SYLLABUS
Human Genetics 2032: Genetic Techniques

Lecture: 1-1:55 pm Tues, A215 Crabtree Hall
Laboratory: 1:00-3:55 pm Wed, A720 Crabtree Hall

Course Director: Quasar Padiath, MBBS, Ph.D.
Office: 3135 Parran Hall (412) 624-7203 qpadiath@pitt.edu
Office Hours: By appointment via email; or immediately after class

Lab Instructor: Dale Lewis, B.S., CG(ASCP)CM (412) 624-5358 dwl2@pitt.edu
Office Hours: Immediately after class or by appointment

Course Description:
Students participate in laboratory exercises to become acquainted with cytogenetics laboratory procedures including cell culture, chromosome preparation, chromosome banding, and karyotyping. Chromosome analysis and karyotype interpretation are practiced.

Purpose of Course:
The purpose of this course is to familiarize graduate students and fellows in Human Genetics and Genetic Counseling with genetic techniques “hands-on” using a variety of genetic techniques with a clinical perspective to facilitate application to and education of future clientele. Students participate in laboratory exercises to become acquainted with cytogenetics laboratory procedures including cell culture, chromosome preparation, chromosome banding, and karyotyping. Chromosome analysis and karyotype interpretation are practiced. Classical and molecular cytogenetic methods covered include cell culture, harvesting, slide making, staining, chromosome banding, chromosome analysis, FISH, karyotyping, and karyotype interpretation and reporting. Molecular genetics techniques include PCR, RNA isolation, Reverse Transcription, gene expression using QRT-PCR, and (RFLP) SNP analysis using restriction enzymes, Microarray, NIPT, preimplantation diagnosis/screening, Sanger Sequencing and Next-Generation Sequencing (NGS) as applied to problems in human genetics, including diagnosis, family studies and mutation analysis. Research applications are also discussed. Co-requisite: HuGen 2031.

Learning Objectives
Upon completion of this course, students will be able to:
• Compare and contrast the strengths and limitations of the various cytogenetic and molecular genetic assays
• Discuss a variety of genetic techniques from a clinical perspective
• Interpret a cytogenetics or molecular diagnostics report
• Critique published cytogenetics literature
• Apply principles of effective written and oral communication to genetics topics.
• Apply cytogenetic and molecular genetics techniques

Textbooks:
There are no textbooks.
Course website:
All readings and course material will be found on Courseweb (http://courseweb.pitt.edu).

Grading Scale:
88-100 A
85-87.9 A-
80-84.9 B+
70-79.9 B
<70  C

Student Performance Evaluation
Student grades are based on
40% Clinical reports
20% Classroom presentations-verbal critique of a self-chosen paper and lab experiment review
20% Class participation
20% Lab notebooks

Academic Integrity:
All students are expected to adhere to the school’s standards of academic honesty. Any work submitted by a student for evaluation must represent his/her own intellectual contribution and efforts. The Graduate School of Public Health’s policy on academic integrity, approved by EPCC on 10/14/08, which is based on the University policy, is available online in the Pitt Public Health Academic Handbook (www.publichealth.pitt.edu/home/academics/academic-requirements). The policy includes obligations for faculty and students, procedures for adjudicating violations, and other critical information. Please take the time to read this policy.

Students committing acts of academic dishonesty, including plagiarism, unauthorized collaboration on assignments, cheating on exams, misrepresentation of data, and facilitating dishonesty by others, will receive sanctions appropriate to the violation(s) committed. Sanctions include, but are not limited to, reduction of a grade for an assignment or a course, failure of a course, and dismissal from the school.

All student violations of academic integrity must be documented by the appropriate faculty member; this documentation will be kept in a confidential student file maintained by the Office of Student Affairs. If a sanction for a violation is agreed upon by the student and instructor, the record of this agreement will be expunged from the student file upon the student’s graduation. If the case is referred to the Pitt Public Health Academic Integrity Hearing Board, a record will remain in the student’s permanent file.

Plagiarism:
University of Pittsburgh policy: “Integrity of the academic process requires that credit be given where credit is due. Accordingly, it is unethical to present as one's own work the ideas, representations, words of another, or to permit another to present one's own work without customary and proper acknowledgement of sources.

A student has an obligation to exhibit honesty and to respect the ethical standards of the profession in carrying out his or her academic assignments. Without limiting the application of this principle, a student may be found to have violated this obligation if he or she:*
10. Presents as one's own, for academic evaluation, the ideas, representations, or words of another person or persons without customary and proper acknowledgment of sources.
11. Submits the work of another person in a manner which represents the work to be one's own.”
Therefore, you must clearly indicate which thoughts are yours and which thoughts belong to others by citing your sources. If you are uncertain, please contact the instructor. Plagiarism detection software will be used in this course. If plagiarism is detected, you will automatically receive a grade of zero for that assignment.

Disabilities:
If you have a disability for which you are or may be requesting an accommodation, you are encouraged to contact both your instructor and Disability Resources and Services, 140 William Pitt Union, 412-648-7890 or 412-383-7355 (TTY) as early as possible in the term.

Course Agenda

Guest lecturers and consultants for the course are listed below and noted on the course schedule by their initials.

Guest Lecturers:
Svetlana Yatsenko, M.D.  Microarray analysis, NIPT, PGS/PGD
Dan Bellisimo, Ph.D.  DNA Sequencing

Consultant:
Susanne M. Gollin, Ph.D.
Urvashi Surti, Ph.D.

<table>
<thead>
<tr>
<th>Lectures/ Other (10-10:55 AM Tues, A215 Crabtree Hall)</th>
<th>Laboratory Sessions (1:00-3:55 PM Wed, A720 Crabtree Hall, or as announced)</th>
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<tbody>
<tr>
<td>Tues, 1/10/17  Introduction to cell culture-DWL</td>
<td>Wed, 1/4/17  Intro to HuGen 2032-QSP/DWL  Bloodborne Pathogens and Chemical Hygiene Safety Training -DWL  Electronic lab notebook introduction-DWL</td>
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<td>Tues, 1/17/17  The cytogenetic harvest-DWL</td>
<td>Wed, 1/11/17  Tissues and amniotic fluid cell culture-DWL  **Blood draw and culture set up will be on Saturday 1/14/17 Time-TBD-QSP/DWL</td>
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<td>Tues, 1/24/17  Introduction to Chromosome Banding and karyotype analysis-DWL</td>
<td>Wed, 1/25/17 (A720 Crabtree Hall)  Trypsin-Giemsa banding-DWL  Selection of abnormal karyotype cases-DWL</td>
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<td>Tues, 1/31/17  Report writing and review of sample cases and reports-DWL  Introduction to Fluorescence In- Situ hybridization-DWL</td>
<td>Wed, 2/1/17 (A720 Crabtree Hall)  FISH for the X and Y chromosomes-DWL</td>
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<td>Tues, 2/7/17</td>
<td>DNA isolation Spectrophotometry Gel Electrophoresis PCR-DWL</td>
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<td>Tues, 2/14/17</td>
<td>Non-Invasive Prenatal Testing (NIPT) PGS/PGD Applications, Methods and Mishaps SY</td>
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<td>Tues, 2/21/17</td>
<td>Introduction to array CGH and SNP microarray-QSP</td>
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<td>Tues, 2/28/17</td>
<td>Introduction to Sanger Sequencing-QSP How to choose articles for case discussions-QSP</td>
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<td>Tues, 3/7/17</td>
<td>SPRING RECESS</td>
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<td>Tues, 3/14/17</td>
<td>Negotiation for articles-QSP</td>
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<td>Tues, 3/21/17</td>
<td>Next Generation Sequencing-QSP</td>
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<td>Tues, 4/4/17</td>
<td>Intro to Reverse transcription and quantitative (QRT-PCR) and DNA dosage analyses-DWL</td>
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<td>Tues, 4/11/17</td>
<td>Introduction to RFLP and restriction enzymes-DWL</td>
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<td>Tues, 4/18/17</td>
<td>Quantitative PCR Analysis-DWL</td>
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<td>Tues, 4/25/17</td>
<td>Student case discussions-QSP</td>
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