ROLLINS SCHOOL OF PUBLIC HEALTH
EMORY

DEPARTMENT: **Biostatistics and Bioinformatics**

COURSE NUMBER: **BIOS 760R** **SECTION NUMBER:** **SEMESTER: SPRING**

CREDIT HOURS: 2

COURSE TITLE: **Advanced Statistical Genetics**

INSTRUCTOR NAME Yijuan Hu, PhD

INSTRUCTOR CONTACT INFORMATION

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SCHOOL ADDRESS OR MAILBOX LOCATION: GCR Building, room 342

OFFICE HOURS Tuesday 5:00pm-6:00pm or by appointment.

BRIEF COURSE DESCRIPTION

This course provides a comprehensive survey of the statistical methods that have been recently developed for the designs and analysis of genetic association studies. Specific topics include genome-wide association studies, likelihood inference and EM algorithm, case-control sampling and retrospective likelihood, secondary phenotypes in case-control studies, haplotypes and untyped SNPs, population stratification, meta-analysis, multiple testing, winner's curse, copy number variants, next-generation sequencing studies, rare variants and trait-dependent sampling.

This course is targeted primarily at the Ph.D. students and will be taught at a rigorous statistical level. The students will learn the theoretical justifications for the methods as well as the skills to apply them to real studies. They will also be exposed to current research topics and open problems.

Prerequisites:
BIOS 560R Introduction to Statistical Genetics
BIOS 511 Statistical Inference I
BIOS 711 Statistical Inference II
or instructor's permission.

LIST SCHOOL LEVEL, DEPARTMENT, AND/ OR PROGRAM COMPETENCIES

- Identify biostatistical aspects in contemporary public health issues
- Collaborate with investigators and statistical colleagues in the analysis of data from biomedical and public health studies
- Demonstrate advanced analytic skills within a collaborative setting
- Demonstrate technical accuracy with advanced analytic methods
- Apply new and existing statistical theory and methods as needed to address public health or medical problems
- Conduct complex statistical analyses for a broad range of applications

LIST LEARNING OBJECTIVES ASSOCIATED WITH THE COMPETENCIES

By the end of the course, students will be able to:

1. Identify statistical problems in comtemporary genetic studies
2. Understand and apply existing statistical theory and methods to genetic studies
3. Develop new statistical methods to solve new problems in genetic studies
4. Collaborate with investigators and statistical colleagues in the analysis of genetic data.

Suggested Textbooks
The Fundamentals of Modern Statistical Genetics (2011) by Nan M. Laird and Christoph Lange
Most lectures will draw materials from journal articles

EVALUATION

Participation (50%)

Journal article presentation (50%)

Tentative Schedule and Topics

Lecture 1	**Likelihood Inference** • Maximum likelihood estimation (MLE), EM algorithm • Nonparametric maximum likelihood estimation (NPMLE)	
Lecture 2	**Study Designs and Trait-Depedent Sampling** • Case-control designs, retrospective likelihood • Cohort, extreme-trait sampling	
Lecture 3	**Secondary Phenotypes Analysis in Case-Control Studies** • Standard methods • New advancements	
Lecture 4	**Haplotyping and Imputation** • Haplotype phasing algorithms • Imputation algorithms	
Lecture 4	**Anaysis of Hapotype-Disease Association** • Gene-environment independence/dependence	
Lecture 5	**Analysis of Untyped SNPs** • Maximum likelihood approach • Single imputation approach	
Lecture 7	**Population Stratification** • Genomic control • Principal components	
Lecture 8	**Meta-Analysis** • Inverse variance weighting • Meta- vs. Mega-analysis	
Lecture 9	**Adjustment for Multiple Testing** • Bonferroni, FDR • A new MCMC method	
Lecture 10	**Winner's Curse** • Effect estimation after significance testing	
Lecture 11	**Copy Number Variants (CNV)** • Copy number calling algorithms • Copy number association	
Lecture 12	**Association Analysis with Rare Variants** • CMC, Burden score, SKAT • Imputation of rare variants	
Lecture 13	**Gene-Set and Pathway Analysis** • Over representation analysis (ORA), Gene-Set Enrichment Analysis • Variance component and kernal machine methods	
Lecture 14	**Student Presentation of Journal Articles**	