THE OHIO STATE UNIVERSITY

COLLEGE OF ARTS AND SCIENCES

Syllabus: STAT 6625
STATISICAL ANALYSIS OF GENETIC EPIDEMIOLOGY DATA
Autumn 2020

Course overview

Instructor

Instructor: Shili Lin
Email address: shili@stat.osu.edu
Class website: http://carmen.osu.edu
Lectures: Via CarmenZoom: Tuesdays and Thursdays 9:10-10:30 am
Office hours: Via CarmenZoom: Tuesdays 10:30-11:30 am; Wednesdays 1:35-2:35 pm

Grader

Ms. Zilu Liu; email: liu.6536@osu.edu

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; analysis of DNA methylation data; other current topics. **Prerequisites:** 6301 and 6302, or permission of instructor.

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

Required

Zigler A and Konig IR (2010) A statistical approach to genetic epidemiology: concepts and applications. 2nd Edition. John Wiley & Sons (electronic, on Carmen)

Links to research papers through OSU library 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

Technology skills necessary for this specific course

- CarmenZoom

Necessary equipment

- Computer: current Mac (OS X) or PC (Windows 10+) with high-speed internet connection
- Webcam: built-in or external webcam, fully installed
- Microphone: built-in laptop or tablet mic or external microphone

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 throughout the semester for most of the lectures through CarmenZoom. Recorded lectures will be posted soon after on class website. For certain topics, however, lectures may be pre-recorded and posted for asynchronous learning – learn at your own speed and at a time of your choosing – such as introductions to genetic software packages.

Detailed instructions for asynchronous learning will be posted prior to the commencing of such activities.

The instructor will hold weekly office hours – times as given above – via CarmenZoom.

Grading and faculty response

Homework, Projects, and Exams

Assignment or category	Percentage
Homework	20
Data analysis projects	15
Exams	35
Final Project	30
Total	100

Homework. There are six homework assignments 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. Violations will be treated as academic misconducts. I would encourage you to talk to me if you have questions after serious attempts have been made to work on an assignment.

Data analysis projects. There are three data analysis projects that will be assigned throughout the semester. Completion of these projects require the use of genetic software that are used and discussed in class. As in completing your homework assignment, you may discuss with other students but you need to complete the work on your own. I would encourage you to talk to me if you have questions after serious attempts have been made to work on the projects.

Exams. There are two one-hour long exams, with the tentative dates given below; any date changes will be communicated well in advance. The exams will be closed book, but formulas deemed necessary will be provided on the exam – each exam will be proctored through webcam.

Exam 1	Thursday, October 1	9:30-10:30 am
Exam 2	Thursday, November 12	9:30-10:30 am

Final project. Teams of 2-3 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 classes, 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 me as soon as possible so that we can work out a solution. For the exams, you need to work independently without any forms of assistance (from the internet, notes, or other people) or communication with anyone except the proctor. 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 his/her own exam in a particular week).

E-mail

I will reply to e-mails within **24 hours on week days**.

Attendance, participation, and discussions

Student participation requirements

For lectures that are delivered synchronously, you are encouraged to join the live lectures and participate in discussions. If you have to miss a live lecture, you are responsible for learning the materials discussed in the recorded lectures, and you may ask questions during virtual office hours. You are also expected to participate in discussion forums.

Discussion and communication guidelines

The following are my expectations for how we should communicate as a class. Above all, please remember to be respectful and thoughtful.

- **Writing style**: While there is no need to participate in class discussions as if you were writing a research paper, you should not use text lingo.
- **Tone and civility**: We need to strive to create a supportive learning community where everyone feels safe and people can disagree amicably. Remember that sarcasm doesn't always come across online.
- **Citing your sources**: When we have academic discussions, please cite your sources to back up what you say. (For the textbook or other course materials, list at least the title and page numbers. For online sources, include a link.)
- **Backing up your work**: Consider composing your academic posts in a word processor, where you can save your work, and then copying into the Carmen discussion.

Other course policies

Health and safety

The Ohio State University Wexner Medical Center's Cornavirus Outbreak site (https://wexnermedical.osu.edu/features/coronavirus) includes the latest information about COVID-19 as well as guidance for students, faculty and staff. Guidelines and requirements for campus safety from the University's COVID-19 Transition Task Force were published on July 1 on the Safe and Healthy website (https://safeandhealthy.osu.edu).

Potential disruptions to instruction

• If the instructor is unable to be present in person because of positive diagnosis, symptoms, or quarantine following contact tracing, a new instructor will be assigned to the course. Details will be given on the course website.

Student academic services

Student academic services on the OSU main campus: http://advising.osu.edu/welcome.shtml.

Student support services

Student support services offered on the OSU main campus: http://ssc.osu.edu.

Academic integrity policy

- **Reusing past work**: In general, you are prohibited in university courses from turning in work from a past class to your current class, even if you modify it. If you want to build on past research or revisit a topic you've explored in previous courses, please discuss the situation with me.
- **Falsifying research or results**: All research you will conduct in this course is intended to be a learning experience; you should never feel tempted to make your results or your library research look more successful than it was.
- **Collaboration and informal peer-review**: While study groups are encouraged, remember that copying solutions from another student or from any other sources is not permitted. If you're unsure about a particular situation, please feel free just to ask ahead of time.

Group projects: This course includes a group project, which provides you the opportunity to formally collaborate with your colleagues. Please work on your group project at a distance, not in person. Each student needs to write up their report separately and individually. No part of the report can be copied from that of another student.

Ohio State's 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 and 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/.

Copyright disclaimer

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, Kellie Brennan, at titleix@osu.edu

Accessibility accommodations for students with disabilities

The University strives to make all learning experiences as accessible as possible. 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 online course requires use of Carmen (Ohio State's learning management system) and other online communication and multimedia tools. If you need additional services to use the following list of technologies, please request accommodations with your instructor.

- Carmen (Canvas) accessibility
- Streaming audio and video
- Synchronous course tools

Your mental health

As a student you may experience a range of issues that can cause barriers to learning, 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. 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 and 24 hour emergency help is also available through the 24/7 National Suicide Prevention Hotline at 1-800-273- TALK or at suicidepreventionlifeline.org

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.

Course schedule (tentative)

Note: Reading assignments are from the text by Zigler and Konig. Research articles will be assigned as reading materials in the second half of the semester.

Week	Dates	Topics	Assigned Readings
1	8/25, 8/27	Molecular Genetics, Mendelian Principles, Genetic Traits	Chapters 1 and 2
2	9/1, 9/3	HWE, Genetic (DNA) Markers, Map Distances	Chapters 3 and 5
3	9/8, 9/10	Linkage Disequilibrium	Chapter 10
4	9/15, 9/17	Population-Based Association Analyses	Chapter 11
5	9/22, 9/24	Pop. and Family-Based Association Analyses	Chapters 11 and 12
6	9/29, 10/1	Family-Based Association Analyses	Chapter 12
7	10/6, 10/8	Haplotype Analysis	Chapter 13
8	10/13, 10/15	Genome-Wide Association Studies (GWAS)	Chapter 14
9	10/20, 10/22	Pop-Based Assoc. Analyses for Rare Variants	Research papers
10	10/27, 10/29	Family-Based Assoc. Analyses for RVs	Research papers
11	11/3, 11/5	DNA Methylation and Epigenome-Wide Association Studies (EWAS)	Research papers
12	11/10, 11/12	Differentially Methylated Regions	Research papers
13	11/17, 11/19	Hi-C Data and Chromatin Interactions	Research papers
14	11/24	ChIA-PET and Long-Range Regulation	
15	12/1, 12/3	Final Project Presentations	