Syllabus

Seminar: Medical Cytogenetics

BIOL 281, SEC 001, Fall 2012

Instructor: Heather Gladwin, M.S.,CG (ASCP)cm
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Course Description

This course will introduce you to the field of human cytogenetics and its application to medicine. Topics covered will include: chromosome structure and function, cell culture and harvest, chromosome banding and staining, chromosome identification, cytogenetics nomenclature, chromosome abnormalities and aberrations, chromosomal syndromes, and Fluorescence in situ Hybridization techniques.

Reading Materials

I will post links to relevant references for each topic covered in this course. You will need to read that information in order to answer questions posted in the Discussion Forum.

Course Organization

We will not meet face-to-face during this course. All communication will be conducted online. There will be no exams. You will be evaluated on your participation in discussion forums and analysis of three case studies. Details about each of these will be posted on the prep page for the week.

I will be available through email for questions/concerns. Please allow 24 hours for a response.

The course is organized into weeks as general categories and subjects as more specific categories starting with the week of 10 September and ending with the week of 3 December. Our week will start on Monday, so I will post the new content no later than Sunday evening. The course is organized around the Preparation Pages model (or prep page for short). Each week there will be a new general prep page with links to required readings, activities, and discussion forum questions. The general prep pages are titled Week 1 through Week 13. Each prep page is divided into Announcements, Overview, Learning Objectives, Required Activities, and Additional References. Clicking on the page title (e.g., Week 1) will show you the exact page with the week's subject pages also included in a separate linked list at the bottom of the page. The first page you see when you visit this site will be the most recent prep page. However, all preparation pages will be available throughout the semester.
You will be expected to read information on the linked sites and answer questions posted in the Discussion Forum each week. Assignments are due no later than 5 pm on Saturday of each week. Assignments posted later than the due date will not receive credit. Each week’s assignment will be listed in that week’s prep page, with a link to the discussion forum to post in.

I will respond to each individual discussion forum post during the first two weeks so that you obtain a better understanding of what is expected. After that, I will only post general comments to the specific discussion forum topic. Please review my comments closely during the first several weeks.

You are expected to be able to use web browsers to access various sites and conduct activities on those sites. Several sites require that your browser be Java enabled and of course the speed of your processor and internet connection will influence the quality of your interactions with the web site.

**Announcements:** Any last minute information (e.g., changes in the schedule) or instructions will be posted here.

**Overview:** A general overview will appear at the top of every prep page.

**Learning Objectives:** This is what you should accomplish for this specific subject.

**Required Activities:** Links to the activities to be conducted during this week will be posted here. Please complete the week's activity and post your response no later than Saturday at 5 PM.

### Grading

#### Scheme

Discussion forum posts and reports will be assigned numerical values of 0, 1, or 2. 0 points is unacceptable, 1 point is acceptable, and 2 points is very good. Final grades for discussion forum posts and case study reports will be calculated through use of an average value.

Final course grades will be calculated according to the following weights:

<table>
<thead>
<tr>
<th>Weight</th>
<th>Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>70%</td>
<td>Contributions to Discussion Forums</td>
</tr>
<tr>
<td>30%</td>
<td>Case Study Reports</td>
</tr>
</tbody>
</table>

#### Scale

Grades will **not** be calculated on a curve. Therefore, you will **not** be competing with your classmates. Your final course grade will be determined as outlined above and assigned a letter grade according to the following scale:
Letter Grade Average Score

<table>
<thead>
<tr>
<th>Grade</th>
<th>Average Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>&gt;1.8</td>
</tr>
<tr>
<td>A-</td>
<td>1.79 - 1.66</td>
</tr>
<tr>
<td>B+</td>
<td>1.65 - 1.52</td>
</tr>
<tr>
<td>B</td>
<td>1.51 - 1.38</td>
</tr>
<tr>
<td>B-</td>
<td>1.37 - 1.24</td>
</tr>
<tr>
<td>C+</td>
<td>1.23 - 1.12</td>
</tr>
<tr>
<td>C</td>
<td>1.11 - 1.00</td>
</tr>
<tr>
<td>C-</td>
<td>0.99 - 0.86</td>
</tr>
<tr>
<td>D+</td>
<td>0.85 - 0.72</td>
</tr>
<tr>
<td>D</td>
<td>0.71 - 0.58</td>
</tr>
<tr>
<td>F</td>
<td>&lt;0.58</td>
</tr>
</tbody>
</table>

Note: These ranges might be revised down, if necessary. They will not be raised. The minimum score for a given letter grade is guaranteed.

Academic Conduct

You are expected to uphold the highest principles of academic honesty as defined in the academic honesty policy section of the College's student handbook:

"Academic Honesty, a necessary foundation of a learning community is expected of all students. Violations are unacceptable and are subject to academic penalties including failure of a course. A record of the violation is submitted to Academic Affairs; repeated violations may result in suspension or dismissal from the college. Violations of academic honesty include cheating on examinations, plagiarism, and submission of papers for credit in two or more courses."
(Westfield State University, Student Handbook, 2010-2011)

You are responsible for familiarizing yourself with the academic policies and procedures of the College.

Participation

Posting to discussion forums and submitting case study reports by the deadlines is expected. Work submitted after the deadline will not be accepted.

Learning Objectives

Knowledge
Learn the major clinical indications for cytogenetic testing and the clinical implications of results
Understand the principles of cytogenetics as applied to diagnostic testing
1. Classification of genetic disorders
2. Disciplines within Clinical Genetics and integration of genetic diagnostic services with other healthcare services (Clinical Genetics, Prenatal Diagnosis, Infertility, Cancer Cytogenetics)
3. Chromosomal basis of heredity
4. Chromosome structure
5. Methods for chromosome study (G-bandng, FISH)

Become familiar with major techniques used in clinical cytogenetics, including culture of peripheral blood, fibroblasts, amniotic fluid, chorionic villus, products of conception, bone marrow, lymph nodes and solid tumor specimens, chromosome banding, karyotyping, and FISH

Understand issues related to the interpretation of normal chromosomal variation, mosaicism, aneuploidy and other chromosomal rearrangements
1. ISCN nomenclature
2. Abnormalities of chromosome number (monosomy, trisomy, triploidy)
3. Partial aneuploidy, microdeletion/contiguous gene syndromes, abnormal chromosomes

Become familiar with major syndromes/pathology caused by chromosomal imbalance
1. Autosomal trisomy syndromes
2. Sex chromosome aneuploidy
3. Common microdeletion syndromes

Understand basic concepts and applications of molecular cytogenetic and molecular genetic techniques to routine cytogenetic analysis
1. Limitations of routine chromosome analysis
2. Microdeletion syndromes
3. Single gene disorders

Skills

The student will demonstrate knowledge of how to obtain reliable information from a variety of sources (e.g., web, popular media, and scientific publications).

Assessment: Knowledge of how to obtain reliable information from a variety of sources (e.g., web, popular media, and scientific publications) will be assessed through use of case study reports.

The student will demonstrate knowledge of how to communicate the results of a scientific investigation.

Assessment: Knowledge of how to communicate the results of a scientific investigation will be assessed through use of case study reports.

Special Needs
Any student enrolled in this course who has a disability that prevents the fullest expression of their abilities should contact me as soon as possible, so that we can discuss accommodations.

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