STAT 494: Statistical Genetics

Macalester College, Spring 2025

Section 01: TTh 9:40 - 11:10 am, Olin Rice (OLRI) 241

Course Overview

This course provides an introduction to the field of *statistical genetics*. Statistical methods for analyzing genetic data and understanding the genetic basis of diseases and traits are at the heart of government-sponsored precision medicine initiatives (e.g., <u>All of Us</u>), genetic testing routinely conducted at health clinics, and direct-to-consumer genetic and ancestry testing offered by companies like 23andMe and AncestryDNA. Statistical geneticists work to answer these important scientific questions while navigating the **statistical challenges** posed by genetic data. In this course, we will explore the statistical methods that have been proposed to address these challenges, applying and extending methods you may have encountered in previous courses (e.g., linear models, principal component analysis) to the context of genetic data and developing a **deeper understanding of statistical methods** along the way. Throughout the course, we will place a particular emphasis on **communication**, **computing**, and **independent learning**.

Course Topics / Tentative Schedule

We will begin the semester with a discussion of **genome-wide association studies** and the challenges posed by the high-dimensional nature of genetic data (Unit 1: Weeks 1–4). From there, we will explore methods for inferring and adjusting for confounding by **genetic ancestry** (Unit 2: Weeks 5–7). As time allows, we will also consider methods for addressing two other challenges posed by genetic data: correlation and sparsity (Unit 3: Weeks 8–11). We'll conclude with independent projects (Weeks 12 – 15).



Your Instructor

Dr. Kelsey E. Grinde

Pronouns: she/her/hers Pronunciation: listen <u>here</u> Office: OLRI 226 Email: <u>kgrinde@macalester.edu</u>



Call me Kelsey

Students sometimes wonder what to call their professors. I prefer to be called by my first name, *Kelsey* (KELL-see), but I am also okay with *Professor Grinde* (GRIN-dee). Please note that I prefer not to be called *Professor* (without my last name attached) or *Ms./Mrs. Grinde*.

Please help me make sure that I call you by your preferred name (with correct pronunciation!) and pronouns, too!

A Letter from Your Professor

Hello, and welcome to a new semester at Macalester!

How to contact me:

- Stop by my **office hours**: see the Google Calendar on Moodle for times and locations
- Email me to request a one-on-one **appointment**
- Post questions about course content, homework assignments, or anything else relevant to the entire class on **Slack**
- Send personal questions or updates (e.g., attendance, grades) via email

I do my best to respond to all messages quickly, but I also work hard to maintain a healthy work/life balance. With that in mind, please allow extra time for a response on evenings and weekends.

I'm excited to spend the next 15 weeks sharing the field of statistical genetics with all of you. I was first introduced to this field through a summer undergraduate research program. Prior to that summer, I had no prior experience with genetics (beyond high school biology) or research, and I had only taken a handful of stats courses (Statistical Modeling, Probability, and Statistical Theory). Needless to say, I had a *lot* of learning to do that summer. It was hard work, but it ended up being one of the most rewarding and transformative educational experiences of my career. By the end of the summer, I decided to apply to PhD programs in biostatistics so I could continue the learning that started that summer. As they say, "the rest is history." (Unfortunately, this particular research program no longer exists, but there are many other great programs out there. Come chat with me about summer research if you're interested in learning more!)

My hope is that this class can provide an educational experience that helps you develop a deeper understanding of statistical methods and the role that (bio)statistics plays in the field of science, and helps you further develop your skills in communication, computing, and independent learning. This is only my second time teaching this course, and it is a unique course offering at the undergraduate level (meaning there are no textbooks or other examples from which we can borrow). This means that there will be times when I am learning right along with you. WIth that in mind, *please let me know if something about the course is not working for you*. I want to hear from you about how I can make adjustments. I'm looking forward to a great semester with all of you!

Kelsey

Learning Goals

To be determined! My goal is to help guide all of you in your continued journey toward become lifelong learners. With that in mind, I believe it is important that you have a say in the goals that you set for yourself this semester. I have some ideas to get us started, but I also want to hear what you are hoping to learn/gain from this class — it may not be the same for all of you! **We will develop these goals together** during the first weeks of class, and then reflect and make adjustments, as needed, throughout the rest of the semester.

Course Structure and Learning Activities

Lecture. The initial introduction to most concepts will come in the form of in-class lectures. I may periodically supplement these lectures with pre-class readings or videos, all of which will be made available on the course website.

Activities/Labs. Most class periods will involve an in-class activity or lab to practice, apply, extend, or synthesize the concepts introduced in lecture. There may be times that we do not finish these activities during class time; in those cases, I expect that you will take time outside of class to complete any remaining parts of the activity/lab. You will submit labs for feedback on an approximately bi-weekly basis.

Journal Club. Approximately once per week, we will spend a class period discussing a journal article or some other piece of scientific/statistical writing. I will lead the first discussion, and you (students) will lead all others. I'll provide a sign-up sheet early in the semester. I will also provide guidance on how to prepare for these discussions and will be available during office hours to help the discussion leaders as needed. The goals of this activity are to learn how to read a scientific article, familiarize yourself with different styles of writing, practice (and get feedback on) your presentation skills, and see examples of how statistical methods are developed and applied "in the real world."

Department Seminars. As a 400-level MSCS capstone course, you are expected to attend, and subsequently submit a short reflection for, *at least two* MSCS seminars (or other related presentations with instructor approval). I encourage you to attend more than two if you are able. These talks serve as an opportunity to learn about research and career opportunities in (bio)statistics and to reflect on effective techniques for communicating statistical concepts.

Major Learning Assessments

Content Summaries. After each of our three major units (see page 1), you will create and submit a summary of the content we covered during that unit. The focus, format, and audience of these "content summaries" are up to you and will depend on the learning goals that you've set for yourself (although I'll provide suggestions!). The goal of these assignments is to demonstrate your understanding of key course concepts, as well as your communication skills.

Learning Reflections and Grading Conferences. You will submit reflections on your learning at three points throughout the semester, starting with an **Initial Reflection** (due February 7) to set your goals for the course, followed by a **Midterm Reflection** (due March 7), and concluding with a **Final Learning Reflection & Portfolio** (due May 5). I will provide written feedback on all three reflections, as well as verbal feedback in the form of two **Learning Conferences** during the weeks of March 10–14 (Midterms) and May 8–12 (Finals).

Project. As a 400-level MSCS capstone course, this course includes a substantial project which you must pass in order to pass this course. During the second half of the semester, you will work in small groups to explore a new topic in statistical genetics or dive deeper into something that we introduced in class. There will be multiple project checkpoints, allowing ample opportunities for practice, feedback, and revision. You will give an **Introduction Presentation** (April 11), a **Pictures-Only Presentation** (April 15–17), and a **Final Presentation** (April 29–May 1), as well as create a **Digital Artifact** that will be due at the end of the the last full week of classes (May 2). Exact scheduling of the presentations will depend on the number of project groups and will be determined later in the semester.

Co-Grading

This course will use a grading system often referred to as *ungrading* (although I personally prefer the term *co-grading*). Read <u>this post</u> for a great introduction to ungrading and the myriad problems with "traditional" grades that it aims to correct. For each of the assignments that you submit this semester, I will focus on *engaging* with your work rather than evaluating it and providing *feedback* rather than grades. We will determine your midterm and final grades *together*, based on a combination of the quality of your content summaries and final project, your engagement in learning activities, and your learning reflections.

If ungrading is new to you (and there's a good chance that it is!), it may take time to adjust. If at any point during the semester you are concerned about your learning (or grade), please get in touch with me to set up a time to discuss!

Advice for Success in STAT 494

Ask questions. When you have questions, please stop me during class, ask your neighbor, post on Slack, and come to office hours. *Saying "I don't understand" is an important part of learning* and it helps your classmates (and the instructor!).

Come to office hours. Office hours are a great time to talk about course material and assignments, study strategies, selecting courses, declaring a major, grad school and/or career planning, or life in general. *You don't need to have a specific question in order to attend office hours*: it can also be a great space to review concepts, talk through examples, or just chat!

Make time. Learning new material requires time: beyond your first introduction to a topic, you also need time to absorb, time to practice, time to revise, time to synthesize, and time to apply. *Start your assignments early.* It is very hard to be creative (or to debug code) when you are in a rush. In addition to the 3 hours we spend together during class, expect to spend about 7 hours per week on this class. If you're spending much more (or less!) time than that, please let me know.

Attend class. Active participation in this class will be key to your learning. We'll use class time to introduce new concepts, ask and answer questions, review material, and practice applying concepts in a collaborative environment. To ensure the best learning experience for you and your classmates, *come prepared, engage in class*, and *make full use of the entire class period*.

If you miss class... I understand that there may be times you are unable to attend class. In those cases, I expect that you will check the course website to see what you missed, review the material, complete the in-class activity on your own, get notes from your classmates, and (after doing all of the above) come to office hours with specific questions.

Prioritize your well-being. Investing time into taking care of yourself will help you engage more fully in your academic experience. Remember that *beyond being a student, you are a human being* carrying your own experiences, thoughts, emotions, and identities with you. If you are having difficulties maintaining your well-being, please contact me and/or check out these <u>resources</u>. As part of prioritizing your well-being (and others around you), it is important that you *stay home if you are are feeling sick*. See the recommendations above (*Attend class*) and below (*Communicate*) if you miss class or need extra time on an assignment.

A quick personal note (please read!): One of my family members is severely immunocompromised due to a recent medical emergency. This means that I have to be very careful not to contract, or at least pass along, any viruses or other illnesses. As a precautionary measure, there may be times this semester when I will choose to wear a mask in office hours, class, or other settings. (So if you see me wearing a mask, it does not necessarily mean that I myself am experiencing respiratory symptoms.) It will be a huge help to me (and my family!) if you could please stay home when you are sick and take care to follow any other suggestions outlined on the <u>Hamre Center</u> and <u>CDC</u> websites. **Thank you!**

Communicate. I will do my best to clearly communicate changes to expectations, deadlines, office hours, or class meetings due to instructor illness or unexpected life issues. Please make sure to *check Moodle and Slack regularly* so you don't miss any important announcements. I know that you may also have issues come up: if so, please get in touch with me to discuss solutions. In particular, I ask that you *please check in with me, as soon as possible, if*:

- You need to miss multiple classes in a row
- You have a conflict (e.g., athletic competition, religious observance) with a major assignment deadline
- You need accommodation(s)
- You are worried about meeting a deadline
- Something about the class is not working for you

Course Policies

All members of this course (students, preceptors, and myself) are expected to adhere to the <u>MSCS</u> <u>Community Guidelines</u>. This will include:

- Being inclusive
- Being present
- Asking for and offering help
- Being collaborative
- Being mindful of academic integrity

If you witness or experience any violations of these guidelines, I encourage you to come chat with me and/or follow the suggestions in the Community Guidelines document to report the issue.

Late Work & Extensions

I set deadlines so that I can get feedback to you in a timely manner, and because the material in this course builds from week to week. I also understand that life happens, and try to be flexible when possible.

If you are worried about meeting a deadline, please get in touch with me to make a plan. If you contact me *before the deadline*, preceptors and I can plan our grading accordingly and make sure that you still get feedback on your work in a timely manner. In particular, all requests related to major learning assessments (e.g., projects, learning reflections, grading conferences) need to be communicated *at least one week in advance* for full consideration. I cannot *guarantee* that I will be able to accommodate all extension requests, but I will do my best to work with you whenever possible.

If you do not get in touch with me, or if you contact me on short notice, *late work may incur a penalty* (e.g., not receiving feedback, delayed feedback, impact on final grade). Please plan accordingly.

Academic Integrity

I expect all of you to be familiar with, and adhere to, the <u>college standards on academic integrity</u>. Please take time to review this policy if you have not done so recently. In this course, I encourage you to work with your classmates to discuss material and ideas for your assignments. With that said, in order for you to receive feedback on YOUR learning, you must *submit your own work (code and writing)* and *explicitly credit all sources (including Al tools)*. I will schedule a meeting with you if I have any concerns that this policy has been violated.

Artificial Intelligence (AI)

When used carefully, AI can be a useful tool. However, it is important to be aware of its limits:

- Al is not always correct. Even when correct, Al may use terminology or coding syntax that is not appropriate for our course. *NEVER copy-paste output from Al into an assignment*. Critically evaluate the response and rewrite it in your own words (or code) before submitting.
- Ideas, language, or code produced by AI *must be cited*, just like any other resource. Failure to do so is in violation of Mac's <u>academic integrity policy</u>.
- The prompts you write are providing data to the AI tool. Don't provide data (prompts or attachments) that you do not have permission to share.
- Al has a *large environmental impact* (<u>evidence</u>). If you can do something in a more energy efficient way (e.g. using pre-existing resources), please do.

Accommodations. I am committed to creating an accessible and inclusive classroom for all students. If you need accommodations, please contact <u>Disability Services</u> to discuss your needs. Once you've met with Disability Services, please then set a time to meet with me to discuss your accommodation plan for this course. It is important to *arrange this meeting as early in the semester as possible* (ideally within the first week), in order to ensure that your accommodations can be implemented early on. It is your responsibility to make sure you are registered with Disability Services. If you wait until later in the course, I will not be able to accommodate you retroactively.

Title IX. If you or anyone you know has experienced harassment or discrimination on the basis of sex or gender, know that you are not alone. Macalester provides staff and resources to help you and support you. More information is available on the <u>Title IX website</u>. Please be aware that *all Macalester faculty and preceptors are mandatory reporters*, which means that if we become aware of incidents or allegations of sexual misconduct, we are required to share the matter with the Title IX Coordinator. Although we have to make that notification, you control how your case is handled, including whether or not you wish to pursue a formal complaint. If you would like to speak to someone *confidentially*, see <u>here</u>.

Religious Observance. Students may wish to take part in religious observances that occur during this semester. I've done my best to schedule deadlines around major holidays, but if you have a religious observance/practice that conflicts with class or an assignment deadline, please let me know and we can discuss accommodations.

Preceptors

Since most of our 400-level STAT courses are offered on an every-other-year rotating basis, it is often difficult for us to hire preceptors for these courses. This particular course was last taught Fall 2022, which means that all of the students who took that course have since graduated.

This semester we are **very** lucky to have one awesome preceptor who will be helping out with our course. Tina has nearly one year of research experience in statistical genetics under her belt (ask her about her summer research & honors projects!) and is thus very familiar with the content we will cover this semester. She will hold office hours, provide feedback on assignments, join us during class time, and, as time allows, answers questions on Slack.

See the STAT 494 Preceptor Office Hours Calendar (at the top of our Moodle page) for up-to-date information about her office hour times and locations.



MSCS Preceptor Expectations. The role of an MSCS preceptor is to help students with content questions, assist in the navigation of available resources, advise on studying approaches for classes, and assist with concepts, tools, and skills needed for problem sets. Students are accountable for their own learning; as such, preceptors are not allowed to share answers to assignments (unless specifically directed by the instructor), are not expected to immediately know the right approach, or provide assistance outside of office hours. Additional guidelines and expectations on how to interact with preceptors can be found <u>here</u>.