

**Graduate School of Public Health
Department of Human Genetics**

**HUGEN 2074
Genome Bioinformatics Capstone**

Fall 2021

Tuesdays and Thursdays · 9:00 AM–11:50 AM
3118 Public Health
6 Credits

COURSE DESCRIPTION

The capstone course provides students in the Master of Science in Genome Bioinformatics with the opportunity to expand their repertoire of analysis and professional skills and communicate results of their internship to faculty and fellow students. As part of this course, students will develop final analyses based on their internship data or use other (faculty-provided) data if the results of their summer internship project are not appropriate thesis material. The capstone course will ensure that the thesis project demonstrates the student's competency in genome bioinformatics as well as communication skills in general.

COURSE GOALS

Upon completion of this course, the student will be able to:

- Gather requirements for a research question of interest and translate them into clear, testable statistical hypotheses
- Develop a data analysis plan to process the applicable data, to test hypotheses that address the question of interest, and use bioinformatics tools to contextualize results
- Communicate orally and visually the analysis plan and analysis results through the description of analytic strategies and the summarization of results
- Write Python code to manipulate data
- Describe relational database and write SQL statements to query databases
- Describe basic machine learning tools and issues in machine learning methods
- Describe ethical issues in data integrity and inclusivity

COURSE PREREQUISITES

Completion of HUGEN 2071, HUGEN 2072 and HUGEN 2073 and summer internship as student in Master of Science in Genome Bioinformatics program.

FACULTY

Course Director

Ryan L. Minster, Ph.D., M.S.I.S.

3118 Public Health

412-624-6928

rminster@pitt.edu

Office hours available upon request

CLASS ACTIVITIES

The course will be in two three-hour sessions (usually) per week. Each session will be divided into a 1¾ hour lecture, a 15 min break, and a 50 min activity. The lectures will be didactic but interactive reviews of topic relevant to genome bioinformatics and the professions that employ it. The activities will include group discussions, presentations, peer critique, role playing, and guest presentations. Approximately ten guest presenters will present on their career paths, job responsibilities and areas of research or business.

Potential Guest Presenters

Karen Cuenco, Senior Specialist, Integration & Quantitative Science, Gates Foundation

Mike Talkowsky, Center for Genomic Medicine, Massachusetts General Hospital

Xiaojing Wang, Associate Director of Metrics, Analytics & Statistics Services at ConvaTec

Purnima Sundar, Principal Scientist, Pfizer

Anthony Roscoe, Director of Enterprise Data Governance & Quality, Highmark Health

Wan Zhu, Sr Machine Learning (ML) Engineer, Paypal; formerly ML Engineer, Google

Celeste Shelton Ohlsen, Director of Clinical Genomics, Ariel Precision Medicine

Brandon Michael Blobner, BlueSphere Bio

Emily Russell, New Hire

EVALUATION AND GRADING

Evaluation will be based on the following components:

Research Question

Students will formulate a research question for their thesis project.

Analysis Plan

Students will develop a written analysis plan for their thesis project and will present the analysis plan to the class.

Descriptive Statistics and Visualizations

Students will generate descriptive statistics and visualizations as appropriate to their analysis plan and project and will submit them in written form and present them to the class.

Analysis Statistics and Visualizations

Students will perform statistical analyses (hypothesis testing or hypothesis generating) and concomitant visualizations as appropriate to their analysis plan and project and will submit them in written form and present them to the class.

Final Analysis Methods and Results

Students will submit a written description of their final thesis project methods and results and will present them to the class.

Python, SQL, and Machine Learning Projects

Students will be given one small coding project in each of basic Python coding, SQL querying and machine learning to assess competency in those domains.

Case Study Engagement (3)

There will be three case studies examined during the course. For each case study, students will submit two engagement questions or comments on the case study to facilitate discussion.

Grading

Grading will be based on specifications grading, a competency-based grading model wherein all assignments are graded as pass/not-pass based specific criteria that align with the course learning objectives. The focus is on demonstrating mastery of the course objectives rather than earning a particular letter grade.

Each assignment is graded as pass/not-pass and may be resubmitted (after receiving feedback) as many as two times within two weeks of the original due date to achieve a pass.

To receive an **H** in this course, all assignments must be passed with only one revision within two weeks of the original due date.

To receive an **S** in this course, all assignments must be passed with no more than two revisions within two weeks of the original due date.

If any assignment is not passed after two revisions within two weeks of the original due date, the assigned course grade will be a **U**.

CANVAS INSTRUCTION

This course will use the University's Canvas site (canvas.pitt.edu). Each lecture will be accompanied by supporting material and further reading, all of which will be made available around the time of the lecture. It is the student's responsibility to check for, and read, this material. The instructors will use Canvas as the primary means of communicating with the students, who are expected to check the site on a regular basis throughout the semester.

Accessibility

Ensuring an accessible and pleasant experience to all users, regardless of disability, is a key focus of Canvas. The Canvas platform was built using the most modern HTML and CSS technologies, and is committed to W3C's Web Accessibility Initiative and [§508](#) guidelines.

COURSE MATERIALS

Required Software (All available free online)

Web Browser

R
r-project.org

R Studio
rstudio.com

Pulse Secure
pulsesecure.net

Required Readings (All available free online through the University of Pittsburgh)

Selected papers from the current literature.

SCHEDULE

Date	Lecture	Activity / Assessment
Tue Aug 31	Introduction & Design	Discuss Internship Experiences
Thu Sep 2	Developing Research Questions	Guest Presentation
Tue Sep 7	Advanced Genetic Data Structures & Analysis	Discuss Potential Research Questions
Thu Sep 9	Developing Analysis Plans	Guest Presentation
<i>Tue Sep 14</i>	<i>No Class</i>	
<i>Thu Sep 16</i>	<i>No Class</i>	
Tue Sep 21	Advanced Unix	
Thu Sep 23	Advanced Topics in DNA Sequencing	
Tue Sep 28	Advanced Topics in RNA Sequencing	Case Study in Genetic Analysis
Thu Sep 30	Public Speaking Fundamentals	Guest Presentation
Tue Oct 5	Public Speaking with Visuals	Present Analysis Plans
Thu Oct 7	Conflict Resolution and Negotiation	Guest Presentation
Tue Oct 12	Project Requirements Gathering	
Thu Oct 14	Project Management	Present Descriptive Statistics
Tue Oct 19	Project Budgeting	Role Play Conflict Resolution
Thu Oct 21	Introduction to Python	
Tue Oct 26	Advanced Python	Role Play Requirements Gathering
Thu Oct 28	Relational Database Design	Guest Presentation
<i>Tue Nov 2</i>	<i>No Class · Election Day</i>	
Thu Nov 4	Structured Query Language (SQL)	Present Analysis Progress
Tue Nov 9	Introduction to Machine Learning 1	Role Play Interviewing
Thu Nov 11	Introduction to Machine Learning 2	Guest Presentation
Tue Nov 16	Advanced Visualizations 1	Case Study in Machine Learning
Thu Nov 18	Advanced Visualizations 2	Guest Presentation
<i>Tue Nov 23</i>	<i>No Class · Thanksgiving Recess</i>	
<i>Thu Nov 25</i>	<i>No Class · Thanksgiving Recess</i>	
Tue Nov 30	Advanced Annotation & Contextualization 1	
Thu Dec 2	Advanced Annotation & Contextualization 2	Present Visualization Project
Tue Dec 7	Data Ethics	Case Study in Data Ethics
Thu Dec 9	Data Ethics	Guest Presentation
Tue Dec 14	Comprehensive Exam	
Thu Dec 16	Student-Chosen Topic	Present Final Analysis Results

ACADEMIC POLICIES

Academic Integrity

All students are expected to adhere to the school's standards of academic honesty. Cheating/plagiarism will not be tolerated. The Graduate School of Public Health's [policy on academic integrity](#), which is based on the University policy, is available online in the Pitt Public Health Academic Handbook. The policy includes obligations for faculty and students, procedures for adjudicating violations, and other critical information. Please take the time to read this policy.

Plagiarism

University 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

To avoid plagiarism, you must give "customary and proper acknowledgment of sources" by appropriately and clearly identifying which thoughts are yours and which are others, and appropriately citing your sources.

Sophisticated plagiarism detection software will be used in this course. If plagiarism is detected, you will automatically receive a grade of zero for that assignment and the incident will be reported, as required, to your Dean.

Covid-19 & Public Health

In the midst of this pandemic, it is extremely important that you abide by public health regulations and University of Pittsburgh health standards and guidelines. While in class, at a minimum, this means you must wear a face covering and comply with physical distancing requirements; other requirements may be added by the University during the semester. These rules have been developed to protect the health and safety of all community members. Failure to comply with these requirements will result in you not being permitted to attend class in person and could result in a Student Conduct violation. For the most up-to-date information and guidance, please visit coronavirus.pitt.edu and check your Pitt email for updates before each class.

Course Recording

This class or portions of this class will be recorded by the instructors for educational purposes. These recordings will be shared only with students enrolled in the course via Canvas and will be deleted at the end of the course.

To ensure the free and open discussion of ideas, students may not record classroom lectures, discussion and/or activities without the advance written permission of the instructor, and any such recording properly approved in advance can be used solely for the student's own private use.

Copyright Notice

These materials may be protected by copyright. United States copyright law, 17 USC § 101, *et seq.*, in addition to University policy and procedures, prohibit unauthorized duplication or retransmission of course materials. See [Library of Congress Copyright Office](#) and the [University Copyright Policy](#).

Disability Resources

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 as early as possible in the term.

Sexual Misconduct, Required Reporting, & Title IX

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:

1. Professional, licensed counselors and pastoral counselors who provide mental-health counseling to members of the University community (and including those who act in that role under the supervision of a licensed counselor) are not required to report any information about an incident to the Title IX coordinator without a victim's permission.

Individuals who work or volunteer on-campus in the Student Health Service, including front desk staff and students, can generally talk to a victim without revealing any personally identifying information about an incident to the University. A victim can seek assistance and support from these individuals without triggering a University investigation that could reveal the victim's identity or that the victim has disclosed the incident, unless required by Pennsylvania law.

For additional information on confidentiality, please contact SHARE at the number below.

2. 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 AM–5:00 PM Mon–Fri) 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](#).

Diversity & Inclusivity

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 and promote social justice. 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 director or course instructor
- the Pitt Public Health Associate Dean for Diversity and Inclusion, Dr Tiffany Gary-Webb, at 412-624-3131 or tgary@pitt.edu
- the University's Office of Diversity and Inclusion at 412-648-7860 or at this [anonymous reporting form](#)