

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

**HUGEN 2073
Genomic Data Visualization and Integration**

Spring 2022

Tuesday · 10:30 AM–11:45 AM
Thursdays · 10:30 AM–11:45 AM

Break · 11:05 AM–11:10 AM

Public Health 1155
3 Credits

COURSE DESCRIPTION

This course will teach principles of data visualization and data visualizations that are specific to genetic and genomic analyses. It will also delve into the integration of data from multiple resources to appropriately annotate genetic associations with relevant information from a variety of repositories of genetic, genomic, and other “omic” data.

COURSE GOALS

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

- Interpret figures and analyze factors that affect their clarity, usefulness, and accessibility, with an emphasis on figures common in genetics and genomics
- Generate appropriate figures to support scientific claims while adhering to ethical standards for visualizations, with an emphasis on figures common in genetics and genomics
- Present visualizations effectively in written or oral form
- Identify important repositories of information relevant to genetic and genomic research questions and describe the available data contained therein.
- Annotate genes, genetic variation, and genomic regions with information from such repositories using local, computational cluster and web-based software interfaces.

COURSE PREREQUISITES

HUGEN 2022 · Human Population Genetics
BIOST 2041 · Introduction to Statistical Methods 1 (or an equivalent biostatistics course)

FACULTY

Hyun Jung (H.J.) Park, Ph.D.

3122 Public Health
412-383-0520
hyp15@pitt.edu
Office hours TBD and upon request

Jonathan M. Chernus, Ph.D.

3124 Public Health
412-723-7097
jonchernus@pitt.edu
Office hours TBD and upon request

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

3118 Public Health
412-624-6928
rminster@pitt.edu
Office hours TBD and upon request

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 (www.section508.gov) guidelines.

EVALUATION AND GRADING

Evaluation will be based on the following components:

Syllabus Review (1)

This assessment will consist of 15 questions about the syllabus. This assessment can be taken repeatedly until passed without costing mulligans (see below). *The initial deadline for this assignment is Thu Jan 13. It can be retaken an unlimited amount of times until passed up to Wed Jan 19 at 11:59 PM EST.*

R Coding Project (1)

This assessment will consist of a set coding tasks with R, aimed at preparing you to create visualizations using R. This assignment is considered passed if 80% coding tasks are successfully completed.

Five-Minute Papers (29)

At the end of each class session students will submit responses to a few questions about the session's content. If a student is unable to attend a class session and needs to leave early, they can submit their five-minute paper up to one week after the session (facilitated by viewing the recorded session via Canvas).

Visualization Critiques (2)

Students will submit a one-page critique of an assigned visualization (figure) from the literature based on the principles for good visualization taught in the class.

Course Projects (6)

There will be six course projects: three projects that will assess proficiency in designing a visualization, coding to produce it, and writing a description of it; and three projects that will assess proficiency with annotating and integrating primary association results with data from standing data repositories and describing the relationships between the primary findings and salient features from those repositories. Course projects will be graded as satisfactory or unsatisfactory based on meeting a specified set of requirements for each project when the project is assigned.

Late Policy & Revision Policy

Each student begins the term with four mulligans. Each mulligan can be used for a 48-hour extension on a project, an opportunity to redo the coding project, or an opportunity to revise a course project. You can use additional mulligans for further extensions on a project, so that you could spend two mulligans for a 96-hour extension.

Grading

Letter grades for the course are assigned based on the number of items in each component (coding project, critiques, and course projects) for which the student earns a 'satisfactory.'

Course Grade	D	C	B	A	A+	
Syllabus Review	80%	80%	80%	80%	80%	Each visualization critique,
R Coding Assignment	80%	80%	80%	80%	95%	five-minute paper,
Five-Minute Papers	17/29	20/29	23/29	26/29	29/29	& course project will be
Visualization Critiques	1/2	1/2	2/2	2/2	2/2	graded satisfactory/
Course Projects	3/6	4/6	5/6	6/6	6/6	unsatisfactory.

For example, to earn a B, a student must satisfactorily complete 80% of the syllabus review, 80% tasks in the R coding project, 23 of 29 five-minute papers, 2 of the 2 visualization critiques, and 5 of the 6 course projects.

Remember that unsatisfactory critiques and projects can be revised by using a mulligan.

COURSE MATERIALS

Required Software (All available free online)

Web Browser

R
r-project.org

R Studio
rstudio.com

Pulse Secure
pulsesecure.net

Required Textbook (Available free online)

Fundamentals of Data Visualization
by Claus O. Wilke
www.clauswilke.com/dataviz

Selected papers from the literature.

SCHEDULE

Date	Topic or Activity
Tue Jan 11	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapters 2–3 Lecture: Introduction; Elements of Visualization Due: Five-Minute Paper 1
Thu Jan 13	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapters 1, 4 Lecture: Introduction to Good Visualization Due: Five-Minute Paper 2 Due: Syllabus Review
Tue Jan 18	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapter 27 Lecture: Visualization with R · Data Frames & Base Graphics Due: Five-Minutes Paper 3
Wed Jan 19	Due: Critique 1
Thu Jan 20	Lecture: Visualization with R · ggplot2 Due: Five-Minutes Paper 4
Tue Jan 25	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapter 6 Lecture: Visualizing Amounts Due: Five-Minute Paper 5
Wed Jan 26	Due: R Coding Assignment
Thu Jan 27	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapters 7–9 Lecture: Visualizing Distributions Due: Five-Minute Paper 6
Tue Feb 1	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapter 12 Lecture: Visualizing Associations Due: Five-Minute Paper 7
Wed Feb 2	Due: Project 1

Date	Topic or Activity
Thu Feb 3	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapters 10–11 Lecture: Visualizing Proportions Due: Five-Minute Paper 8
Tue Feb 8	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapter 14 Lecture: Visualizing Trends Due: Five-Minute Paper 9
Wed Feb 9	Due: Critique 2
Thu Feb 10	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapter 16 Lecture: Visualizing Uncertainty Due: Five-Minute Paper 10
Tue Feb 15	Read: Saxena <i>et al.</i> <i>Neurocomputing</i> 2017 https://dx.doi.org/10.1016/j.neucom.2017.06.053 Lecture: Visualizing High Dimensions Due: Five-Minute Paper 11
Wed Feb 16	Due: Project 2
Thu Feb 17	Read: Nusrat <i>et al.</i> <i>Comput Graph Forum</i> 2019 https://dx.doi.org/10.1111/cgf.13727 Lecture: Visualization in Genetics & Genomics 1 Due: Five-Minute Paper 12
Tue Feb 22	Lecture: Visualization in Genetics & Genomics 2 Due: Five-Minute Paper 13
Thu Feb 24	Lecture: Ethical Issues in Visualization Due: Five-Minute Paper 14
Tue Mar 1	Lecture: Presenting Visualizations Due: Five-Minute Paper 15
Wed Mar 2	Due: Project 3
Thu Mar 3	Read: <i>Fundamentals of Data Visualization</i> · www.clauswilke.com/dataviz Chapter 28 Lecture: Visualization with Tableau & Python Due: Five-Minute Paper 16
Tue Mar 8	Lecture: No Class · Spring Break
Thu Mar 10	Lecture: No Class · Spring Break
Tue Mar 15	Lecture: Introduction to Annotation Due: Five-Minute Paper 17
Thu Mar 17	Lecture: SNV- & LD-Based Annotation

Date	Topic or Activity
	Due: Five-Minute Paper 18
Tue Mar 22	Lecture: Annotation & Interpretation Due: Five-Minute Paper 19
Wed Mar 23	Due: Project 4
Thu Mar 24	Lecture: UCSC Genome Browser Visualization & Tables Due: Five-Minute Paper 20
Tue Mar 29	Lecture: Constructing UCSC Genome Browser Custom Tracks Due: Five-Minute Paper 21
Thu Mar 31	Lecture: SQL & Querying UCSC Genome Browser Tables Due: Five-Minute Paper 22
Tue Apr 5	Lecture: Gene Set/Pathway Analysis Due: Five-Minute Paper 23
Wed Apr 6	Due: Project 5
Thu Apr 7	Lecture: Gene Set/Pathway Analysis Due: Five-Minute Paper 24
Tue Apr 12	Lecture: eQTLs & Transcriptome-Wide Association Studies Due: Five-Minute Paper 25
Thu Apr 14	Lecture: Phenome-Wide Association Studies & Metabolomics Due: Five-Minute Paper 26
Tue Apr 19	Lecture: Open Targets Due: Five-Minute Paper 27
Thu Apr 21	Lecture: Case Study: GWAS Finemapping Due: Five-Minute Paper 28
Tue Apr 26	Lecture: Case Study: Multiomics Due: Five-Minute Paper 29
Wed Apr 27	Due: Project 6
Thu Apr 28	Lecture: No Class

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:

<https://bc.pitt.edu/policies/policy/02/02-03-02.html>

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.

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.

Websites:

www.copyright.gov
www.provost.pitt.edu/faculty-handbook/ch3_uni_copyright

Disability Resources

www.studentaffairs.pitt.edu/drs

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 at:

www.diversity.pitt.edu

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 www.diversity.pitt.edu