Graduate School of Public Health
Department of Human Genetics

HUGEN 2073
Genomic Data Visualization and Integration

Spring 2023

Tuesday · 10:30 AM–11:50 AM
Thursdays · 10:30 AM–11:50 AM

Break · 11:05 AM–11:10 AM

Public Health A425
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

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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

Evaluation will be based on the following components. How they determine the letter-grade is explained further below.

Syllabus Review (1)

This assessment can be taken repeatedly until passed without costing mulligans (see below). *The initial deadline for this assignment is Thu Jan 12. It can be retaken an unlimited number of times until passed up to Wed Jan 18 at 11:59 PM EST.*

R Coding Project (1)

This assessment will consist of a set coding tasks in 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 (approximately 1 per class)

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 48 hours after the end of the class session (facilitated by viewing the recorded session via Canvas). Each is graded as either satisfactory or not.

Visualization Critique (1)

Students will critique visualizations from the scientific literature based on principles of good visualization. Graded as satisfactory or not.

Other homework assignments (7)

There will be seven other homework assignments. Several will assess proficiency in designing a visualization, coding to produce it, and writing a description of it; several will assess proficiency with annotating and integrating association results via "-omics" repositories and interpreting the findings. Each will be graded either satisfactory or unsatisfactory based on specific requirements.

Paper Critique (1)

Students will critique a recent genetics article, in particular describing and evaluating how the results are annotated and integrated with "-omics" data and discussing the implications. Graded as satisfactory or not. (Only required for an A+ grade.)

Late Policy & Revision Policy

Each student begins the term with four "mulligans." Each mulligan can be used for a 48-hour extension on an assignment or an opportunity to revise and resubmit either the coding project or another homework assignment (see instructor for revision deadline). You can use additional mulligans for further extensions on an assignment (e.g., 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 other homework) for which the student earns a 'satisfactory.'

Course Grade	D	C	B	A	A+
Syllabus Review	80%	80%	80%	80%	80%
R Coding Assignment	80%	80%	80%	80%	95%
Five-Minute Papers	All but 12	All but 9	All but 6	All but 3	All
Visualization Critique	1/1	1/1	1/1	1/1	1/1
Other homework	4/7	5/7	6/7	7/7	7/7
Paper critique	0/1	0/1	0/1	0/1	1/1

Each critique, five-minute paper, & homework will be graded satisfactory/ unsatisfactory.

For example, to earn a B, a student must do all of the following:

- score 80% on the syllabus review and complete 80% of the tasks in the R coding project
- fail to complete no more than 6 five-minute papers
- satisfactorily complete the visualization critique
- satisfactorily complete 6 of the 7 other homework assignments.

Remember that unsatisfactory critiques and assignments 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

Some lecture topics are likely to change. While some readings are listed below, others will be announced throughout the term on Canvas. Assigned reading for a class is expected to be completed before that class session.

Homework assignments and due dates will be announced on Canvas. Students are expected to check Canvas announcements regularly.

Students are expected to attend class in person and on time. Numerous absences or excessive tardiness will necessitate a meeting with the instructor.

Class	Date	Topic or Activity
1	**Tue Jan 10**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapters 2–3
		Lecture: Introduction; Elements of Visualization
		Due: Five-Minute Paper 1
2	**Thu Jan 12**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapters 1, 4
		Lecture: Introduction to Good Visualization
		Due: Five-Minute Paper 2
		Due: Syllabus Review
3	**Tue Jan 17**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapter 27
		Lecture: Visualization with R · Data Frames & Base Graphics
		Due: Five-Minutes Paper 3
4	**Thu Jan 19**	**Lecture:** Visualization with R · ggplot2
		Due: Five-Minutes Paper 4
5	**Tue Jan 24**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapter 6
		Lecture: Visualizing Amounts
		Due: Five-Minute Paper 5
6	**Thu Jan 26**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapters 7–9
		Lecture: Visualizing Distributions
		Due: Five-Minute Paper 6
7	**Tue Jan 31**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapter 12
		Lecture: Visualizing Associations
		Due: Five-Minute Paper 7
8	**Thu Feb 2**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapters 10–11

Class	Date	Topic or Activity
		Lecture: Visualizing Proportions
		Due: Five-Minute Paper 8
9	**Tue Feb 7**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapter 14
		Lecture: Visualizing Trends
		Due: Five-Minute Paper 9
10	**Thu Feb 9**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapter 16
		Lecture: Visualizing Uncertainty
		Due: Five-Minute Paper 10
11	**Tue Feb 14**	**Read:** Saxena *et al. Neurocomputing* 2017 https://dx.doi.org/10.1016/j.neucom.2017.06.053
		Lecture: Visualizing High Dimensions
		Due: Five-Minute Paper 11
12	**Thu Feb 16**	**Read:** Nusrat *et al. Comput Graph Forum* 2019 https://dx.doi.org/10.1111/cgf.13727
		Lecture: Visualization in Genetics & Genomics 1
		Due: Five-Minute Paper 12
13	**Tue Feb 21**	**Lecture:** Visualization in Genetics & Genomics 2
		Due: Five-Minute Paper 13
14	**Thu Feb 23**	**Lecture:** Ethical Issues in Visualization
		Due: Five-Minute Paper 14
15	**Tue Feb 28**	**Lecture:** Presenting Visualizations
		Due: Five-Minute Paper 15
16	**Thu Mar 2**	**Read:** *Fundamentals of Data Visualization* · www.clauswilke.com/dataviz Chapter 28
		Lecture: Visualization with Tableau & Python
		Due: Five-Minute Paper 16
	Tue Mar 7	**Lecture:** No Class · Spring Break
	Thu Mar 9	**Lecture:** No Class · Spring Break
17	**Tue Mar 14**	**Lecture:** Introduction to Annotation
		Due: Five-Minute Paper 17
18	**Thu Mar 16**	**Lecture:** SNV- & LD-Based Annotation
		Due: Five-Minute Paper 18
19	**Tue Mar 21**	**Lecture:** Annotation & Interpretation
		Due: Five-Minute Paper 19
20	**Thu Mar 23**	**Lecture:** UCSC Genome Browser Visualization & Tables
		Due: Five-Minute Paper 20

Class	Date	Topic or Activity
21	**Tue Mar 28**	**Lecture:** Constructing UCSC Genome Browser Custom Tracks
		Due: Five-Minute Paper 21
22	**Thu Mar 30**	**Lecture:** SQL & Querying UCSC Genome Browser Tables
		Due: Five-Minute Paper 22
23	**Tue Apr 4**	**Lecture:** Gene Set/Pathway Analysis 1
		Due: Five-Minute Paper 23
24	**Thu Apr 6**	**Lecture:** Gene Set/Pathway Analysis 2
		Due: Five-Minute Paper 24
25	**Tue Apr 11**	**Lecture:** eQTLs & Transcriptome-Wide Association Studies
		Due: Five-Minute Paper 25
26	**Thu Apr 13**	**Lecture:** Phenome-Wide Association Studies & Metabolomics
		Due: Five-Minute Paper 26
27	**Tue Apr 18**	**Lecture:** Open Targets
		Due: Five-Minute Paper 27
28	**Thu Apr 20**	**Lecture:** Case Study: GWAS Fine-mapping
		Due: Five-Minute Paper 28
29	**Tue Apr 25**	**Lecture:** Case Study: Multiomics
		Due: Five-Minute Paper 29
	Thu Apr 27	**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

If you are experiencing sexual assault, sexual harassment, domestic violence, and stalking, please report it to me and I will connect you to University resources to support you.

University faculty and staff members are required to report all instances of sexual misconduct, including harassment and sexual violence to the Office of Civil Rights and Title IX. When a report is made, individuals can expect to be contacted by the Title IX Office with information about support resources and options related to safety, accommodations, process, and policy. I encourage you to use the services and resources that may be most helpful to you.

As your professor, I am required to report any incidents of sexual misconduct that are directly reported to me. You can also report directly to Office of Civil Rights and Title IX: 412-648-7860 (M–F 8:30 AM–5:00 PM) or via the Pitt Concern Connection at: Make A Report

An important exception to the reporting requirement exists for academic work. Disclosures about sexual misconduct that are shared as a relevant 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 wish to make a confidential report, Pitt encourages you to reach out to these resources:

- The University Counseling Center:
  412-648-7930 (M–F 8:30 AM–5:00 PM) and
  412-648-7856 (*after business hours*)

- Pittsburgh Action Against Rape (community resource): 866-363-7273 (24/7)

If you have an immediate safety concern, please contact the University of Pittsburgh Police, 412-624-2121.

Any form of sexual harassment or violence will not be excused or tolerated at the University of Pittsburgh.

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