CS 291 - **PERSONAL GENOMICS FOR BIOINFORMATICIANS** - TR 3:30-4:50 MCGIL 2315
Instructors: Melissa Gymrek OH: WF 3:00-4:50 CSE 4216

COURSE DESCRIPTION

Genome-sequencing is quickly becoming a commodity, and more than a million people have already analyzed their own genomes through direct-to-consumer companies. This course provides an introduction to current bioinformatics techniques for analyzing and interpreting human genomes. We will learn how to interpret a single genome in the context of an entire population, based on the often quoted concept: interpreting one genome requires tens of thousands of genomes. Topics covered include an introduction to human medical and population genetics, human ancestry, finding and interpreting disease-causing variants, genome-wide association studies, genetic risk prediction, analyzing next generation sequencing data, and how to scale current genomics techniques to analyze hundreds of thousands of genomes. We will also discuss the social impact of the personal genomics revolution.

Class meetings will consist of short lectures, journal club discussions, and hands on experience using and developing bioinformatics methods. All details of the course will be posted at the course website:
https://gymreklab.github.io/teaching/personal_genomics/personal_genomics_2017.html

SCHEDULE

Module	Date	Lecture	Homework
Intro to personal genomics	01-05 (Th)	Introduction to your genome	PS1 out
	01-10 (T)	Social impact of personal genomics	PS1 due
What can I do with my genome?	01-12 (Th)	Basic population genetics	PS2 out
	01-17 (T)	Determining ancestry	
	01-19 (Th)	Phasing and imputation	
	01-24 (T)	Web 2.0 Genomics	PS2 due
Complex traits	01-26 (Th)	Introduction to GWAS	PS3 out
	01-31 (T)	Controlling for confounders	Project out
	02-02 (Th)	Scaling GWAS to millions of genomes	
	02-07 (T)	Missing heritability	PS3 due
Next-gen sequencing and big data	02-09 (Th)	Introduction to NGS	PS4 out
	02-14 (T)	Short read alignment strategies	Proposal due
	02-16 (Th)	Variant calling from NGS	
	02-21 (T)	Functional genomics - RNAseq, ChIPseq	PS4 due
	02-23 (Th)	Storing, querying, visualization	
	02-28 (T)	Long read sequencing technologies	
Mutation hunting	03-02 (Th)	Introduction to genetic mapping	PS5 out
	03-07 (T)	Filtering and prioritizing variants	
	03-09 (Th)	*De novo* mutations and constraint	
	03-14 (T)	Interpreting non-coding variants	PS5 due
Final Project	03-16 (Th)	Project presentations	Project due

GENOTYPING FAQ

The goal of this course is to teach you how to analyze your genome. At this time, we are not able to provide genotyping or sequencing services to students, and instead homeworks will be performed on publicly available human genomes.

You are welcome to analyze your own genome using a direct-to-consumer service such as 23andme or Ancestry.com. I recommend 23andme, which performs SNP genotyping arrays for $200. You will also get access to a range of fun tools on their website that will complement some of the analyses we do in class. Homeworks for modules 1-3 are built to analyze the type of data available from 23andme.

Please keep in mind the following:

- It is **NOT** required that you analyze your own genome. Your grade is in no way influenced by whether or not you participate in 23andme.

- To ensure the previous point, **do not tell me whether you are analyzing your own genome.**

- If you do 23andme, it is at your own risk and at your expense. We do not have funding to pay for the test.

- While you are free to analyze your genome, you must still complete the homework assignments with the provided genomes as well.

- 23andme can take up to six weeks to get your data back, so sign up early.

GRADING

Component	Percentage
Participation	10%
Attendance	10%
PS1	5%
PS2	10%
PS3	10%
PS4	10%
PS5	10%
Project proposal	5%
Final project	35%
	100%