

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

## ADVANCED GENE MAPPING COURSE

January 10 - 14, 2022

The course will be held every day from 9:00AM - 5:00 PM in 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 (HRP Consulting Group) 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 – January 10<sup>th</sup>**

Instructors – Heather Cordell (9:00-12:30) and Suzanne Leal (1:30-5:00)

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 2.0, GCTA-MLMA, FaST-LMM, and REGENIE

### **Evening Program**

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

### **TUESDAY – January 28<sup>th</sup>**

Instructor - Heather Cordell (9:00-12:00) and Suzanne Leal (1:00-3:50)

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: SKAT R library (power analysis), Armitage-test for trend tool, R 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 – January 12<sup>th</sup>**

Instructor – Gao Wang (9:00-5:00 PM)

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

Exercises: tensorQTL; FUSION; LD Score Regression (LDSC)

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

Exercises: LD-clumping, SuSiE and mvSuSiE

## **THURSDAY – January 13<sup>th</sup>**

Instructor – Andrew DeWan (9:00-3:50)

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

Exercises: MultiPhen, PLINK2.0, MR-base

Lecture – Jurg Ott (4:00-5:00)

Association analysis of genotype patterns with digenic traits.

## **FRIDAY – January 14<sup>th</sup>**

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, LDpred, etc