial Inheritance and Common Diseases	12
March 12	Genetic Testing and Gene Therapy Gene Mapping:Linkage Analysis (exclude LOD scores) **March 12 – "Patients in Waiting" paper due (5 pts)**	13 8 (up to p.164-5th ed)
March 19	**Spring Break – NO CLASS ALL WEEK**	
March 26	Cancer Genetics Video – BRC genes and Gleevac Bayes analysis worksheet	11
April 2	Clinical Genetics and Genetic Counseling	15
April 9	**EXAM #2 (Tuesday) – 50 pts.** **April 11 - Digital Stories presentations (30 pts)**	
April 16	**Digital Stories presentations continued**	
April 23	Whole genome/whole exome sequencing	
April 30	Variant Interpretation	Separate worksheet
May 7	work in groups on variants	Separate worksheet
May 14	Immunogenetics **May 14 - Variant Interpretation Reports Due (40 pts)** **(May 14 = Last day of class)**	9
May 21	**FINAL EXAM (Tuesday 11:15 – 1:15) – 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 in 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 February 26
   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- 30 pts. (Rubric for grading to be posted on Blackboard) – Presentations start April 11**
   - 5 minutes if a solo project;10 mins if you work with one other partner
   - Tell a story (either about you, another person, groups of people), or create an engaging 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),
     - experience with direct-to-consumer genetic testing through companies such as 23andme, Ancestry, etc
     - nutrigenomics (need to tease out fake news vs peer reviewed literature)
     - genetic disease support groups
     - etc.
   - Example:of a digital story
     a) Description: A digital story about a CSU Stanislaus student with mosaic Turner syndrome. Created by Fatima Feroze and Harpreet Mann (interview starts at 6:06)

4. **Activities (In class/homework)– 50 pts total**
   There will be two class activities in which you will break up into groups and conduct activities in smaller groups.

   a) Bayes risk assessment worksheet (10 pts)
   b) variant interpretation- students will be assigned gene variants that they need to curate and write up a report using worksheets provided – (40 pts)

5. **Writing assignment (5 pts)**
   Read the article "Patients in Waiting" posted on Blackboard
   Due**: March 12**

5. **Total points and Grading**

   Total points = 245 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