Course: Biomedical Sciences 5334, Medical Genetics
Monday & Wednesday 3:30 – 4:45 pm      ST 106

Faculty: Suzzette F. Chopin, Ph.D.
CS 130; 825-6022
Office Hours:
   Mondays & Wednesdays 5:00-6:30 pm
   Tuesdays & Thursdays 1:00 – 2:00 pm
   Other times by appointment
Email: suzzette.chopin@tamucc.edu

Course Description:
This course is an examination of the genetic basis of health and disease. We will examine
the hereditability, diagnosis and treatment of disorders arising from chromosomes abnormalities
and individual genes. We will discuss ethical considerations related to genetics.

Supplies:
Syllabus, PowerPoint presentations and handouts printed from Blackboard
Journal articles printed from Blackboard
Binder for notes, handouts, articles and outlines; bring your binder to class every day.

Text
Nussbaum, McInnes and Willard. 2007. Thompson and Thompson Genetics in
Medicine 7th edition. Saunders: Canada ($62 from Amazon.com)

Course Objectives/Student Learning Outcomes: At the conclusion of the course, the student will
be able to:
1. Describe the genetic foundations of certain pathological conditions;
2. Distinguish between gross chromosomal abnormalities and single gene mutations;
3. Discuss cytogenetics;
4. Identify molecular techniques relevant to clinical practice;
5. Explain the principles of pharmacogenetics;
6. Correlate clinical conditions, demographics and pharmacogenetics;
7. Discuss genetic counseling; and
8. Evaluate ethical considerations related to genetic testing, genetic counseling and gene
patents.

Course Requirements And Grading Points:
You need to structure your personal schedule so that you are present for all activities that result in a
grade. Doctor appointments, vacations, work schedules, etc. are secondary to your commitment to
academics. Manage your time so that you are able to submit all work on the due date, take tests on
the date in the agenda, be present in class when we are doing in class work.

Written work is due during the class period on the day indicated in the agenda. For example,
journal article reflections are due at the beginning of class (3:30 pm); late work is not accepted.
Excuses such as my printer broke, I ran out of ink, I ran out of paper are not acceptable.

THERE IS NO OPPORTUNITY FOR MAKING UP WORK

Chopin July 2011
**GRADED ACTIVITIES:** Work is due on the day/time indicated in the table below.

<table>
<thead>
<tr>
<th>Activity</th>
<th>Date</th>
<th>Points</th>
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</thead>
<tbody>
<tr>
<td><strong>Tests: Lowest grade dropped</strong></td>
<td></td>
<td>300</td>
</tr>
<tr>
<td>Test 1</td>
<td>W 9/21</td>
<td>100</td>
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<tr>
<td>Test 2</td>
<td>W/10/21</td>
<td>100</td>
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<tr>
<td>Test 3</td>
<td>M 11/21</td>
<td>100</td>
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<tr>
<td>Comprehensive Final Exam</td>
<td>W 12/?</td>
<td>100</td>
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<tr>
<td><strong>Article Exercises</strong></td>
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<td>100</td>
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<tr>
<td>Exercise 1: <em>Non-invasive Prenatal Diagnosis</em></td>
<td>M 9/12</td>
<td>25</td>
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<tr>
<td>Exercise 2: Consumers’ Views of Direct-to-Consumer Genetic Information</td>
<td>W 9/28</td>
<td>25</td>
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<tr>
<td>Exercise 3: To Know Or Not to Know: A Review of Behaviour and Suicidal Ideation in Preclinical Huntington’s Disease</td>
<td>W 10/26</td>
<td>25</td>
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<tr>
<td>Exercise 4: Should selecting saviour siblings be banned?</td>
<td>W 11/30</td>
<td>25</td>
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<tr>
<td><strong>REFLECTIONS</strong></td>
<td></td>
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<td>Speaker Summary</td>
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<td>Homework: Sexual orientation, fraternal birth order, and the maternal immune hypothesis: A review</td>
<td>W 9/7</td>
<td>25</td>
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<tr>
<td>Homework: <em>The Pathophysiology of Fragile X Syndrome</em></td>
<td>W 10/5</td>
<td>25</td>
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<tr>
<td>Video: Genetically Correct: Ensuring Perfect Babies</td>
<td>M 10/24</td>
<td>25</td>
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<tr>
<td>Video</td>
<td>TBA</td>
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<tr>
<td>Homework: Pharmacogenomics &amp; Individualized Drug Therapy</td>
<td>W 11/2</td>
<td>25</td>
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<tr>
<td>Homework: <em>Defining the Cancer Master Switch</em></td>
<td>W 11/16</td>
<td>25</td>
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<tr>
<td>Homework: Preimplantation Genetic Diagnosis</td>
<td>M 12/5</td>
<td>25</td>
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<td><strong>PRESENTATIONS</strong></td>
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<tr>
<td>Presentation 1</td>
<td>W 9/14</td>
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<td>M 9/19</td>
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<td>Presentation 2</td>
<td>M 11/28</td>
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<td><strong>Total</strong></td>
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<td>700</td>
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**The Points Required for Each Letter Grade Are:**

- 700 – 630 = A
- 629 – 560 = B
- 559 - 490 = C
- 489 – 420 = D
- 419 and below = F
TESTS:
There are three intra-term tests and a comprehensive final exam. The lowest grade is dropped.

Each exam may contain questions in the following formats: multiple choice; true/false; matching; fill in the blank; short answer questions; and essay questions. Everything that is discussed in class may on the test. This includes information from journal articles, presentations, exercises, speakers and videos.

ARTICLE EXERCISES:
Journal articles are available on Blackboard. You must read and understand the assigned article before coming to class so that you can work efficiently in the time allotted for the exercise. You do not have time to read the article and complete the exercise during the time allotted in class. You will work more efficiently if you bring your annotated article to class.

For exercises 1, 2 and 3, you will work individually and will have the last 20 minutes of class to answer questions. For exercise 4, you will work with a partner to formulate an argument and a counter-argument related to the reading. Then you and your partner will present the arguments to the class.

REFLECTIONS:

HOMEWORK:
Synopses done for homework will consist of two paragraphs. Paragraph one will be a brief summary of the article; paragraph two will be the longer paragraph in which you discuss what about the article was significant to you. Do not re-write the article’s abstract as your synopsis. The synopsis is to be your own work in your own words -- do not plagiarize. Plagiarism = 0 grade

Format: One-page-computer generated; 1" margins; 1-1.5 line spaces; ≤ 12 pt. (If you submit a half-page summary, you can earn only a maximum of half the points)

VIDEO SUMMARIES:
Twice during the course, you will write a reflection after viewing a video. The one-page reflection will be due after the video and be a discussion of what you learned from the video and what about the video was significant to you.

SPEAKER SUMMARIES:
We will have two guest speakers. At the end of each presentation, you will be given 15 minutes to write a one-page summary of the presentation.

PRESENTATIONS:
Presentations are done individually. You will deliver two 30-minute PowerPoint presentations chosen from the list of topics. You are expected to gather reference material from scientific journals or textbooks. Occasionally information from the popular press is acceptable.

Your presentation should be professionally produced and delivered. Dress professionally: you do not have to wear a suit, but jeans, T-shirts, caps are not acceptable. Email your PowerPoint to me by noon of the day of your presentation and I will make copies for your classmates. Otherwise, you will have to provide copies for your classmates.

Presentation Scoring:
Visual Aids (2.5 points)
Were the PowerPoint slides appropriate for what the presenter discussed?
Were they clear and readable?
Did they have bulleted information instead of sentences?
Were all words spelled correctly?
Did the slides look professional?

**Handouts (5 points)**
Did the presenter provide copies of the PowerPoint to each classmate?

**Delivery (7.5 points)**
Did the speaker engage the audience?
Did the speaker look at everyone?
Did the speaker speak clearly, distinctly, slowly?
Did the speaker avoid reading the slides?
Did the speaker know his/her presentation without using note cards?

**Professional Demeanor (2.5 points)**
Did the presenter project an image of a professional?
Was he/she properly dressed?

**Evidence of Use of Several Sources (2.5 points)**
Did the speaker use multiple sources?
Did the speaker use resources in addition to that provided on BlackBoard?

**Evidence of knowledge of the topic (25 points)**
Did the presenter know what he/she was discussing?
Did the presenter's explanation foster understanding by the audience?
Did the presenter respond appropriately to questions from the audience?

**Length (5 points)**
Presentation was 30 minutes

Presentation Topics – see agenda for dates of presentation

Presentation 1. Techniques
Presentation 2. Techniques
Presentation 3. GINA
Presentation 4. Gene Patents

**Class Standards:**
The student is expected to attend each class. If absent, it is the responsibility of the student to obtain missed information from a classmate. Missed information includes not only missed lecture notes, but also any possible information regarding syllabus changes.

The student has several opportunities to demonstrate competency in the course. Assessment and computation are performed by the instructor according to objective criteria. If a student experiences difficulty in the course, the instructor is available for consultation. However, it is the responsibility of the student to seek help, preferably when the investment made by the student can still be salvaged.

Academic integrity is expected. Cheating is not acceptable behavior and the student is not to give or receive help during testing. All students are expected to conform to University standards of ethics, academic integrity, grammar and spelling; review the 2011-2012 A&M-CC catalog for more information.
Cell Phone/Electronic Device Usage:  No cell phone conversations or Internet usage during class.

Academic Integrity/Plagiarism
University students are expected to conduct themselves in accordance with the highest standards of academic honesty. Academic misconduct for which a student is subject to penalty includes all forms of cheating, such as illicit possession of examinations or examination materials, falsification, forgery, complicity or plagiarism. (Plagiarism is the presentation of the work of another as one’s own work.) In this class, academic misconduct or complicity in an act of academic misconduct on an assignment or test will result in a grade of 0 for the work.

Dropping a Class
Events can sometimes occur that make dropping a course necessary. Consult with me before you drop the course. Should dropping the course be the best course of action, you must initiate the process to drop the course by going to the Student Services Center and filling out a course drop form. Just stopping attendance and participation will not automatically result in your being dropped from the class.

Classroom/Professional Behavior
Some general considerations include:
• Arrive on time for class;
• Allow others to speak; do not monopolize the discussion;
• Focus on the discussion; do not do other work during our meetings;
• Evaluate other points of view; and
• Respect others’ opinions.

Texas A&M University-Corpus Christi, as an academic community, requires that each individual respect the needs of others to study and learn in a peaceful atmosphere. Under Article III of the Student Code of Conduct, classroom behavior that interferes with either (a) the instructor’s ability to conduct the class or (b) the ability of other students to profit from the instructional program may be considered a breach of the peace and is subject to disciplinary sanction outlined in article VII of the Student Code of Conduct. Students engaging in unacceptable behavior may be instructed to leave the classroom. This prohibition applies to all instructional forums, including classrooms, electronic classrooms, labs, discussion groups, field trips, etc.

Grade Appeals
As stated in University Rule 13.02.99.C2, Student Grade Appeals, a student who believes that he or she has not been held to appropriate academic standards as outlined in the class syllabus, equitable evaluation procedures, or appropriate grading, may appeal the final grade given in the course. The burden of proof is upon the student to demonstrate the appropriateness of the appeal. A student with a complaint about a grade is encouraged to first discuss the matter with the instructor. For complete details, including the responsibilities of the parties involved in the process and the number of days allowed for completing the steps in the process, see University Rule 13.02.99.C2, Student Grade Appeals, and University Procedure 13.02.99.C2.01, Student Grade Appeal Procedures. These documents are accessible through the University Rules Web site at http://www.tamucc.edu/provost/university_rules/index.html. For assistance and/or guidance in the grade appeal process, students may contact the Office of Student Affairs.

Disabilities Accommodations
The Americans with Disabilities Act (ADA) is a federal anti-discrimination statute that provides comprehensive civil rights protection for persons with disabilities. Among other things, this legislation requires that all students with disabilities be guaranteed a learning environment that provides for reasonable accommodation of their disabilities. If you believe you have a disability requiring an accommodation, please call or visit Disability Services at (361) 825-5816 in Driftwood 101.

If you are a returning veteran and are experiencing cognitive and/or physical access issues in the classroom or on campus, please contact the Disability Services office for assistance at (361) 825-5816.

References:
## Tentative Agenda

<table>
<thead>
<tr>
<th>DATE</th>
<th>ACTIVITY</th>
<th>RESOURCE</th>
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<tbody>
<tr>
<td>August</td>
<td></td>
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<tr>
<td>W 24</td>
<td>Review Syllabus; Course Overview</td>
<td>Always refer to the information Blackboard</td>
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<tr>
<td>M 29</td>
<td>Review: DNA &amp; Cell Division</td>
<td>Chapter 2</td>
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<tr>
<td>W 31</td>
<td>Review: Gametogenesis &amp; Fertilization</td>
<td>Chapter 2</td>
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<tr>
<td>September</td>
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<tr>
<td>M 5</td>
<td>Holiday</td>
<td></td>
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</table>
| W 7        | Homework Due: Sexual orientation,  Fraternal birth order, and the maternal immune hypothesis: A review  
Chromosome Analysis  
Chromosome Abnormalities | Chapter 3  
Non-Invasive Prenatal Genetic Diagnosis |
| M 12       | Imprinting  
Exercise 1: pairs                                           | Chapter 3  
Non-Invasive Prenatal Genetic Diagnosis |
| W 14       | Graduate Presentation: Techniques  
Presentation 1: Savior Siblings                                      | Chapter 4                                                                |
| M 19       | Graduate Presentation: Techniques  
Presentation 2: Designer Babies                                        | Chapter 4                                                                |
| W 21       | TEST 1                                                                   | Chapters 2, 3 & 4; PPTs; handouts; Non-Invasive Prenatal Genetic Diagnosis (NIPD); Graduate Presentations; Presentations 1 & 2 |
| M 26       | Trisomies                                                                | Chapter 6                                                                |
| W 28       | Trisomies  
Presentation 3: Collection of DNA Data from Military Personnel  
Exercise 2: pairs                                           | Chapter 6  
Consumers’ Views of Direct-to-Consumer Genetic Information |
|            |                                                                           |                                                                          |
| October    |                                                                           |                                                                          |
| M 3        | Sex Chromosome Disorders  
Mendelian Inheritance                                                       | Chapter 6  
Chapter 7                                                             |
| W 5        | Homework Due: The Pathophysiology of Fragile X Syndrome  
Mendelian Inheritance  
Presentation 4: Phenylketonuria                                       | Chapter 7                                                                |
| M 10       | Hemoglobin biochemistry  
Presentation 5: Sickle cell disease/anemia                                | Chapter 7                                                                |
| W 12       | Presentation 6: Thalassemias  
Presentation 7: Cystic Fibrosis                                            | Chapter 7                                                                |
| M 17       | Pedigree Patterns  
Presentation 8: Diabetes mellitus                                          | Chapter 7                                                                |
<table>
<thead>
<tr>
<th>Week</th>
<th>Assignment/Activity</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>W 19</td>
<td><strong>TEST 2</strong></td>
<td>Chapters 6 &amp; 7; PPTs; handouts; Presentations 3, 4, 5, 6, 7, 8; Consumers’ Views of Direct-to-Consumer Genetic Information</td>
</tr>
<tr>
<td>M 24</td>
<td>Video Reflection: Genetically Correct: Ensuring Perfect Babies 54 m</td>
<td>Chapter 8 To know or not to know: A review of behaviour and suicidal ideation in preclinical Huntington’s disease</td>
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<tr>
<td>W 26</td>
<td>Genes and Environment <strong>Exercise 3: pairs</strong></td>
<td>Chapter 8 To know or not to know: A review of behaviour and suicidal ideation in preclinical Huntington’s disease</td>
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<tr>
<td>M 31</td>
<td>Cancer Genetics <strong>Presentation 10: Huntington’s Disease</strong></td>
<td>Chapter 16</td>
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<td>November</td>
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<tr>
<td>W 2</td>
<td>Homework Due: <strong>Pharmacogenomics &amp; Individualized Drug Therapy</strong> Cancer Genetics</td>
<td>Chapter 16</td>
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<tr>
<td>M 7</td>
<td>Pharmacogenomics</td>
<td>Chapter 17 &amp; 18</td>
</tr>
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<td>W 9</td>
<td>Pharmacogenomics</td>
<td>Chapter 17 &amp; 18</td>
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<tr>
<td>M 14</td>
<td>Pharmacogenomics <strong>Presentation 12: Amyotrophic Lateral Sclerosis Presentation 13: Fetal Testing</strong></td>
<td>Chapter 17 &amp; 18</td>
</tr>
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<td>W 16</td>
<td>Homework Due: <strong>Defining the Cancer Master Switch</strong> Cancer Genetics</td>
<td>Chapter 16</td>
</tr>
<tr>
<td>M 21</td>
<td><strong>TEST 3</strong></td>
<td>Chapters 8, 16, 17 &amp; 18; PPTs; handouts; To know or not to know: A review of behaviour and suicidal ideation in preclinical Huntington’s disease; Presentations 9 &amp; 10</td>
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<tr>
<td>W 23</td>
<td>Holiday</td>
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<tr>
<td>M 28</td>
<td>Graduate Presentation: GINA Graduate Presentation: Sexual Orientation</td>
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<tr>
<td>W 30</td>
<td><strong>Exercise 4: in groups of 3 students</strong></td>
<td>Should selecting saviour siblings be banned?</td>
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<td>December</td>
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<tr>
<td>M 5</td>
<td>Homework Due: <strong>Preimplantation Genetic Diagnosis</strong> Pre-natal Diagnosis</td>
<td>Chapter 15</td>
</tr>
<tr>
<td>W 11 ??</td>
<td><strong>Final Exam 1:45-4:45 pm</strong></td>
<td>Comprehensive</td>
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