STAT 494: Statistical Genetics

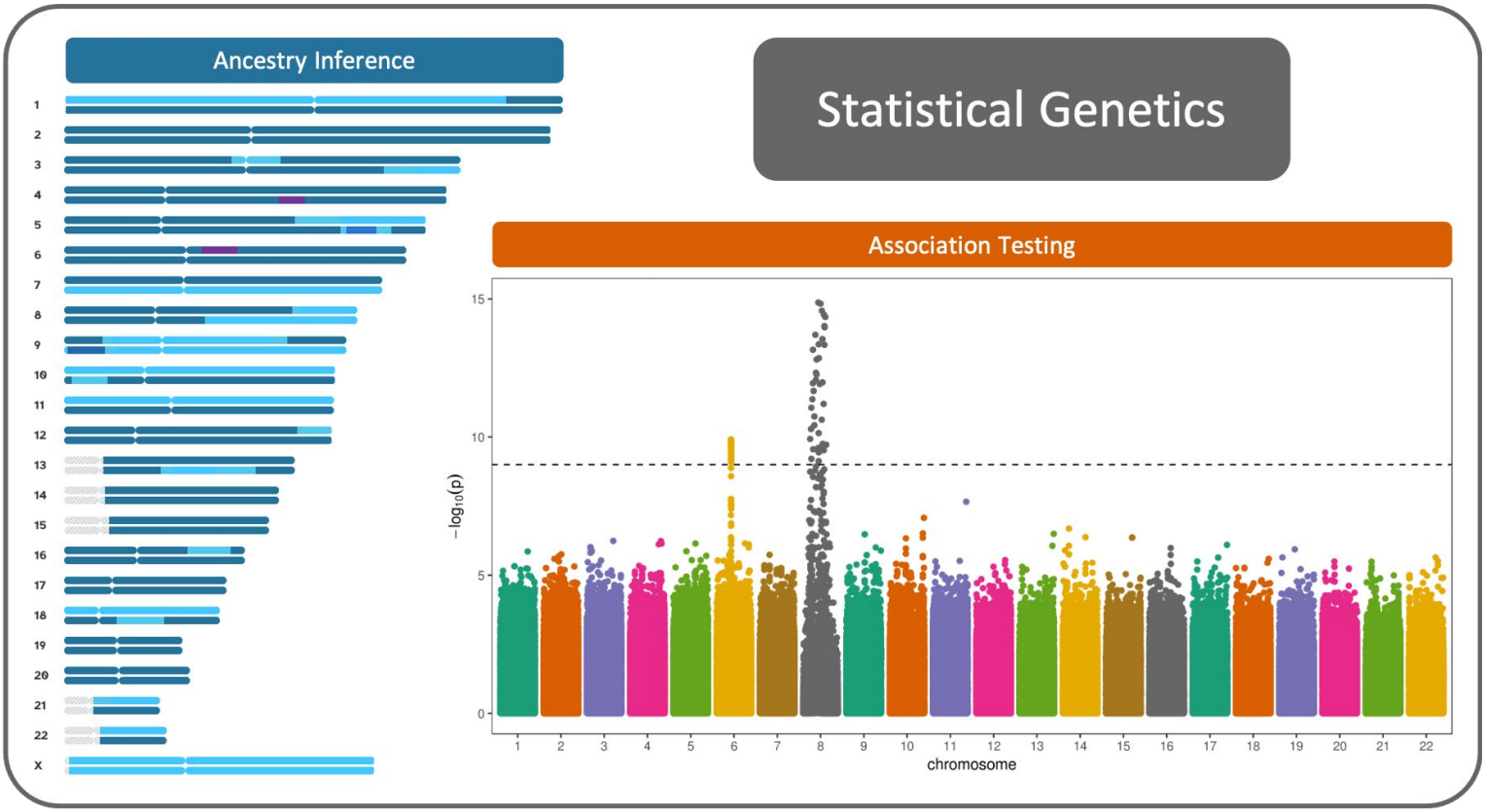
Macalester College, Fall 2022

Section 01: MWF 1:10 pm – 2:10 pm, OLRI 254

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., All of Us), 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 **unique 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

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. From there, we will move on to explore methods for inferring and adjusting for **confounding** by **genetic ancestry**. As time allows, we will also consider methods for addressing two other challenges posed by genetic data: **correlation** and **sparsity**.

Your Instructor

Dr. Kelsey E. Grinde

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

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 in an ongoing effort to maintain a healthy work/life balance, please allow extra time for a response on evenings and weekends.

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!

I'm very 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 research program in biostatistics and statistical genetics during the summer between my junior and senior years of college. 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 courses in statistics (Statistical Modeling, Probability, and Mathematical Statistics). 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 was inspired, and 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 my first time teaching this course, so there will be times when I am learning right along with you. ***If something about the course is not working for you, please let me know***. I have built this course to be flexible and 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 course concepts and material will come in the form of in-class lectures. I may periodically supplement these in-class lectures with short videos or readings that will be made available on Moodle.

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. I will not collect or grade these assignments, but I will post solutions. Make these as useful for yourself as possible!

Journal Club. Every Monday—with a handful of exceptions—we will spend our entire class period discussing a journal article or some other piece of scientific/statistical writing. Each week, a different group of students will sign up to lead the discussion. I will 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."

Guest Speakers and Department Seminars. We will have two guest speakers that will be joining our class this semester to talk with us about their research in statistical genetics and career path. Throughout the semester, I also encourage you to attend Department Seminars, Beyond Mac Sessions, and related events to learn about research and career opportunities in (bio)statistics and reflect on effective techniques for communicating statistical concepts.

Major Learning Assessments

Learning Reflections (x3) and Grading Conferences (x2). I will ask you to submit reflections on your learning at three points throughout the semester, starting with an **Initial Reflection** (due September 9) to set your goals for the course, followed by a **Midterm Reflection** (due October 17), and concluding with a **Final Learning Reflection** (due December 12). I will provide written feedback on all three reflections, as well as verbal feedback in the form of two **Grading Conferences** during the weeks of October 17 and December 12.

Content Summaries (x3). At three points throughout the semester (September 23, October 14, November 4) you will submit a summary of the content we covered during the preceding weeks of the course. 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.

Project. During the second half of the semester, you will work individually or 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** (November 7 – 11), an **Update Presentation** (November 14 – 21), and **Final Presentation** (November 28 – December 9), as well as create a **Digital Artifact** that will be due on the last day of class (December 9). Exact scheduling of the presentations will depend on the number of project groups. We will discuss the format and expectations for presentations and digital artifacts later in the semester.

Ungrading

This course will use a grading system known as *ungrading*. Read this post 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***. There may be times you are unable to attend class: in those cases, I expect that you will check Moodle 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 resources. 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.

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

Shared Community Commitment

We all have a role to play in keeping each other safe. As a member of the Macalester community, you are expected to follow the practices outlined in the Shared Community Commitment: Mac Stays Safer 3.0. Included in this is the expectation that all individuals will wear a well-fitted face covering (e.g, N95, KN95, and K94 masks/respirators) in public indoor spaces, regardless of vaccination status, at least through September 14. This means that we all need to ***wear masks during class time***. I also ask that you ***please wear a mask whenever visiting my office***; I will do the same. I'll inform you of updates to our classroom and office hour mask policies as campus and public health guidelines change.

Important Course Policies

I expect you to adhere to the **MSCS Community Guidelines** in all of your interactions with classmates (and me). 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.

Academic Integrity

I expect all of you to be familiar with the college standards on academic integrity. Please take time to review this policy if you have not done so recently. I encourage you to work with your classmates to discuss material and ideas for your assignments, but in order for you to receive feedback on YOUR learning, ***all submitted work (including code!) must be written in your own words.*** I take academic integrity very seriously and will schedule a meeting with you if I have any concerns that this policy has been violated.

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. That said, I will accept late work, without any penalty to your grade, provided that it is submitted before grading begins (typically within 1–2 days of the deadline). If you get in touch with me to request an extension ***before the deadline***, I can plan my grading accordingly and make sure that you get feedback on your work. However, I cannot guarantee that I will be able to accommodate extension requests that are made after a deadline has passed—or shortly (e.g., less than 24 hours) before something is due, so please plan accordingly. 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.

Accommodations

I am committed to creating an accessible and inclusive classroom for **all** students. If you need any accommodations, please contact Disability Services (visit their website, call 651-696-6275, or email disabilityservices@macalester.edu) to make an appointment 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 Title IX website. 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***, contact the Hamre Center (651-696-6275), chaplains (651-696-6298), or other local and national resources listed here.

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 appropriate accommodations.