Spring 2023 - PM 570: Statistical Methods in Human Genetics

COURSE INFORMATION:

Instructor:	Nicholas Mancuso, Ph.D.
E-mail:	nmancuso@usc.edu
Office hours:	TBD
Office location:	NRT G517G
Course Schedule:	Friday, 1pm – 5pm
Course Location:	SSB 111

CONTENT:

The aim of this course is to provide an introductory course in the statistical methods used in the analysis of human genetic data. Emphasis will be placed on applied analysis, but the underlying statistical theory will be presented and discussed when needed to help further understanding. We will also introduce and use the statistical environment R, scripting environment bash, and various tools (e.g., PLINK, GCTA, etc) to implement some of the methods covered.

Lectures and related material will be posted and updated on the blackboard system

http://blackboard.usc.edu

METHODS OF INSTRUCTION:

We will have lectures to provide general background and details on each topic. Lectures will be followed with a student-led presentation of a recent manuscript with a "journal club style" discussion.

ASSESSMENT:

Assessment will be based on a group-based project (60%), journal club presentation (30%), and class participation (10%).

Group-based project: Groups of 2-3 individuals will carry out a research project on selected topics. Research project can be the development of a novel method and its application to real or simulated data, or the application of an existing method to real data.

Journal-club presentation: Starting on the 4th week, a group of 2-3 individuals will present a manuscript and lead a journal-club style presentation and discussion. It is expected that the manuscript will be related to the topic introduced by the previous week's lecture.

Course final grades will be determined using the following scale:

A	95-100
A-	90-94
B+	87-89
B	83-86
B-	80-82
C+	77-79
C	73-76
C-	70-72
D+	67-69
D	63-66
D-	60-62

F	59 and below

TEXTBOOKS (*There is no required textbook*):

McCulloch CE, Searle SR, Neuhaus JM. *Generalized, Linear, and Mixed Models*. Wiley Series in Probability and Statistics, 2008.

Searle SR, Casella G, McCulloch CE. *Variance Components*. Wiley Series in Probability and Statistics, 2006.

Thomas DC. *Statistical Methods in Genetic Epidemiology*. Oxford: Oxford University Press, 2004.

Siegmund D., Yakir B. *The Statistics of Gene Mapping.* New York: Springer, 2007.

Stram, D. *Design, Analysis, and Interpretation of Genome-Wide Association Scans.* New York: Springer, 2014.

PREREQUISITES:

Talk to instructor. In general, PM533 (Genetic and Molecular Epidemiology), PM534 (Statistical Genetics) and a background in basic statistical analysis (PM 510, PM 511) are highly recommended.

General Topics and Syllabus:

Date	Topic
Week 1: 01/13/2023	**Lecture:** Intro to statistical genetics. Basic molecular genetics, linkage and association concepts. **Lab:** R/Linux tutorial. Allele frequency estimation.
Week 2: 01/20/2023	**Lecture:** Intro to classical methods in statistical genetics. **Lab:** Pedigree Analysis, Linkage analysis
Week 3: 01/27/2023	**Lecture:** Intro to genome-wide association studies (GWAS) Part I. **Lab:** Phenotype simulation. PCA, GLM
Week 4: 02/03/2023	**Lecture:** Intro to genome-wide association studies (GWAS) Part II. **Journal Club:** *TBD*
Week 5: 02/10/2023	**Lecture:** Intro to SNP heritability Part I **Journal Club:** *TBD*
Week 6: 02/17/2023	**Guest Lecture:** *TBD*
Week 7: 02/24/2023	**Lecture:** Intro to SNP heritability Part II **Journal Club:** *TBD*
Week 8: 03/03/2023	**Lecture:** Inferring SNP heritability from summary statistics **Journal Club:** *TBD*
Week 9: 03/10/2023	**Lecture:** Inferring SNP heritability from summary statistics **Journal Club:** *TBD*
Week 10: 03/17/2023	**!!!Spring Break!!!**
Week 11: 03/24/2023	**Lecture:** Intro to molecular-QTL analyses **Journal Club:** *TBD*

Week 12: 03/31/2023	**Lecture:** Intro to transcriptome-wide association studies (TWAS), Mendelian randomization **Journal Club:** *TBD*
Week 13: 04/07/2023	**Lecture:** Intro to transcriptome-wide association studies (TWAS), Mendelian randomization **Journal Club:** *TBD*
Week 14: 04/14/2023	**Final Project Presentation Pt I**
Week 15: 04/28/2023	**Final Project Presentation Pt II**

STATEMENT ON ACADEMIC CONDUCT AND SUPPORT SYSTEMS:

STATEMENT FOR STUDENTS WITH DISABILITIES:
Any student requesting academic accommodations based on a disability is required to register with Disability Services and Programs (DSP) each semester. A letter of verification for approved accommodations can be obtained from DSP. Please be sure the letter is delivered to me (or to TA) as early in the semester as possible. DSP is located in STU 301 and is open 8:30 a.m.–5:00 p.m., Monday through Friday. The phone number for DSP is (213) 740-0776.

STATEMENT ON ACADEMIC INTEGRITY:
USC seeks to maintain an optimal learning environment. General principles of academic honesty include the concept of respect for the intellectual property of others, the expectation that individual work will be submitted unless otherwise allowed by an instructor, and the obligations both to protect one's own academic work from misuse by others as well as to avoid using another's work as one's own. All students are expected to understand and abide by these principles. *Scampus,* the Student Guidebook, contains the Student Conduct Code in Section 11.00, while the recommended sanctions are located in Appendix A: http://web-app.usc.edu/scampus/gov/. Students will be referred to the Office of Student Judicial Affairs and Community Standards for further review, should there be any suspicion of academic dishonesty. The Review process can be found at: http://www.usc.edu/student-affairs/SJACS/.

Academic Conduct:
Plagiarism – presenting someone else's ideas as your own, either verbatim or recast in your own words – is a serious academic offense with serious consequences. Please familiarize yourself with the discussion of plagiarism in *SCampus* in Part B, Section 11, "Behavior Violating University Standards" https://policy.usc.edu/scampus-part-b/. Other forms of academic dishonesty are equally unacceptable. See additional information in *SCampus* and university policies on scientific misconduct, http://policy.usc.edu/scientific-misconduct.

Support Systems:
Student Counseling Services (SCS) - (213) 740-7711 – 24/7 on call (https://engemannshc.usc.edu/counseling/)
Free and confidential mental health treatment for students, including short-term psychotherapy, group counseling, stress fitness workshops, and crisis intervention.

National Suicide Prevention Lifeline - 1-800-273-8255 (http://www.suicidepreventionlifeline.org)
Provides free and confidential emotional support to people in suicidal crisis or emotional distress 24 hours a day, 7 days a week.

Relationship & Sexual Violence Prevention Services (RSVP) - (213) 740-4900 - 24/7 on call (https://engemannshc.usc.edu/rsvp/)
Free and confidential therapy services, workshops, and training for situations related to gender-based harm.

Sexual Assault Resource Center (http://sarc.usc.edu/)
For more information about how to get help or help a survivor, rights, reporting options, and additional resources.

Office of Equity and Diversity (OED)/Title IX compliance – (213) 740-5086 (https://equity.usc.edu/)
Works with faculty, staff, visitors, applicants, and students around issues of protected class.

Bias Assessment Response and Support (https://studentaffairs.usc.edu/bias-assessment-response-support/)
Incidents of bias, hate crimes and microaggressions need to be reported allowing for appropriate investigation and response.

Student Support & Advocacy – (213) 821-4710 (https://studentaffairs.usc.edu/ssa/)
Assists students and families in resolving complex issues adversely affecting their success as a student EX: personal, financial, and academic.

Diversity at USC – (https://diversity.usc.edu/)
Tabs for Events, Programs and Training, Task Force (including representatives for each school), Chronology, Participate, Resources for Students