

Rockefeller University
Welch – The Great Hall
1230 York Avenue, New York

ADVANCED GENE MAPPING COURSE May 22-26, 2023

The course will be held every day from 9:00AM - 5:00 PM The Great Hall of the Welch Building on the Rockefeller University campus, located at 1230 York Avenue (entrance at 66th Street). On Monday evening a social gathering will be held at The Rockefeller University Faculty Club.

Each session will start with a theoretical introduction followed by practical exercises. Instructors for the course are Heather Cordell (Newcastle University), Andrew Dewan (Yale University), Suzanne Leal (Rockefeller University & Columbia University), Shamil Sunyaev (Harvard University), and Gao Wang (Columbia University).

Judy Matuk will lecture on ethics and the regulation of human subject research. A special guest lecture will also be given by Jurg Ott (Rockefeller University).

MONDAY – May 22nd

Instructors – Heather Cordell (Morning) and Suzanne Leal (Afternoon)

Statistical framework for association testing common and rare variants (population and family based); linear and logistic regression; variance components analysis; linear mixed models (LMM) and generalized LMM; Controlling for population admixture and substructure; Data quality control of genotype and sequence data

Exercises: PLINK, GCTA, FaST-LMM, and VAT (Part 1)

Evening Program

5:10 PM Get-together for course participants and instructors at The Rockefeller Faculty Club

TUESDAY – May 23rd

Instructors - Heather Cordell (Morning) and Suzanne Leal (Afternoon)

Rare variant association analysis using exome and sequence data for population- and family-based data; rare variant association tests; meta-analysis, detecting gene x gene and gene x environment interactions, power and sample size analysis

Exercises: VAT (Part 2), PSEQ, REGENIE, Armitage-test for trend tool, and Cassi

Special Lecture - Responsible Conduct of Research

Instructor - Judy Matuk (4:00-5:00)

Conflict of interest, research ethics, human subjects, and data security

WEDNESDAY – May 24th

Instructor – Gao Wang (9:00 - 2:00 and 3:00-5:00)

Molecular QTL mapping; Transcriptome-wide association studies (TWAS); Estimation of heritability

Exercises: MR-JTI

Statistical Fine-mapping (concepts and application: trans-ancestry, functional annotation, colocalization and multi-trait analysis).

Exercises: LD-clumping and SuSiE

THURSDAY – May 25th

Instructor – Andrew DeWan (9:00-5:00)

Biological, mediated, and spurious pleiotropy; Methods to detect pleiotropy; Mediation analysis; Mendelian Randomization

Exercises: MultiPhen, PLINK2.0, MR-base

Lecture – Jurg Ott (4:15-5:00)

Association analysis of genotype patterns with digenic traits.

FRIDAY – May 26th

Instructor – Shamil Sunyaev (9:00-5:00)

Population and evolutionary genetics; prediction of variant functionality; incorporating functional information in rare variant association tests; analysis of epigenomic data; polygenic risk scores

Exercise: ANNOVAR, CADD, GERP, Polyphen-2, SIFT, LDpred2