

THE OHIO STATE UNIVERSITY COLLEGE OF ARTS AND SCIENCES

Syllabus: STAT 6625 STATISTICAL ANALYSIS OF GENETIC EPIDEMIOLOGY DATA Autumn 2024

Course overview

Instructor

Instructor: Professor Shili Lin

Email address: lin.328@osu.edu

Class website: <http://carmen.osu.edu>

Lectures: Journalism Building 295, Tuesdays and Thursdays 9:10-10:30 AM

Office hours: Cockins Hall 440K, Tuesdays and Thursdays 10:45-11:45 AM, and by prior appointments

Grader

Ms. Yongqi Liu; email: liu.8224@buckeyemail.osu.edu; Office hours: TBD

Prerequisites

STAT 6301 and 6302, or permission of instructor

Course description

Introduction to genetic epidemiology; molecular genetics and Mendelian principles; genetic markers and distances; model-based and model-free population and family based (genome wide) association studies; association analysis using haplotypes; multi-trait analysis, causal inference, and other advanced topics.

Course learning outcomes

By the end of this course, students should successfully be able to:

- Understand a repertoire of problems addressed in genetic epidemiology
- Develop strategies for solving problems in genetic epidemiology
- Select an appropriate methodology for solving a specific problem

- Conduct genetic data analysis using software
- Communicate results to subject-area researchers

Course materials

References

Zigler A and Konig IR (2012) A statistical approach to genetic epidemiology: concepts and applications. 2nd Edition. John Wiley & Sons (electronic, on Carmen)

Laird N and Lange C (2011) The fundamentals of modern statistical genetics. Springer

Mukhopadhyay I and Majumder P (2023) Statistical methods in human genetics. Springer

Links to research papers will also be posted on Carmen throughout the semester.

Course technology

For help with your password, university e-mail, Carmen, or any other technology issues, questions, or requests, contact the OSU IT Service Desk. Standard support hours are available at <https://ocio.osu.edu/help/hours>, and support for urgent issues is available 24x7.

- **Self-Service and Chat support:** <http://ocio.osu.edu/selfservice>
- **Phone:** 614-688-HELP (4357)
- **Email:** 8help@osu.edu
- **TDD:** 614-688-8743

Baseline technical skills necessary for online courses

- Basic computer and web-browsing skills
- Navigating Carmen

Necessary software

- This class requires you to use the free statistical software package R (The R Project for Statistical Computing; <http://www.r-project.org/>).
 - You can download R for Windows, Mac, and Linux, from the CRAN archive at <https://cran.r-project.org>.
 - An in-depth introduction to R is available at <http://cran.r-project.org/doc/manuals/R-intro.pdf>
 - Hands-on tutorials are available in the Swirl system, which you can learn about at <http://swirlstats.com/>. In particular, “R Programming: The basics of

programming in R” is an appropriate first tutorial for students who have never used R.

- An easier to use interface to R is available in the free software package RStudio. This package is available for Windows, Mac, and Linux and can be downloaded from <http://rstudio.org>. **Note that RStudio requires R to be installed.**
- Software packages for analyzing genetic data will be introduced in class and in handouts posted on Carmen.

Course delivery

We will meet at our regularly scheduled class time in person throughout the semester for most of the lectures. However, some lectures may be pre-recorded and posted for asynchronous learning such as introductions to genetic software packages.

Grading and faculty response

Homework, Projects, and Exams

Assignment or category	Percentage
Participation in class discussion	5
Homework / Data analysis projects	35
Midterm Exam	30
Final Project	30
Total	100

Homework/Data analysis projects. Homework assignments, including several that involves data analysis, will be administered throughout the semester. You may discuss with other students, but DO NOT simply copy any part of someone else's work or solutions from any other sources, including, but are not limited to, generative AI. Violations will be treated as academic misconducts. I would encourage you to talk to the instructor and/or the grader if you have questions after serious attempts have been made to work on an assignment.

Midterm exam. The exam will take place in class, tentatively scheduled to be on Thursday, November 7, 9:10-10:30 AM. The exam will be closed book, but formulas deemed necessary will be provided on the exam.

Final project. Teams of two students will be formed to work on a Final Project together – details will be discussed after the first exam. The team will present their results in the last week of the semester, and each student is expected to write an individual (not team) report summarizing their findings.

Logistics and policies. Homework and projects will be submitted through the class website. Typically, no late homework/projects will be accepted, and no make-up exams will be given. However, if you are unable to complete an assignment on time or have an emergency that prevents you from taking the exam on the date specified, please get in touch with the instructor as soon as possible. For the exam, a basic calculator is permitted; however, using a cell phone, tablet, laptop or any other communication device for this purpose is not permitted.

Faculty feedback and response time

The following sections delineate my intended availability throughout the course.

Grading and feedback

Sample solutions to homework assignments will be posted soon after all the papers are submitted. You can generally expect feedback within **7 days**, but there may be exceptions (e.g. grader has her own exam in a particular week).

E-mail

The instructor will reply to e-mails within **24 hours on weekdays**.

Other course policies

Academic integrity policy

It is the responsibility of the Committee on Academic Misconduct to investigate or establish procedures for the investigation of all reported cases of student academic misconduct. The term “academic misconduct” includes all forms of student academic misconduct wherever committed; illustrated by, but not limited to, cases of plagiarism and dishonest practices in connection with examinations. Instructors shall report all instances of alleged academic misconduct to the committee (Faculty Rule 3335-5-487). For additional information, see the Code of Student Conduct <http://studentlife.osu.edu/csc/>.

If I suspect that a student has committed academic misconduct in this course, I am obligated by university rules to report my suspicions to the Committee on Academic Misconduct. If COAM determines that you have violated the university's *Code of Student Conduct* (i.e., committed academic misconduct), the sanctions for the misconduct could include a failing grade in this course and suspension or dismissal from the university.

If you have any questions about the above policy or what constitutes academic misconduct in this course, please contact me.

Other sources of information on academic misconduct (integrity) to which you can refer include:

- Committee on Academic Misconduct web page (go.osu.edu/coam)
- *Ten Suggestions for Preserving Academic Integrity* (go.osu.edu/ten-suggestions)

Copyright for instructional materials

The materials used in connection with this course may be subject to copyright protection and are only for the use of students officially enrolled in the course for the educational purposes associated with the course. Copyright law must be considered before copying, retaining, or disseminating materials outside of the course.

Statement on Title IX

Title IX makes it clear that violence and harassment based on sex and gender are Civil Rights offenses subject to the same kinds of accountability and the same kinds of support applied to offenses against other protected categories (e.g., race). If you or someone you know has been sexually harassed or assaulted, you may find the appropriate resources at <http://titleix.osu.edu> or by contacting the Ohio State Title IX Coordinator at titleix@osu.edu

Commitment to a diverse and inclusive learning environment

The Ohio State University affirms the importance and value of diversity in the student body. Our programs and curricula reflect our multicultural society and global economy and seek to provide opportunities for students to learn more about persons who are different from them. We are committed to maintaining a community that recognizes and values the inherent worth and dignity of every person; fosters sensitivity, understanding, and mutual respect among each member of our community; and encourages each individual to strive to reach his or her own potential. Discrimination against any individual based upon protected status, which is defined as age, color, disability, gender identity or expression, national origin, race, religion, sex, sexual orientation, or veteran status, is prohibited.

Land Acknowledgement

We would like to acknowledge the land that The Ohio State University occupies is the ancestral and contemporary territory of the Shawnee, Potawatomi, Delaware, Miami, Peoria, Seneca,

Wyandotte, Ojibwe and Cherokee peoples. Specifically, the university resides on land ceded in the 1795 Treaty of Greeneville and the forced removal of tribes through the Indian Removal Act of 1830. I/We want to honor the resiliency of these tribal nations and recognize the historical contexts that has and continues to affect the Indigenous peoples of this land.

More information on OSU's land acknowledgement can be found at <https://mcc.osu.edu/about-us/land-acknowledgement>

Your mental health

As a student you may experience a range of issues that can cause barriers to learn, such as strained relationships, increased anxiety, alcohol/drug problems, feeling down, difficulty concentrating and/or lack of motivation. These mental health concerns or stressful events may lead to diminished academic performance or reduce a student's ability to participate in daily activities. The Ohio State University offers services to assist you with addressing these and other concerns you may be experiencing. If you or someone you know are suffering from any of the aforementioned conditions, you can learn more about the broad range of confidential mental health services available on campus via the Office of Student Life's Counseling and Consultation Service (CCS) by visiting ccs.osu.edu or calling [614-292-5766](tel:614-292-5766). CCS is located on the 4th Floor of the Younkin Success Center and 10th Floor of Lincoln Tower. You can reach an on call counselor when CCS is closed at [614-292-5766](tel:614-292-5766) and 24 hour emergency help is also available 24/7 by dialing 988 to reach the Suicide and Crisis Lifeline.

Religious accommodations

It is Ohio State's policy to reasonably accommodate the sincerely held religious beliefs and practices of all students. The policy permits a student to be absent for up to three days each academic semester for reasons of faith or religious or spiritual belief.

Students planning to use religious beliefs or practices accommodations for course requirements must inform the instructor in writing no later than 14 days after the course begins. The instructor is then responsible for scheduling an alternative time and date for the course requirement, which may be before or after the original time and date of the course requirement. These alternative accommodations will remain confidential. It is the student's responsibility to ensure that all course assignments are completed.

Disclaimer

This syllabus should be taken as a fairly reliable guide for the course content. However, you cannot claim any rights from it and in particular we reserve the right to change due dates, exam dates or the methods of grading and/or assessment if necessary. Any changes will be communicated to you through official course announcements.

Accessibility accommodations for students with disabilities

Requesting accommodations

The university strives to make all learning experiences as accessible as possible. In light of the current pandemic, students seeking to request COVID-related accommodations may do so through the university's request process, managed by Student Life Disability Services. If you anticipate or experience academic barriers based on your disability including mental health, chronic or temporary medical conditions, please let me know immediately so that we can privately discuss options. To establish reasonable accommodations, I may request that you register with Student Life Disability Services. After registration, make arrangements with me as soon as possible to discuss your accommodations so that they may be implemented in a timely fashion. **SLDS contact information:** slds@osu.edu; 614-292-3307; slds.osu.edu; 098 Baker Hall, 113 W. 12th Avenue.

Accessibility of course technology

This course requires use of CarmenCanvas (Ohio State's learning management system) and other communication and multimedia tools. If you need additional services to use these technologies, please request accommodations with your instructor.

- Canvas accessibility (go.osu.edu/canvas-accessibility)

Course schedule (tentative)

Note: Reading assignments are from the text by Zigler and Konig; however, materials from other sources will be supplemented as well.

Week	Dates	Topics	Assigned Readings
1	8/20, 8/22	Molecular genetics, Mendelian principles, genetic traits	Chapters 1 and 2
2	8/27, 8/29	HWE, genetic (DNA) markers, map distances	Chapters 3 and 5
3	9/3, 9/5	Linkage analysis – an overview Linkage disequilibrium	Chapter 7 Chapter 10
4	9/10, 9/12	Population-based association analyses	Chapter 11
5	9/17, 9/19	Population-based association analyses Family-based association analyses	Chapters 11 Chapter 12
6	9/24, 9/26	Family-based association analyses	Chapter 12
7	10/1, 10/3	Haplotype analysis	Chapter 13
8	10/8	Genome-wide association studies (GWAS) and multiple comparison	Chapter 14
9	10/15, 10/17	Pop-based assoc. analyses for rare variants	Other reading
10	10/22, 10/24	Family-based assoc. analyses for rare variants	Other reading
11	10/29, 10/31	Multi-trait analysis	Other reading
12	11/5 11/7	Polygenic risk score Midterm	Other reading
13	11/12, 11/14	Mendelian randomization Mediation analysis	Other reading
14	11/19, 11/21	Ancestry inference	Other reading
15	11/26	Final project presentations	
16	12/3	Final project presentations	