

SYLLABUS
Human Genetics 2032: Genetic Techniques 2022
The instructor reserves the right to make changes to the schedule.

Lecture: 10-10:55 am Tues, 3121C Pitt Public Health (Formerly Parran Hall)

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



Course Director: Quasar Padiath (QSP), MBBS, Ph.D.

Assistant Director: Guillermo Rodreguez Bey (GRB), Ph.D.



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. Specifically, students become acquainted with a variety of techniques including cytogenetics and molecular genetics laboratory procedures and techniques including cell culture, chromosome preparation, chromosome banding, karyotyping, PCR, RNA isolation, Reverse Transcription, gene expression using QRT-PCR, (RFLP) SNP analysis using restriction enzymes, Microarray, NIPT, preimplantation diagnosis/screening, Sanger Sequencing and Next-Generation Sequencing (NGS). These genetics techniques are applied to problems in human genetics, such as diagnosis, determining prognosis, family studies and mutation analysis; and used to determination test results, interpretation and reporting findings. Research applications are also discussed. Co-requisite: HuGen 2060: Chromosomes – Structure and Function.

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 genetics 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+
75-79.9 B
70-74.9 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 July 2013, which is based on the University policy, is available online in the Pitt Public Health Academic Handbook (<http://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.

The appropriate faculty member must document all student violations of academic integrity; 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."

Source: <http://www.bc.pitt.edu/policies/policy/02/02-03-02.html>

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.

Accommodation for Students with 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.

Sexual Misconduct, Required Reporting and Title IX Statement

The University is committed to combatting sexual misconduct. As a result, you should know that University faculty and staff members are required to report any instances of sexual misconduct, including harassment and sexual violence, to the University's Title IX office so that the victim may be provided appropriate resources and support options. What this means is that as your professor, I am required to report any incidents of sexual misconduct that are directly reported to me, or of which I am somehow made aware.

There are two important exceptions to this requirement about which you should be aware: A list of the designated University employees who, as counselors and medical professionals, do not have this reporting responsibility and can maintain confidentiality, can be found here: www.titleix.pitt.edu/report/confidentiality

An important exception to the reporting requirement exists for academic work. Disclosures about sexual misconduct that are shared as part of an academic project, classroom discussion, or course assignment, are not required to be disclosed to the University's Title IX office.

If you are the victim of sexual misconduct, Pitt encourages you to reach out to these resources:

- Title IX Office: 412-648-7860
- SHARE @ the University Counseling Center: 412-648-7930 (8:30 A.M. TO 5 P.M. M-F) and 412-648-7856 (AFTER BUSINESS HOURS)

If you have a safety concern, please contact the University of Pittsburgh Police, 412-624-2121. Other reporting information is available here: www.titleix.pitt.edu/report-0

Statement from the Department of Gender, Sexuality, and Women's Studies

[This statement was developed by Katie Pope, Title IX Coordinator, in conjunction with GSWS instructors.]

Diversity Statement

The University of Pittsburgh Graduate School of Public Health considers the diversity of its students, faculty, and staff to be a strength and critical to its educational mission. Pitt Public Health is committed to creating and fostering inclusive learning environments that value human dignity and equity. Every member of our community is expected to be respectful of the individual perspectives, experiences, behaviors, worldviews, and backgrounds of others. While intellectual disagreement may be constructive, no derogatory statements, or demeaning or discriminatory behavior will be permitted.

If you feel uncomfortable or would like to discuss a situation, please contact any of the following:

- the course instructor;
- the Pitt Public Health Associate Dean for Diversity at 412-624-3506 or nam137@pitt.edu;
- the University's Office of Diversity and Inclusion at 412-648-7860 or <https://www.diversity.pitt.edu/make-report/report-form> (anonymous reporting form).

Course Agenda

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

Course Director : Quasar Padiath (QP), M.B.B.S., Ph.D.
 Assistant Director: Guillermo Rodreguez Bey (GRB), Ph.D.
 Guests Lecturers: Mahmoud Aarabi (MA), M.D., Ph.D. Cytogenetic techniques
 Svetlana Yatsenko (SY), M.D. (SY), Microarray analysis, NIPT
 Dan Bellissimo (DB), Ph.D. (DB) DNA Sequencing
 Ansuman Chattopadhyay (AC), PhD – RNA sequencing analysis

Lectures/ Other 10-10:55 AM (Tuesdays) 3121C, Pitt Public Health	Laboratory Sessions 1:00-3:55 PM (Wednesdays) A720 Crabtree Hall, or as announced
Tues, 1/11/22 Intro to HuGen 2032-QSP Electronic lab notebook introduction Introduction to cell culture (Virtual)	Wed, 1/12/22 (A720 Crabtree Hall) Blood borne Pathogens and Chemical Hygiene Safety Training (Virtual)
Tues, 1/18/22 The cytogenetic harvest and chromosome analysis - MA (Virtual)	Wed, 1/19/22 Introduction to Chromosome Banding and Fluorescence <i>In- Situ</i> hybridization (FISH)-MA (Virtual)
Tues, 1/25/22 Report writing and review of sample cases and reports-MA FISH and G Banding analysis- MA (Virtual)	Wed, 1/26/22 Harvest blood lymphocyte chromosomes and slide making-GRB/QSP
Tues, 2/1/22 Trypsin-Giemsa banding-GRB/QSP	Wed, 2/2/22 Cell culture GRB/QSP
Tues, 2/8/22 DNA isolation Spectrophotometry Gel Electrophoresis PCR-QSP	Wed, 2/9/22 Isolate DNA from blood. Evaluate purity and concentration of DNA. Set up PCR reaction- GRB/QSP
Tues, 2/15/22 Introduction to Sanger Sequencing-QSP How to choose articles for case discussions- QSP	Wed, 2/16/22 Run Gel of PCR products- GRB/QSP Prepare products for Operon sequencing PCR for RFLP samples

Tues, 2/22/22 Non-Invasive Prenatal Testing (NIPT) PGS/PGD Applications, Methods and Mishaps-SY	Wed, 2/23/22 Sanger sequence analysis- GRB/QSP QSP-Lab
Tues, 3/1/22 Introduction to array CGH and SNP microarray-QSP	Wed, 3/2/22 Microarray analysis, interpretation and reporting SY
Tues, 3/8/22 SPRING RECESS	Wed, 3/9/22 SPRING RECESS
Tues, 3/15/22 Next Generation Sequencing-QSP	Wed, 3/16/22 NGS analysis, interpretation, and report writing-DB
Tues, 3/22/22 RNA isolation RNA gel-QSP	Wed, 3/23/22 Isolate RNA from cells. Evaluate purity and concentration by spectrophotometry and gel electrophoresis- GRB/QSP
Tues, 3/29/22 RNA Sequencing and analysis- AC	Wed, 3/30/22 NGS analysis resources- AC
Tues, 4/5/22 Intro to Reverse transcription and quantitative (QRT-PCR) and DNA dosage analyses-QSP	Wed, 4/6/22 Produce cDNA by reverse transcription (RT) for gene expression - GRB/QSP Run gel on PCR product
Tues, 4/12/22 Introduction to RFLP and restriction enzymes-QSP	Wed, 4/13/22 Set up QRT-PCR plates Set up Restriction Enzyme digestions
Tues, 4/19/22 Quantitative PCR Analysis-QSP	Wed, 4/20/22 Run gel on RE digested product. Interpret RE digest findings- GRB/QSP Review molecular techniques, experiments and findings- GRB/QSP
Tues, 4/26/22- Student case discussions-QSP	Wed, 4/27/22 - Student case discussions-QSP