

ADVANCED GENE MAPPING COURSE

January 25 - 29, 2021

The course will be held every day via zoom. On Monday evening a social gathering will be held at via Zoom.

Each session will start with a theoretical introduction followed by practical exercises. The practical exercises will be performed using cloud computing. Each student will have their own account to perform the exercises during and up to three weeks after the completion of the course. Instructors for the course are Heather Cordell (Newcastle University), Nancy Cox (Vanderbilt 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 25th

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

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

Exercises: PLINK 2.0, GCTA-MLMA, FaST-LMM, REGENIE, Variant Association Tools (VAT), PSEQ

Evening Program

5:35PM Zoom Social – Get to know your colleagues

TUESDAY – January 26th

Instructor - Heather Cordell (9:00-12:00) and Suzanne Leal (1:00-4:00)

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, linear mixed models, power and sample size analysis

Special session on responsible conduct of research: data management (security) and ethical use of human research subjects

Exercises: VAT, PSEQ, SKAT R library (power analysis), Armitage-test for trend tool, R and Cassi

Special Lecture- Ethics and the Regulation of Human Subject Research

Presenter - Judy Matuk (4:00-5:30)

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

WEDNESDAY – January 27th

Instructor- Gao Wang (9:00-12:00)

Fine Mapping continued - expressionQTLs and multiple traits

Exercise: SuSie and LD-clumping

Instructor – Andrew DeWan (1:00-5:30)

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

Exercises: MultiPhen, PLINK2.0, MR-base

THURSDAY – January 28th

Instructor – Nancy Cox (9:00-3:00)

Linear mixed models; eQTL mapping; predication models using RNAseq and array data; challenges and inferences for heritability estimation and prediction

Exercises: Maxtix-eQTL, GCTA-MLMA, PrediXcan

Instructor- Gao Wang (3:00 -4:00)

Fine Mapping - continued

Special lecture- Frequent Pattern Mining (FPM) Methods for Finding SNP-SNP Interactions in the Absence of Single-SNP Effects

Presenter - Jurg Ott (4:00-5:00)

FRIDAY – January 29th

Instructor – Shamil Sunyaev (9:00 - 5:30)

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

Exercise: Polygenic risk score prediction using nonparametric shrinkage. LDpred, ANNOVAR, CADD, GERP, Polyphen-2, SIFT, etc