CMPB 5005 Functional Interpretation of High-Throughput Data

Course Instructors:

Andrea Sboner, PhD, ans2077@med.cornell.edu Jan Krumsiek, PhD, jak2043@med.cornell.edu Doron Betel, PhD, dob2014@med.cornell.edu

Course Description:

Vast amounts of biological and medical data are generated by high-throughput techniques, but these data sets require careful analysis and interpretation. It is, for example, challenging to the decipher the functional consequences of the myriad variants obtained from whole genome sequencing.

In this course, students will learn applications of high-throughput screens across different disciplines, in both basic and clinical research. The course will focus on the interpretation of data from Next-Generation Sequencing technologies, epigenome data, proteomics data, and metabolomics data. During the course, students will learn how to apply computational tools to data to guide illumination of biological function.

Learning Objectives:

By the end of the course, students will be able to:

- Have a deep appreciation of how biological and medical data from various highthroughput technologies requires careful analysis and interpretation
- Have a deep appreciation of the complexity of the human genome
- Learn tools for functional interpretation of a variety of data sets and learn to critically assess results obtained from these tools

Course Format: lectures, computer labs, and journal club presentations

Semester: Spring

Prerequisites: CMPB 5004 *Analysis of Next-Generation Sequencing Data* (can be taken concurrently) or familiarity with Next-Generation Sequencing and R.

Credits: 4

Zoom: https://weillcornell.zoom.us/j/94905544629

This is an in-person course. The Zoom link is for students unable to participate in person. Students must notify the module director of in-person absence.

Grading: Pass/fail

Time: Wednesdays 3-5pm

Location: WCMC C611

TAs: Annalise Schweickart, aas4002@med.cornell.edu Jonathan Rub, jor4008@med.cornell.edu

Schedule:

Module 1, Weeks 1-5: Precision Medicine Module Director: Andrea Sboner, PhD

In this module, students will be introduced to precision medicine with an overview of several high-throughput methods, e.g., next-generation sequencing, and computational approaches used in this context. Students will learn what key elements to consider when designing a research experiment or interpreting clinical results. This module is structured with a lecture and "journal club" presentations by students (in groups) with discussions.

- 1/4: Introduction to next-generation sequencing (NGS) for precision medicine
- 1/11: Processing and analysis of NGS data
- 1/18: Repositories and knowledge bases for the interpretation of the results
- 1/25: Integration of NGS data into clinical practice
- 2/1: The future of precision medicine: AI, mixed reality, etc.

Module 2, Weeks 6-11: Pathways and Networks Module Director: Jan Krumsiek, PhD TA: Annalise Schweickart

In this module, we will talk about applied metabolomics data analysis, pathway analysis, and network analysis. The module contains hands-on coding parts, where students replicate analyses from published research papers.

- 2/8: Metabolomics
- 2/15: coding: Metabolomics
- 2/22: Pathway Analysis
- 3/1: No class, spring recess
- 3/8: coding: Pathway Analysis
- 3/15: Network Analysis
- 3/22: coding: Network Analysis

Module 3, Weeks 12-16: Epigenetics, single cell analyses Module Director: Doron Betel, PhD TA: Jonathan Rub This module will focus on specialized omics topics, including single-cell genotyping and phenotyping, lineage tracing, and spatial transcriptomics. Students will do a final project.

3/29: TBD
4/5: TBD
4/12: TBD
4/19: TBD
4/26: TBD