

Association Mapping: GWAS and Sequencing Data

Instructors: Joelle Mbatchou and Loic Yengo

Summer Institute in Statistical Genetics (SISG)

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Introduction: Course Goals

This is a course on statistical methods and software for genetic association studies of complex traits. We aim to cover:

- ▶ Genetic Association Testing with Case-Control & Quantitative Traits
- ▶ Population Structure/Ancestry Inference
- ▶ Genetic Association Testing in Samples with Structure
- ▶ Conditional analyses, Colocalization, Fine-mapping & Gene and Pathway Level Analysis
- ▶ Polygenic Risk Scores
- ▶ Association Testing for Rare Variant Analysis
- ▶ Interaction Analysis, GWAX, Time-to-event & Multi-trait Analysis
- ▶ Power and Sample Size, Design Considerations and Emerging Issues

Introduction: Resources

Importantly, the class site is:

https://joellembatchou.github.io/SISG2023_Association_Mapping/

Contains (or will contain):

- ▶ Link to PDF copies of slides
- ▶ Practical exercises for you to try
- ▶ Link to datasets used in exercises
- ▶ Our solutions to exercises (later!)
- ▶ Links to software packages

Introduction: About Loic



- ▶ Statistical Geneticist,
the University of Queensland, Australia
- ▶ Research interests:
 - ▶ GWAS of anthropometric traits
 - ▶ GWAS transferability
 - ▶ Non-random mating in humans

Introduction: About Joelle



- ▶ Statistical Geneticist,
Regeneron Genetics Center
- ▶ Research interests:
 - ▶ Genetic Association Studies
 - ▶ Genetic Data with Structure
 - ▶ Mixed Models methods
 - ▶ Association methods for large-scale datasets

Slack Channel

Slack channel for class:

<https://uwbiostatisticssisg.slack.com/archives/C05EB2ZDDSN>

Will contain:

- ▶ Key announcements
- ▶ Link to the class website
- ▶ PDF copies of slides

Cloud server

We will use a cloud server to do the practical exercises. For more info on getting set up on the server:

https://joellembatchou.github.io/SISG2023_Association_Mapping/using_server.html

Things to note:

- ▶ **Let us know if you cannot access the server**
- ▶ We will run exercises on the server
- ▶ Datasets used in practicals are at:
/data/SISG2023M15/data/
- ▶ RStudio server can be accessed at:
<http://si2023-compute.biostat.washington.edu:8787/>

Introduction: Course Structure

- ▶ Full schedule on class website (**Seattle time, PDT**)
- ▶ 10 sessions, 60-90 minutes each, over 2.5 days
- ▶ What to expect in a typical session;
 - ▶ 45 mins teaching/lecture
 - ▶ 30 mins hands-on exercises
 - ▶ 15 mins summary/discussion
- ▶ For the practicals:
 - ▶ Students will be split into smaller groups
 - ▶ You can run them on the server or your own machine