

**Medical Genetics - BIOL 4820  
Spring 2017**

**Instructor:** Dr. Janey Youngblom  
**Office Hours:** Tues. & Thurs. 2:15-3:45  
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**Required Text:** Medical Genetics, by Jorde, Carey, and Bamshad 2016 (5<sup>th</sup> edition)

| <u>DATE</u>     | <u>TOPIC</u>   | <u>Chapter Readings</u>                  |
|-----------------|--|--|
| January 26      | Background and History   | 1  |
| January 31      | Autosomal Dominant and Recessive Inheritance   | 4  |
| February 7      | Sex-Linked and Nontraditional Modes of Inheritance   | 5  |
| February 14     | <b>Instructor in SF on Feb 14, but class will be held</b><br>Clinical Cytogenetics: The Chromosomal Basis of Human Disease | 6  |
| February 21     | Clinical Cytogenetics: (continued)<br><b>February 22 – LAST DAY TO DROP</b>  | 6  |
| February 28     | Biochemical Genetics <sup>2</sup> : Disorders of Metabolism<br>Genetics and Personal Medicine: Pharmacogenomics            | 7<br>14                                  |
| <b>March 7</b>  | <b>EXAM #1 (Tuesday) – 50 pts.</b><br>Multifactorial Inheritance and Common Diseases                                       | 12                                       |
| March 14        | Genetic Testing and Gene Therapy<br>Gene Mapping: Linkage Analysis (exclude LOD scores)                                    | 13<br>8 (up to p.164-5 <sup>th</sup> ed) |
| <b>March 21</b> | <b>Spring Break – NO CLASS ALL WEEK</b>  |  |
| March 28        | <b>Digital Stories presentations (5 mins) – all week</b>   |  |
| April 4         | <b>Digital Stories continued</b><br>Cancer Genetics<br>Video – BRC genes and Gleevac (~ one hour)                          | 11                                       |
| <b>April 11</b> | <b>EXAM #2 (Tuesday) – 50 pts.</b><br>Clinical Genetics and Genetic Counseling   | 15                                       |
| April 18        | Whole genome/whole exome sequencing<br><b>Family history and pedigree due using US Surgeon General Tool - 10 pts</b>       |  |
| April 25        | Variant Interpretation   | Separate worksheet                       |
| May 2           | work in groups on variants   | Separate worksheet                       |
| May 9           | Immunogenetics   | 9  |
| May 16          | Developmental Genetics   | 10                                       |
| <b>May 23</b>   | <b>FINAL EXAM (Tuesday 11:15 - 1:15) – 50 pts.</b>   |  |

## 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 April 18

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. Digital Story- 40 pts. (Rubric for grading to be posted soon) – week of March 28<sup>th</sup>

- 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, e.g.
  - 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.
- Examples:
  - a) Description: A personal reflection on the family effects of cardiovascular disease, smoking, and cancer. (7:30)  
[http://digitalstorytelling.coe.uh.edu/view\\_story.cfm?vid=86&categoryid=4&d\\_title=Health/Medical](http://digitalstorytelling.coe.uh.edu/view_story.cfm?vid=86&categoryid=4&d_title=Health/Medical)  
– not great and too long, but gives you some idea of a personal story
  - b) Description: A digital story that helps patients understand the anatomy, physiology and treatment of hypoplastic left heart syndrome, a rare congenital heart defect. (4:01)  
[http://digitalstorytelling.coe.uh.edu/view\\_story.cfm?vid=414&categoryid=4&d\\_title=Health/Medical](http://digitalstorytelling.coe.uh.edu/view_story.cfm?vid=414&categoryid=4&d_title=Health/Medical)

### 4. Activities – 40 pts total

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.

- a) variant interpretation of a genetic result associated with a medical case
- b) Bayes risk assessment worksheet
- c) option for a third activity- to be announced

### 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  |