

Teaching genomics and command-line basics at a primarily undergraduate institution using browser-based activities

Bárbara D. Bitarello

Resources and Programs for
Undergraduate Education in
Genomics, PAG, 2024

Slides!



About me

- 2021-Present: Assistant professor at Bryn Mawr College (BMC), a small women's liberal arts college
- Research: evolutionary & statistical genomics (humans and other primates)
- Bitarello (dry) Lab: currently 7 undergraduate researchers working on diverse projects in evolutionary & statistical genetics & phylogenetics
- Teaching:
 - 100-level: Intro Bio
 - 200-level: Genomics (6h/week, 1/2 lab), Biostatistics with R
 - 300-level: Evolutionary Genetics & Genomics

Outline

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 - A. A soft-introduction to the command line and FASTQ files
 - B. The Genomics Education Partnership (GEP) and how I've adapted and contributed materials
- Bonus:** A quick mention about a third project involving R programming!

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Browser-only activities bypass all of these hurdles!

Examples

- Biostatistics with R: Posit (Studio) Cloud ✓
- Genomics: UCSC Genome Browser, Galaxy, etc ✓

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But what about running software and learning about the command-line?

PROJECT 1: A SOFT INTRODUCTION TO THE COMMAND-LINE AND FASTQ FILES

Motivation

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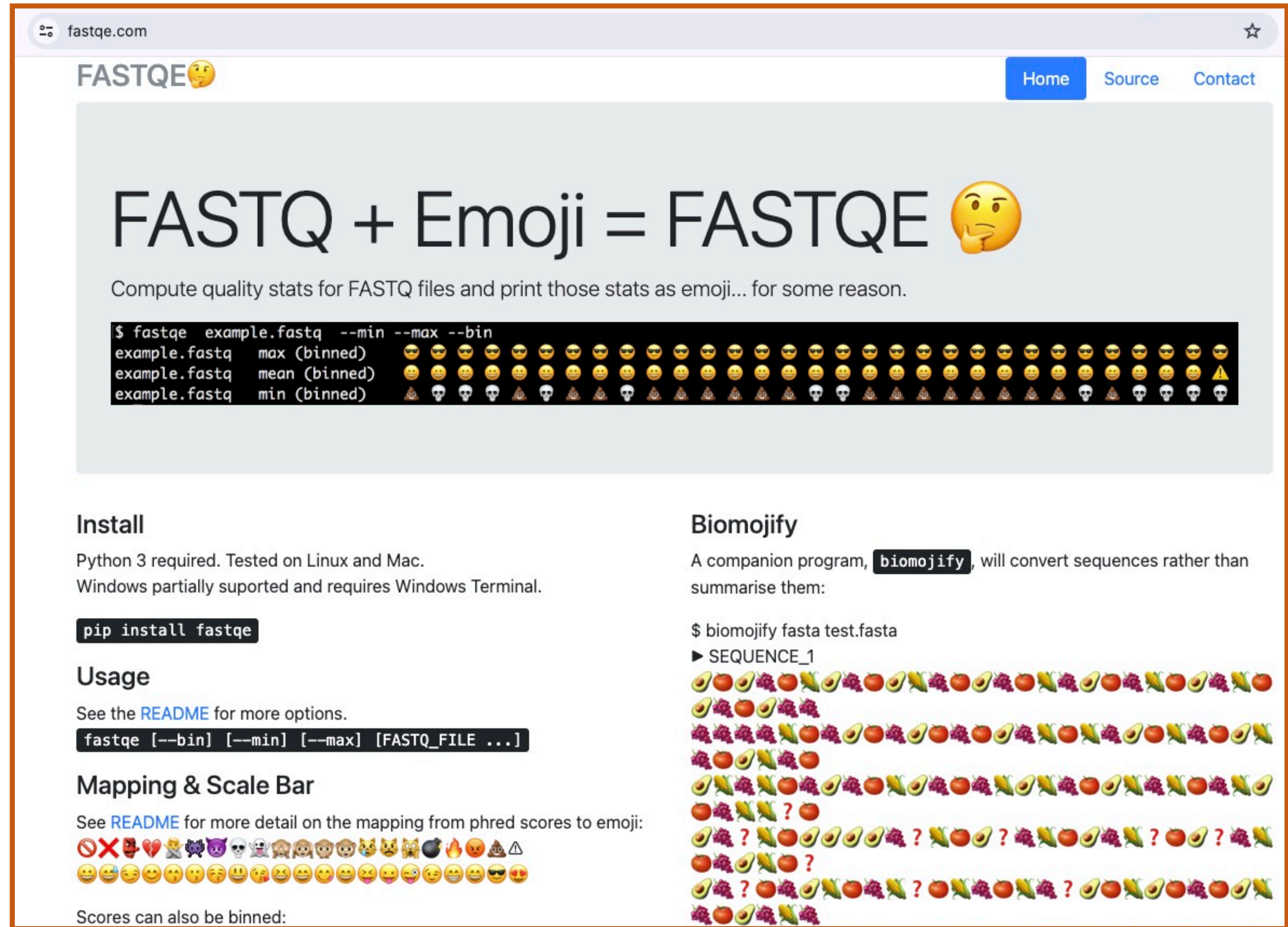
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Motivation

- Excellent materials from St. Jaquest et al. (2021), published in *CourseSource*
- Introduces the command-line and FASTQ files by using the FASTQE software

FASTQE: FASTQ + EMOJI

- Official Page:
fastqe.com
- Github:
github.com/fastqe/fastqe
- St. Jacques et al. (2021). CourseSource.
doi.org/10.24918/cs.2021.17



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- 2022: lost one entire class installing locally for each students and one student still could not get it to work; Cyverse did not work for any of them despite many efforts over several days
- FASTQE on galaxy: did not have time to test; removes the command-line experience

UPDATED IMPLEMENTATION

Solution: using mybinder.org

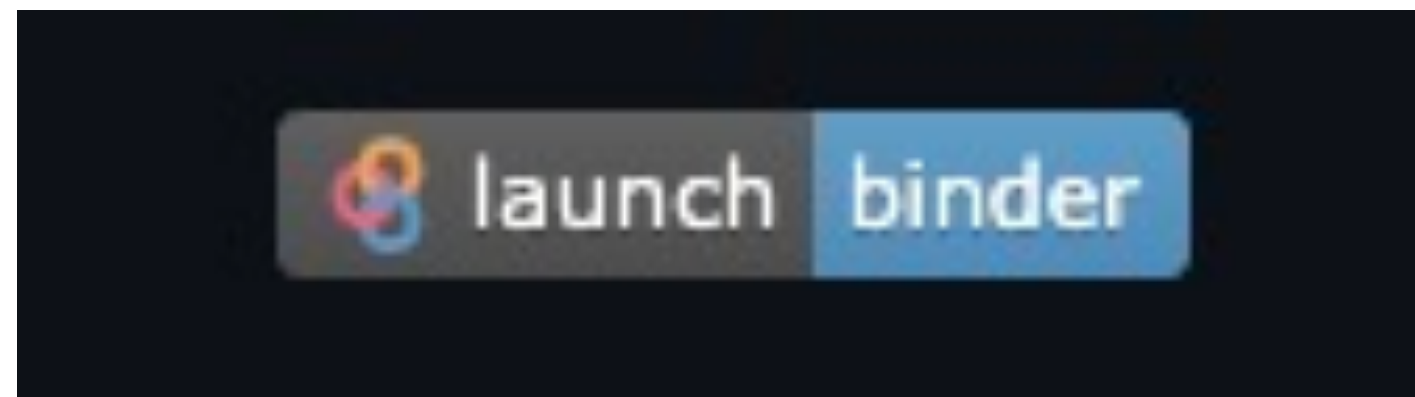
- Binder allows you to create **custom computing environments** that can be shared and used by many remote users.
- A Binder service is powered by [BinderHub](https://BinderHub.com), an open-source tool.
- One such deployment lives at mybinder.org, and is free to use.

Freely available

- The Github repo https://github.com/bitarellolab/Genomics_Teaching contains:
 - The code in R markdown and html formats
 - A slide deck for students
 - A direct link to launch the url for the activity

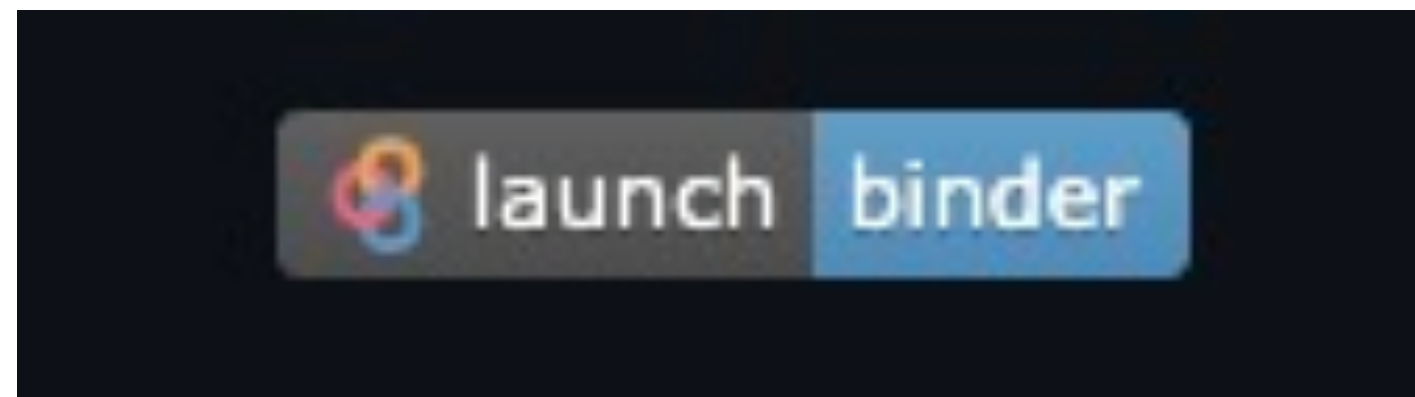
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- Shortened link I made for this presentation (try it!):
<http://tinyurl.com/33wkwjwt>

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- `ls`

- `mkdir`

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- `pwd`

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2) Have a soft introduction to FASTA and FASTQ files

3) Build an intuition around next-generation sequencing quality scores based on emojis

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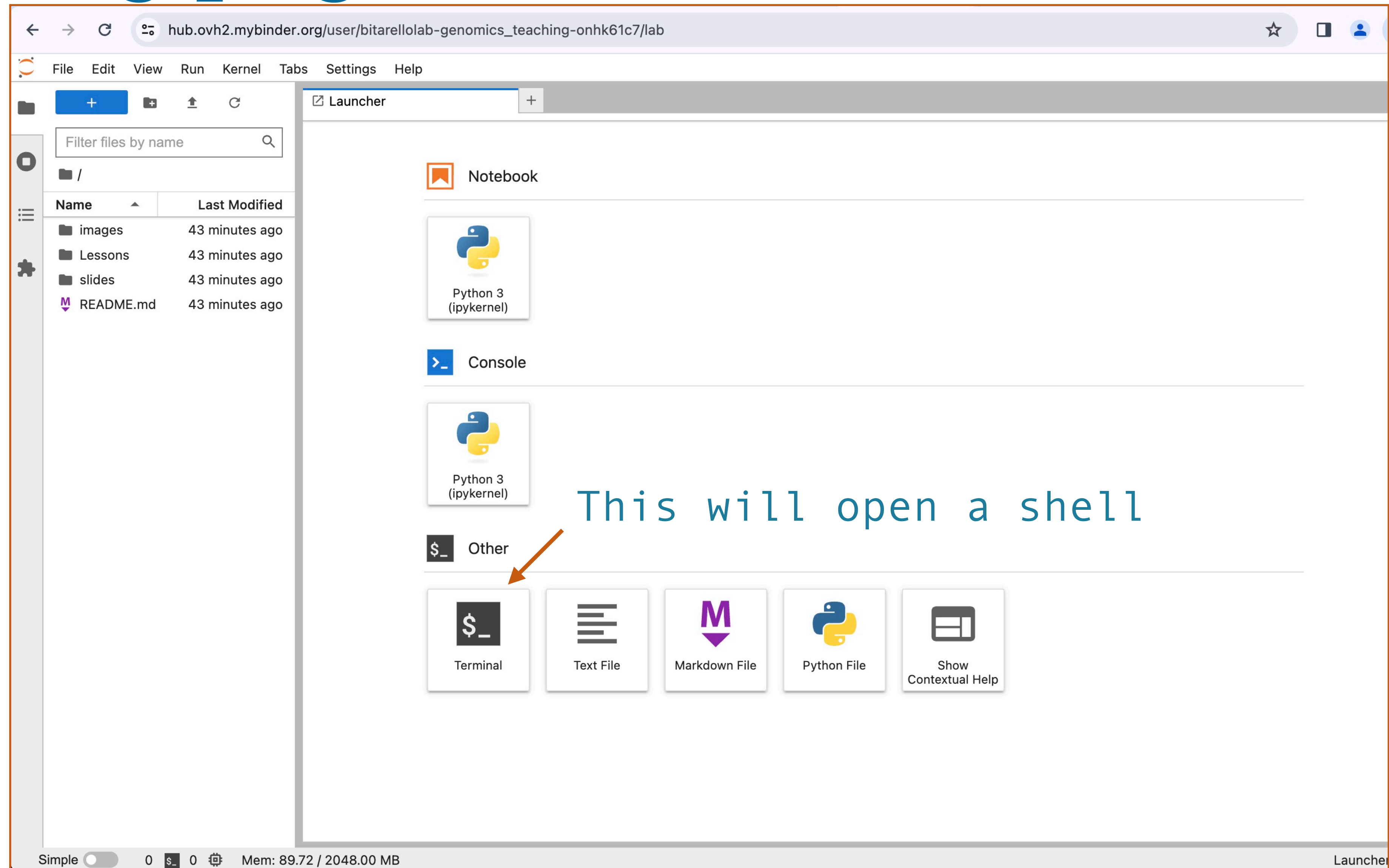
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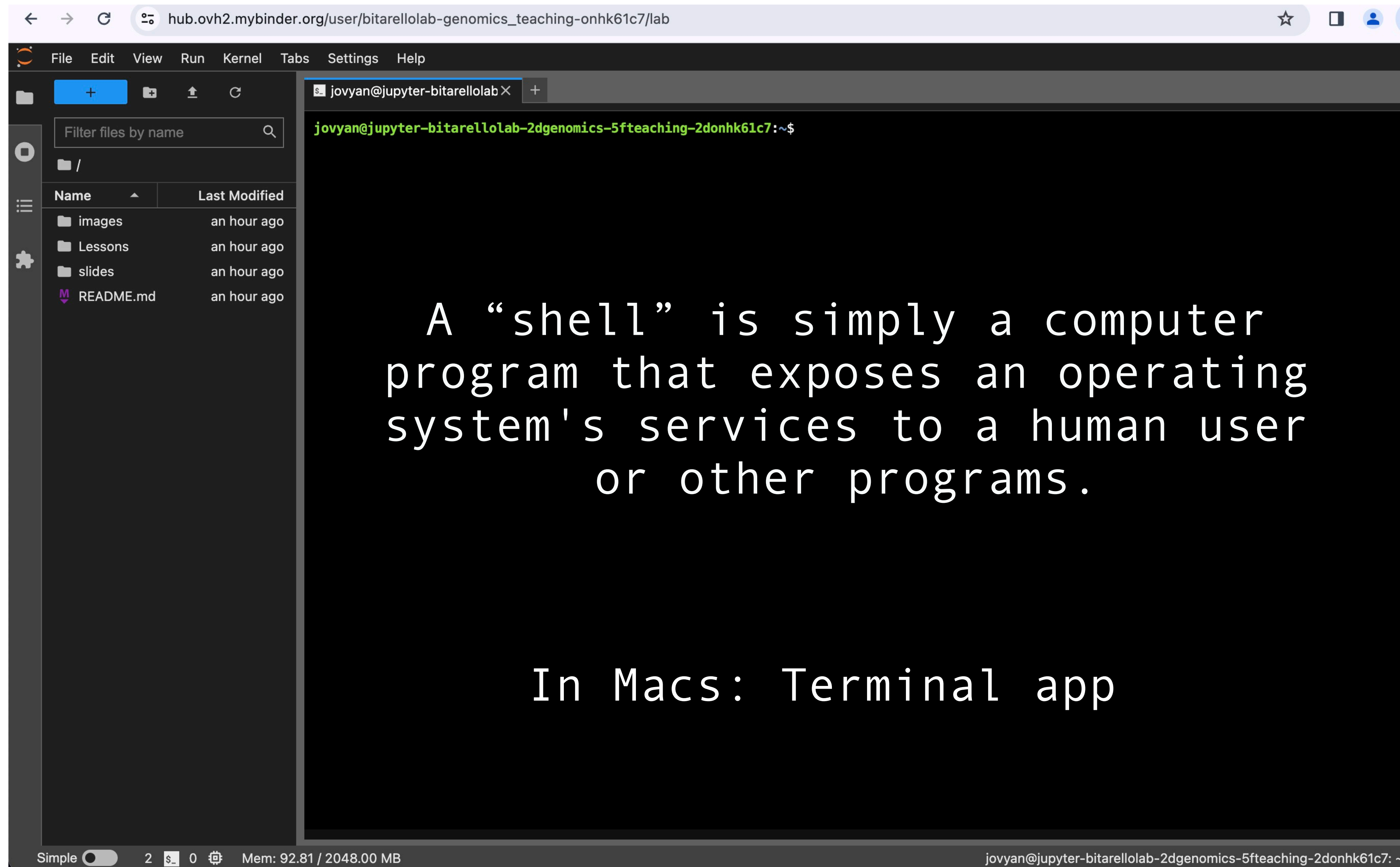
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 - Bash Basics tutorial
 - FASTQE tutorial
- Students hand in answer sheet at the end of class
- Later in the semester: problem set question where they had to go back to this and analyze different sequence files

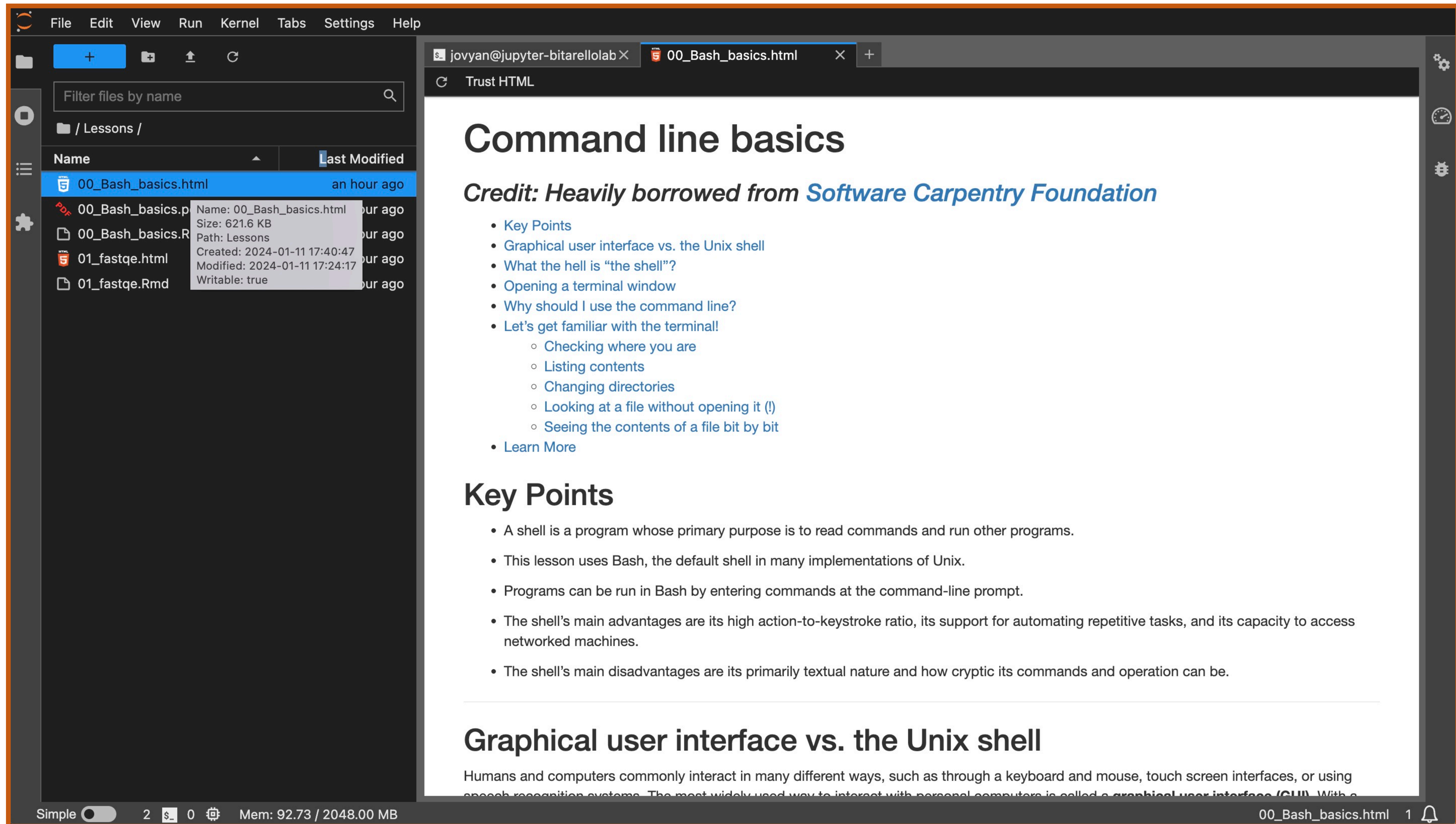
Landing page



A shell is now open



Soft introduction to the command line



The screenshot displays the JupyterLab interface. On the left, the file browser shows a directory named 'Lessons' containing several files. The file '00_Bash_basics.html' is selected, and its details are shown in a tooltip: Name: 00_Bash_basics.html, Size: 621.6 KB, Path: Lessons, Created: 2024-01-11 17:40:47, Modified: 2024-01-11 17:24:17, Writable: true. On the right, the document editor shows the content of '00_Bash_basics.html'.

Command line basics

Credit: Heavily borrowed from [Software Carpentry Foundation](#)

- [Key Points](#)
- [Graphical user interface vs. the Unix shell](#)
- [What the hell is “the shell”?](#)
- [Opening a terminal window](#)
- [Why should I use the command line?](#)
- [Let’s get familiar with the terminal!](#)
 - [Checking where you are](#)
 - [Listing contents](#)
 - [Changing directories](#)
 - [Looking at a file without opening it \(!\)](#)
 - [Seeing the contents of a file bit by bit](#)
- [Learn More](#)

Key Points

- A shell is a program whose primary purpose is to read commands and run other programs.
- This lesson uses Bash, the default shell in many implementations of Unix.
- Programs can be run in Bash by entering commands at the command-line prompt.
- The shell’s main advantages are its high action-to-keystroke ratio, its support for automating repetitive tasks, and its capacity to access networked machines.
- The shell’s main disadvantages are its primarily textual nature and how cryptic its commands and operation can be.

Graphical user interface vs. the Unix shell

Humans and computers commonly interact in many different ways, such as through a keyboard and mouse, touch screen interfaces, or using speech recognition systems. The most widely used way to interact with personal computers is called a [graphical user interface \(GUI\)](#). With a

Learning about FASTQ files with FASTQE

The screenshot shows a JupyterLab environment. On the left is a file browser pane for the directory `/ Genomics_Teaching / Lessons /`. It contains a table of files:

Name	Last Modified
00_Bash_basics.html	5 minutes ago
00_Bash_basics.pdf	5 minutes ago
00_Bash_basics.Rmd	5 minutes ago
01_fastqe.html	5 minutes ago
01_fastqe.Rmd	5 minutes ago

The main pane displays the notebook `01_fastqe.html`. The content is as follows:

Next Generation Sequencing (NGS) quality Analysis with Emoji

Developed by: Ray Enke, Rachael St. Jaques¹, Max Maza, Caylin Murray, Sabrina Robertson, Andrew Lonsdale, & Jason Williams. Modified by Bárbara Bitarello

- [Goals](#)
- [Background](#)
 - [FASTQ files](#)
- [The FASTQC software](#)
 - [What about FASTQE?](#)
- [The Data we are looking at](#)
- [Let's install fastqe](#)
- [Step 1: Install the fastqe software](#)
- [Step 2: Running fastqe](#)
- [Step 3: Using the help page](#)
- [Step 4. Scale](#)
- [Step 5. Quality](#)
- [Step 6. FASTP](#)
- [Learn More](#)

Goals

- Introduce students to writing basic command line scripts
- Analyze & assess the quality of FASTQ formatted NGS data
- Trim/filter low quality reads in FASTQ files

FastQC is a popular software used to provide an overview of basic quality metrics for NGS data. In this lesson, you will use an even more universal form of communication to analyze FASTQ files, THE EMOJI!

Background

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- Hopefully useful to instructors with different degrees of programming experience
- Upcoming: expanding/modifying the intro to command-line portion
- Possibility: publish on QUBES/CourseSource to increase visibility

PROJECT 2: THE GENOMICS EDUCATION PARTNERSHIP (GEP)

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 - participating in a genomics CURE centered around comparative gene annotation

Basic Curriculum: Understanding Eukaryotic Genomes (UEG)

All use the UCSC genome browser:

- Module 1: Intro to the UCSC Browser
- Module 2: Transcription
- Module 3: Post-transcription processing
- Module 4: Removal of introns
- Module 5: Translation
- Module 6: Alternative Splicing
- Pre and post-course, students fill out a survey that shows how much they know the content.
- Helps keep funding and assess learning gains

My experience with UEG materials

- Very customizable - materials are already great but all is easily available and free for anyone to edit
- Questions and keys are provided to instructors
- Each module fits well into a 3 hour lab
- Very positive feedback from students

Beyond UEG materials

- There are other highly curated materials + faculty-generated curriculum
- Particularly awesome for junior faculty (my opinion)
- Very helpful when designing new activities without having to start from scratch
- Lots of space to offer suggestions and contributions
- Conferences

Example 1: Mixing and adding



Genomics Education Partnership
BIOL B216 Genomics
Spring 2023
Bitarello

A hands-on Introduction to sequence homology with BLAST¹

- Adapted by Bárbara Bitarello from two GEP lessons:
 - *An introduction to NCBI* (By: Wilson Leung)
 - *Introduction to Blast using human leptin* (By: Justin DiAngelo & Alexis Nagengast)
- Optional: submit this for evaluation by GEP to become part of curated curriculum

Example 2: New but GEP-inspired

BIOL B216 Genomics

Spring 2023

Bitarello

Using UNIPROT to explore protein sequences and build a phylogenetic tree

Using UNIPROT to explore protein sequences and build a phylogenetic tree	1
Preparation	1
A very brief overview of Hemoglobin	1
Activity 1: Learning your way around UniProt	2
Activity 2: Making a phylogenetic tree of HBA1 orthologs	3

Example 2: New but GEP-inspired

- Activity in a google doc with links to:
 - Uniprot: <https://www.uniprot.org/>
 - Clustal Omega, Blast (inside Uniprot)
 - iTol Interactive Tree of Life page: <https://itol.embl.de/>

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Activity 1: Learning your way around UniProt

- Go to <https://www.uniprot.org/>
- Search for "hemoglobin subunit alpha" (in quotes)
- You will get something like 2,991 results (as of April 9th, 2023).
- Scroll down and find the human entry.
- Click on the human entry (it should be the first one): P69905
- This takes you to a page with a lot of information about this specific protein.

Function	Protein ⁱ	Hemoglobin subunit alpha	Amino acids	142
Names & Taxonomy	Gene ⁱ	HBA1; HBA2	Protein existence ⁱ	Evidence at protein level
Subcellular Location	Status ⁱ	UniProtKB reviewed (Swiss-Prot)	Annotation score ⁱ	5/5
Disease & Variants	Organism ⁱ	Homo sapiens (Human)		
PTM/Processing	Entry	Feature viewer	Publications	External links
Expression	BLAST	Download	Add	Community curation (3)
Interaction			Add a publication	Entry feedback
Structure				
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Sequence				
Similar Proteins				

Functionⁱ

Involved in oxygen transport from the lung to the various peripheral tissues.

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Hemopressin acts as an antagonist peptide of the cannabinoid receptor CNR1 (PubMed:18077343).

Hemopressin-binding efficiently blocks cannabinoid receptor CNR1 and subsequent signaling (PubMed:18077343). 1 Publication

- Take some time to explore the data in this page.

Questions:

- 1) How many aminoacids long is this protein?
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Bonus: Other projects

- Digital Scholarship Grant from BMC: Developing an R package with materials for the *B215: Biostatistics with R* course.
- Many great resources out there, but not very specific to biology
- \$5,000\$ to pay two UG students to help me with this (2023-2024)
- Currently not a package but materials were used this fall
- Considering making this a mybinder.org repo instead

Acknowledgments

For the command-line activity portion, I took heavy inspiration from:

- The Software Carpentry. <https://swcarpentry.github.io/shell-novice/01-intro.html> (Accessed March 22, 2023)
- <https://thegep.org/>



BRYN
MAWR
COLLEGE

Thank you!

Questions?

Email: bbitarello@brynmawr.edu

Website: <https://bitarellolab.digital.brynmawr.edu/>

GitHub: <https://github.com/bitarellolab>

Twitter (X): [@dudutchy](https://twitter.com/dudutchy)

THANK YOU!

QUESTIONS?

Email: bbitarello@brynmawr.edu (happy to share materials!)

Website: <https://bitarellolab.digital.brynmawr.edu/>

GitHub: <https://github.com/bitarellolab>

Twitter (X): @dudutchy