Principles and Applications of Modern DNA Sequencing

EEEB GU4055

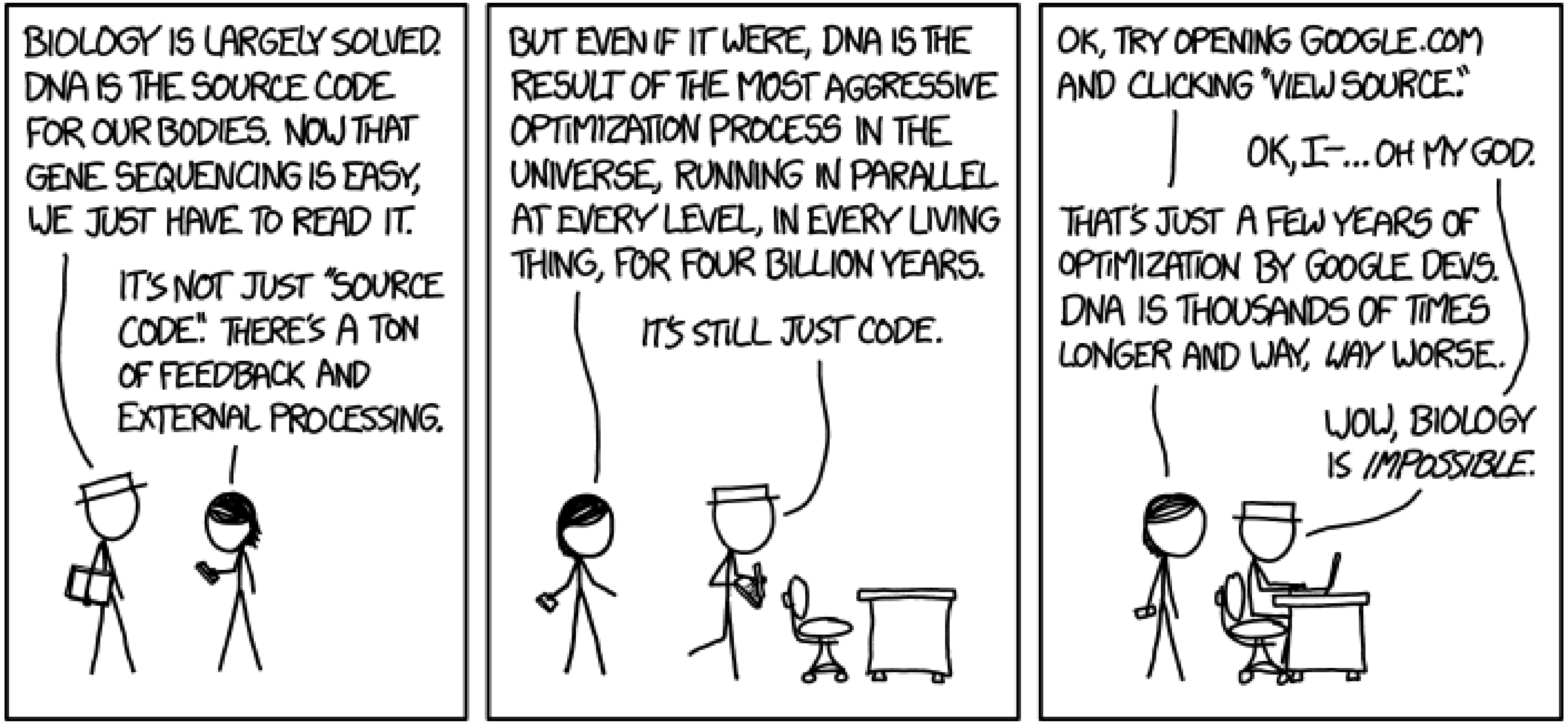
Session 1: Introduction

Today's topics

1. Introductions
2. Syllabus
3. Class structure
4. Computational resources

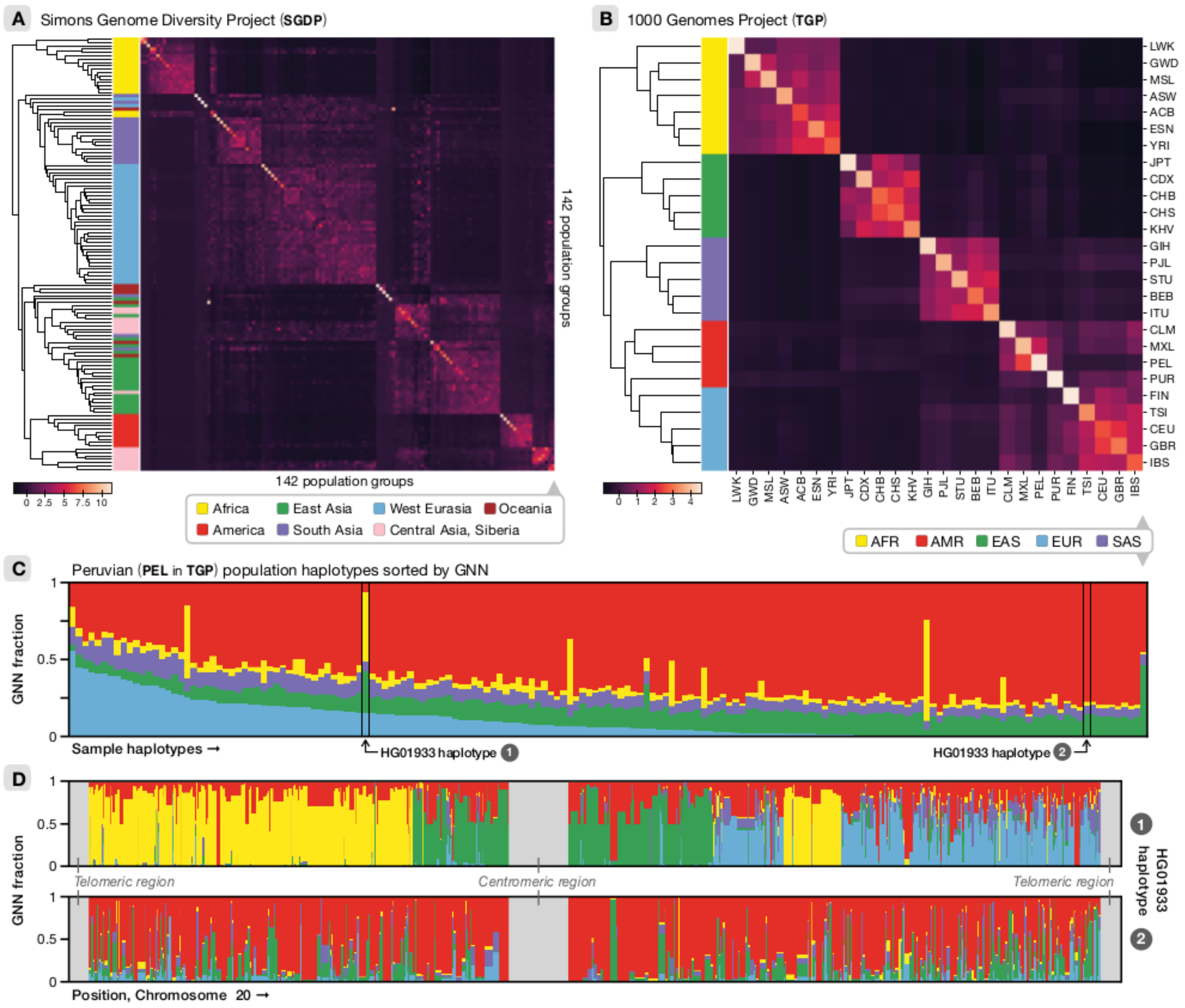
Learning objectives in this courese

To understand that **genomes are data** -- a set of instructions, and a record of history -- and to learn to use this information to test hypotheses.



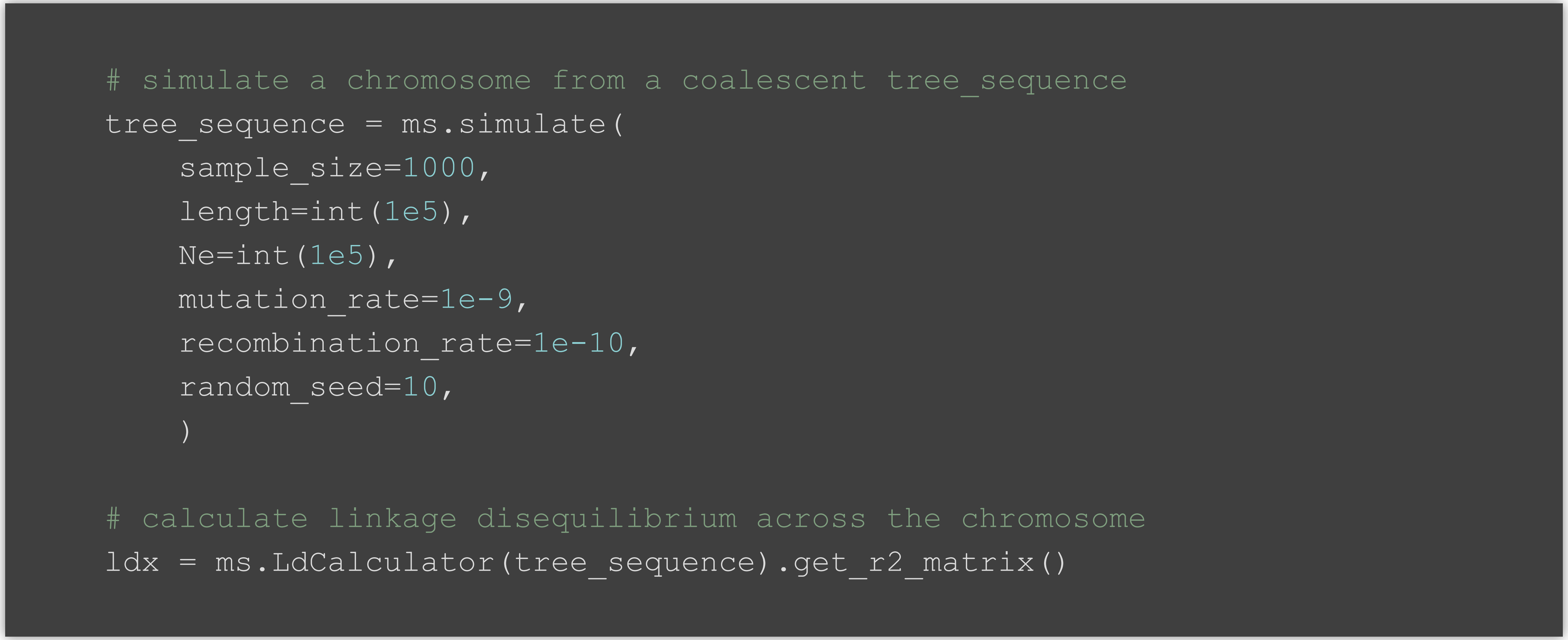
Learning genomics from the primary literature

We will read and discuss empirical papers and reviews of the application of genomics methods for studying evolution and medicine.



Learn genomics through hands-on computational exercises

We will use code exercises to see and touch real genomic data to understand how biological processes and information are translated and interpreted as data.

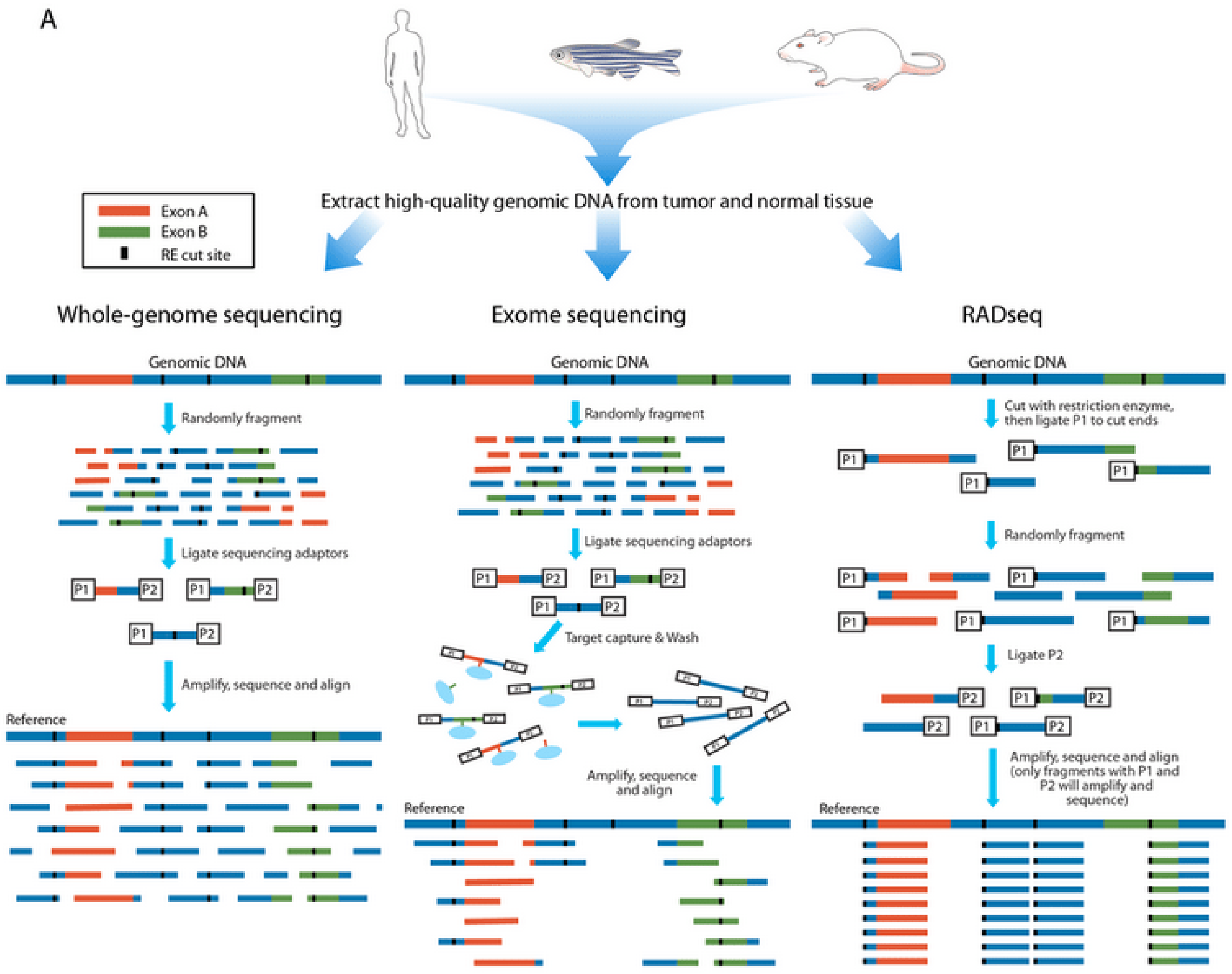


```
# simulate a chromosome from a coalescent tree_sequence
tree_sequence = ms.simulate(
    sample_size=1000,
    length=int(1e5),
    Ne=int(1e5),
    mutation_rate=1e-9,
    recombination_rate=1e-10,
    random_seed=10,
    )

# calculate linkage disequilibrium across the chromosome
ldx = ms.LdCalculator(tree_sequence).get_r2_matrix()
```

Learn about modern genomics technologies

We will discuss state-of-the-art technologies. Why are these methods useful, what came before, and what is coming next? Why should you choose one method over another?



In summary: Learning objectives

Learn to design, conduct, and analyze genomic experiments. By the end of class you should be able to:

- Describe the structure of genomes; what information can be extracted.
- Choose appropriate technologies for genomic experiments.
- Analyze genomic data using computational methods.

When poll is active, respond at **PollEv.com/dereneaton004**

What is your interest in genomics: enter keyword technologies

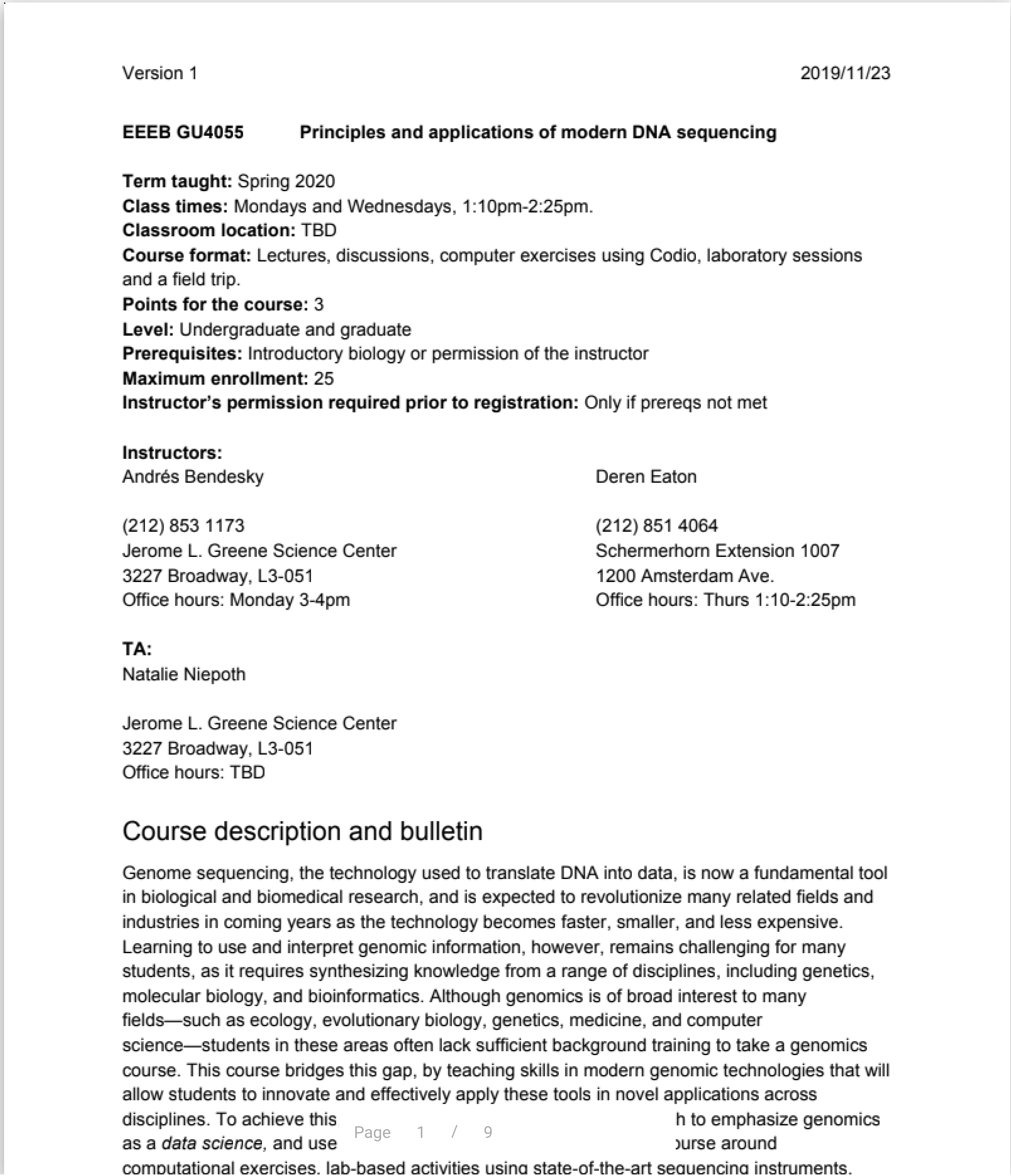
Class format: In each class we will

1. Discuss previous reading and review previous assignments.

2. Introduce new topics.

3. Assign readings and assignments on the new topic.

- Mon. assigned work load will be light, Wed. will be intensive.
- Assignments are due before the start of next class, else score=0.



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EEEB GU4055 Principles and applications of modern DNA sequencing

Term taught: Spring 2020
Class times: Mondays and Wednesdays, 1:10pm-2:25pm.
Classroom location: TBD
Course format: Lectures, discussions, computer exercises using Codio, laboratory sessions and a field trip.
Points for the course: 3
Level: Undergraduate and graduate
Prerequisites: Introductory biology or permission of the instructor
Maximum enrollment: 25
Instructor's permission required prior to registration: Only if prereqs not met

Instructors:

Andrés Bendesky	Deren Eaton
(212) 853 1173	(212) 851 4064
Jerome L. Greene Science Center	Schermerhorn Extension 1007
3227 Broadway, L3-051	1200 Amsterdam Ave.
Office hours: Monday 3-4pm	Office hours: Thurs 1:10-2:25pm

TA:
Natalie Niepoth

Jerome L. Greene Science Center
3227 Broadway, L3-051
Office hours: TBD

Course description and bulletin

Genome sequencing, the technology used to translate DNA into data, is now a fundamental tool in biological and biomedical research, and is expected to revolutionize many related fields and industries in coming years as the technology becomes faster, smaller, and less expensive. Learning to use and interpret genomic information, however, remains challenging for many students, as it requires synthesizing knowledge from a range of disciplines, including genetics, molecular biology, and bioinformatics. Although genomics is of broad interest to many fields—such as ecology, evolutionary biology, genetics, medicine, and computer science—students in these areas often lack sufficient background training to take a genomics course. This course bridges this gap, by teaching skills in modern genomic technologies that will allow students to innovate and effectively apply these tools in novel applications across disciplines. To achieve this h to emphasize genomics as a *data science*, and use ourse around computational exercises, lab-based activities using state-of-the-art sequencing instruments.

Project proposal

Propose a novel use/question/investigation using a modern genomic technology; or propose an idea for a new technology/method, how it would work, and why it would be useful.

This activity will require synthesizing knowledge about technologies we have learned, and about the data contained within genomes.

Field trip and report

Black Rock Forest Hands-on Portable Genomic Sequencing in the Field

4/17-4/18 (Fri-Sat) Let us know immediately if you cannot make it.

Grading

Assignments (50%)

Midterm (15%)

Participation/Quizzes (15%)

Project Proposal (5%)

Project Presentation (5%)

Final trip report (10%)

Our policy on working in groups

You can discuss the assignment with each other, including on the course chatroom on Courseworks. However, you should not post complete answers on the chatroom, and you cannot work together in groups to complete assignments or share answers. We have office hours available between each class where you should seek extra help with assignments.

Introduction to bash/jupyter/the-cloud

Throughout this course will assign *online computational notebooks* to complete between sessions. These are called jupyter notebooks, which combine text and code together into a single document. They are a great tool for teaching and for doing science.

Codio, binder, and cloud hosting

The focus of this class is on genomics. Coding and bioinformatics are an integral part of genomics, and so we will use them as a tool to learn more about the subject. However, this is not a computer science course. We do not require you to have prior coding experience. We will not require you to install any software on your computer.

To make it as easy as possible to jump right into doing science we are hosting all of the assignments on cloud-based servers. This means you will be able to login to complete your assignments online without having to install anything on your computer.

You should have access to codio: https://codio.com
And we will also use a free alternative, binder: example

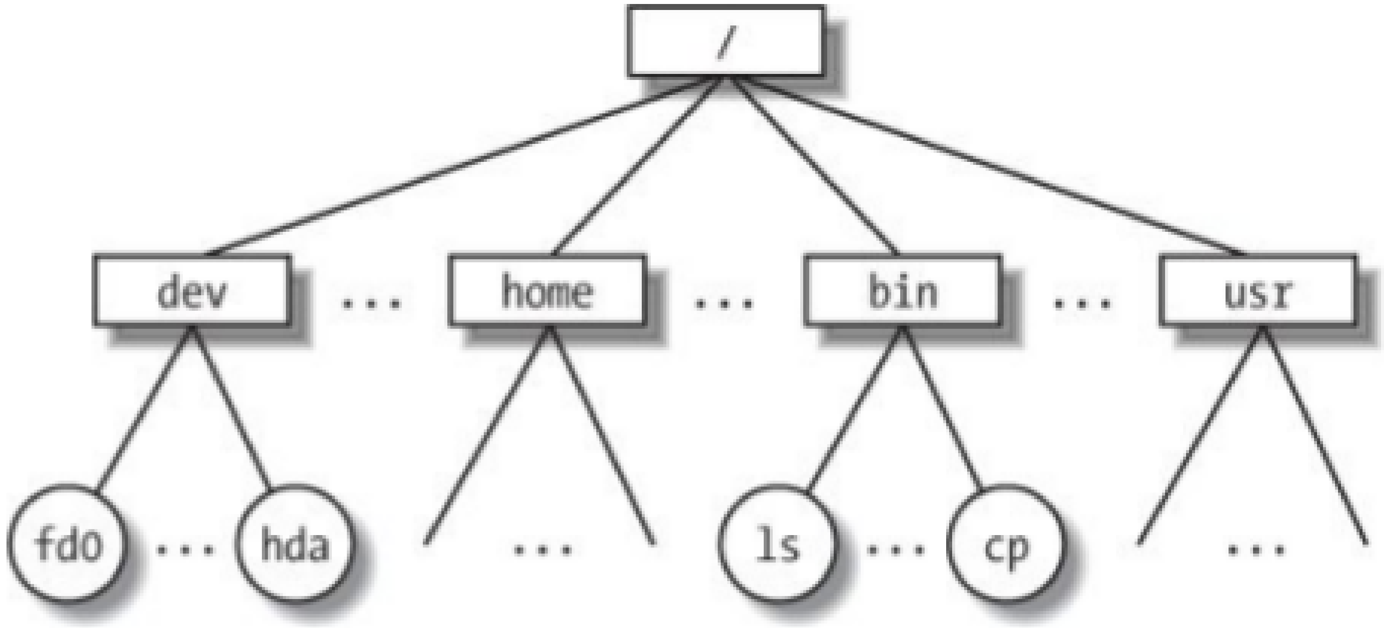
Introduction to the bash terminal

The system is composed of a hierarchical file system, just like the folder within folders in your own computer. There is a way of specifying the location of any file on your computer with text by describing its *path*.

```
# The root (top) of the entire filesystem (used for writing full paths).
$ /

# Here, in my current directory (used for writing relative paths).
$ ./

# Up one directory from my current directory (a relative path).
$ ../
```

Hierarchical file system

The beginning of the path starts at the *root*, which is represented by a forward slash (/). From there you can see file and folders of your system, as well as folders leading to your personal file. When you open a terminal you are located somewhere in this file system. You can ask where am I? What is here?



The bash command line

Bash is a language for interacting with your system from a terminal. From bash you can call a large number of software programs (which we will learn about) to accomplish a large number of tasks, including data analysis.

```
# the common syntax of bash commands
$ [program name] [-options] [target]

# an example with the program 'ls'
$ ls -l ./

# the same without using the optional flag -l
$ ls ./

# the same without the optional target (it uses the default target ./)
$ ls
```

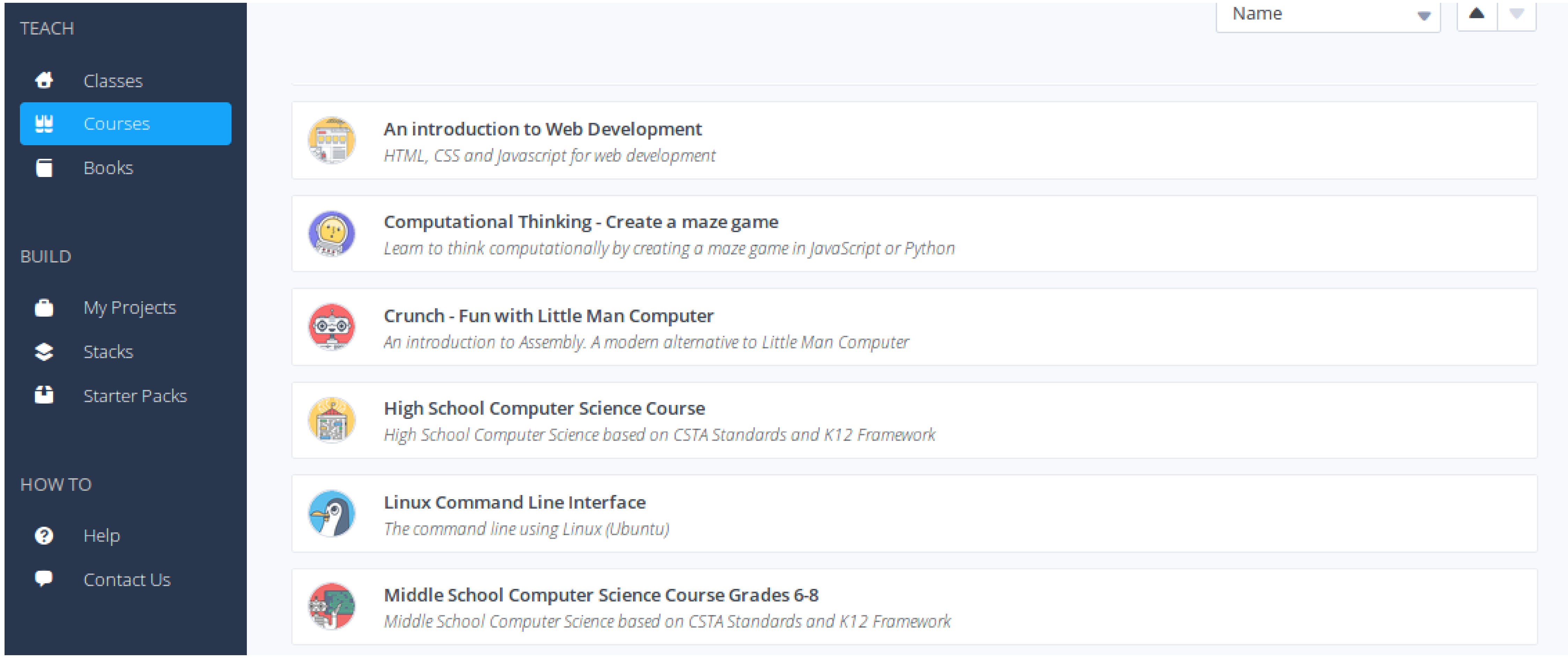
Hierarchical file system

You should always know where you are in the filesystem. This is bioinformatics skill number one. You need to know where your data is located to anything with it.

```
# show the files in your current directory
$ ls -l

# show the files in a different location on the filesystem
$ ls -l /bin/

# move yourself to a new location. This becomes your new cur dir.
$ cd folder

# print the path to your current location
$ pwd
```

Learning bash command line tools

There are many great tutorials, and google always has an answer. If you have zero experience in using a terminal then you may want to complete the Linux Command Line Tutorial on Codio, listed under the Courses tab on the left.



An introduction to Web Development
HTML, CSS and Javascript for web development

Computational Thinking - Create a maze game
Learn to think computationally by creating a maze game in JavaScript or Python

Crunch - Fun with Little Man Computer
An introduction to Assembly. A modern alternative to Little Man Computer

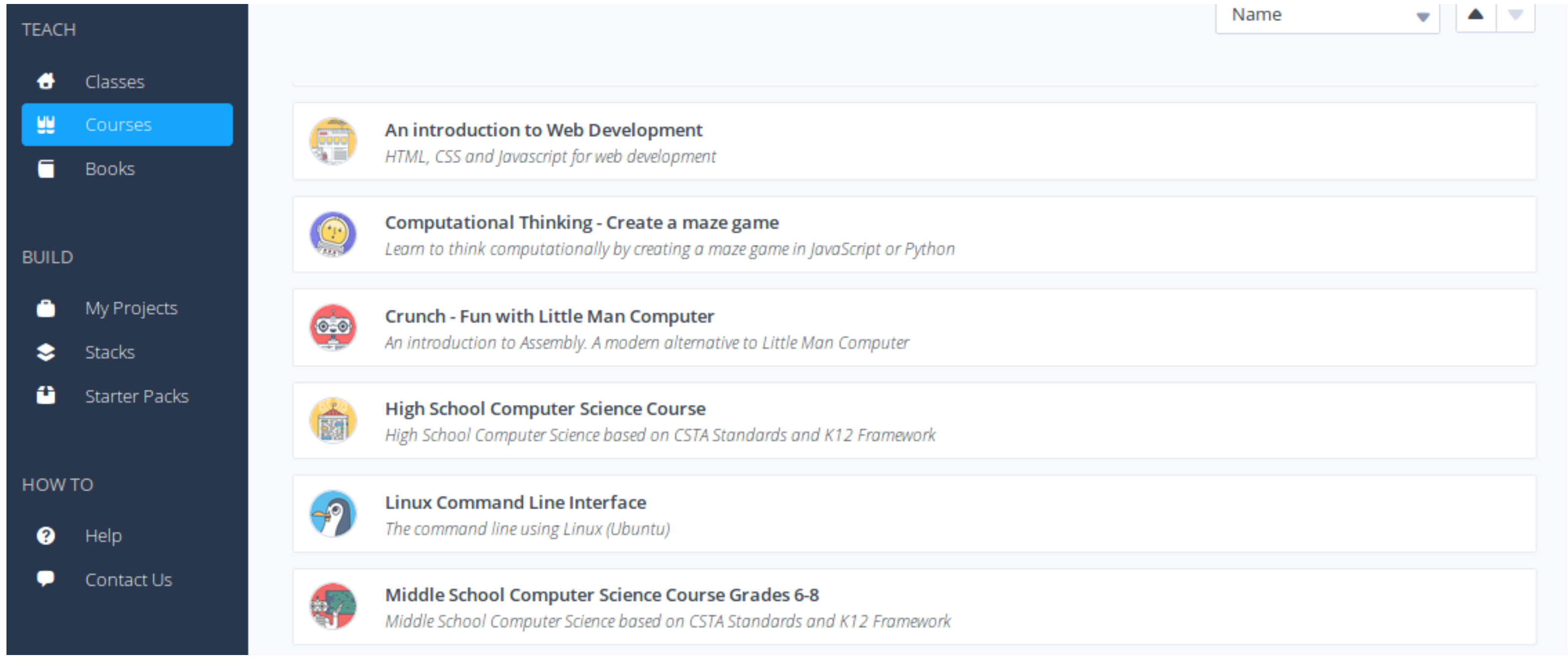
High School Computer Science Course
High School Computer Science based on CSTA Standards and K12 Framework

Linux Command Line Interface
The command line using Linux (Ubuntu)

Middle School Computer Science Course Grades 6-8
Middle School Computer Science based on CSTA Standards and K12 Framework

Your assignment for Monday

You have several notebooks to complete and an assigned paper to read.



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