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

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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

1. Why browser-based?
2. Two projects/experiences from B216 (Genomics) that only require a browser
   - 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!

Why browser-based?

Challenges for teaching genomics/bioinformatics:

1. getting all tools installed in a variety of OS and versions: often **frustrating** and **time-consuming**
2. campus computers: often **lack permissions** to get all the **required updates** and **installations** in a timely manner
3. some students use machines that **lack space or capability** for local installations (e.g. Chromebook)
4. technical challenges intimidate students even more; the browser **keeps it familiar/simple**

Browser-only activities bypass all of these hurdles!

Examples

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

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

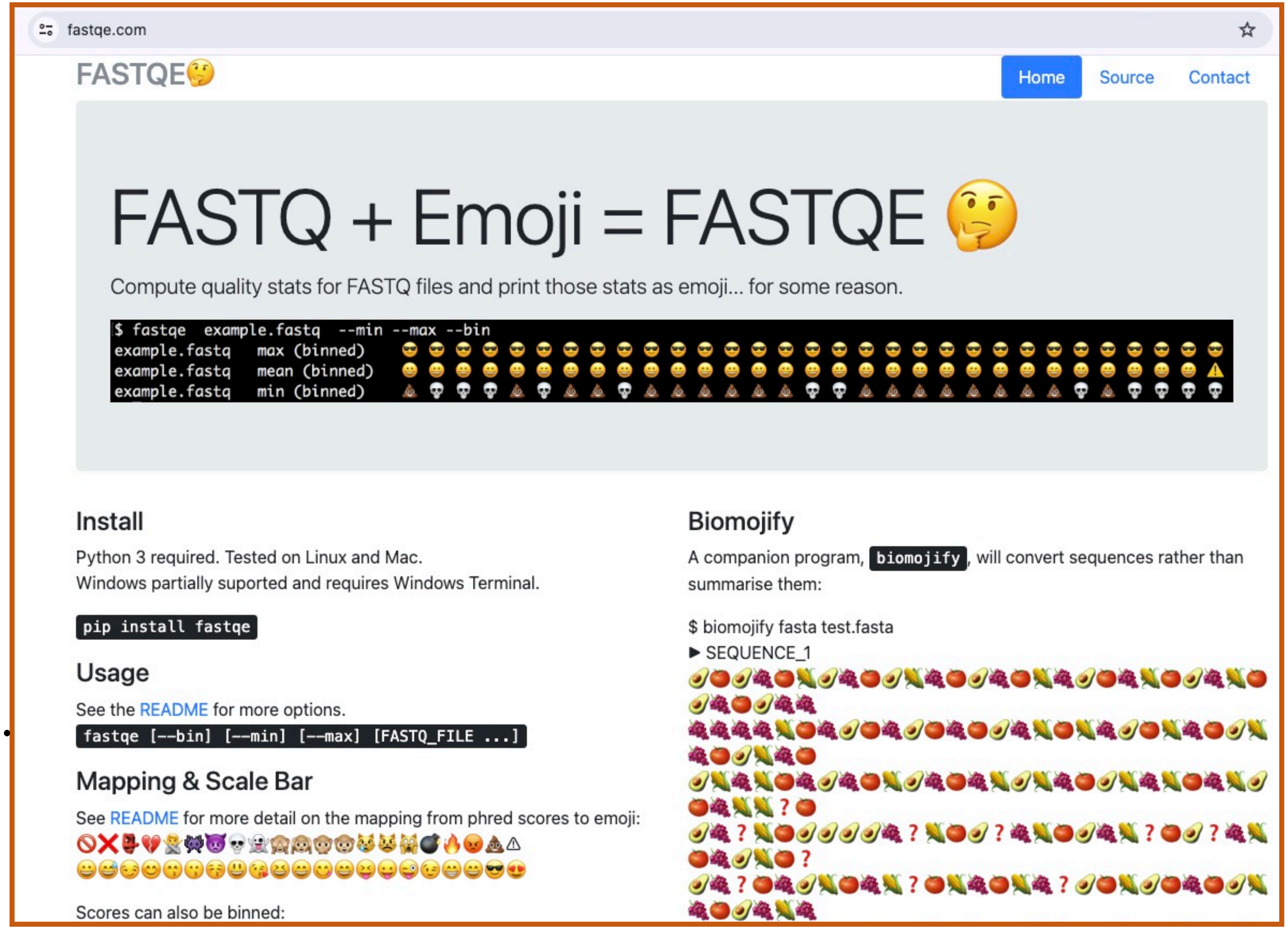
Project 1: A soft introduction to the command-line and FASTQ files

Motivation

- Excellent materials from St. Jaquest et al. (2021), published in *CourseSource*
- Introduces the command-line and FASTQ files by using the FASTQE software
- I reached out to senior author Ray Enke about my adapted materials and here we are!

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](doi.org/10.24918/cs.2021.17)



FASTQ + Emoji = FASTQE 🤔

Compute quality stats for FASTQ files and print those stats as emoji... for some reason.

```
$ fastqe  example.fastq  --min --max --bin
example.fastq   max (binned)   😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎 😎
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example.fastq   min (binned)   💩 💀 💀 💀 💩 💀 💩 💩 💀 💩 💩 💩 💩 💩 💩 💀 💀 💩 💩 💩 💩 💩 💩 💩 💩 💀 💩 💀 💀 💀 💀
```

Install

Python 3 required. Tested on Linux and Mac.
Windows partially suported and requires Windows Terminal.

```
pip install fastqe
```

Usage

See the README for more options.

```
fastqe [--bin] [--min] [--max] [FASTQ_FILE ...]
```

Mapping & Scale Bar

See README for more detail on the mapping from phred scores to emoji:
🚫❌👺💔🙅👾👿💀👻🙈🙉🙊🐵😿😾🙀💣🔥😡💩⚠
😀😅😏😊😙😗😚😃😘😆😄😋😄😝😛😜😉😁😄😎😍

Scores can also be binned:

Biomojify

A companion program, `biomojify`, will convert sequences rather than summarise them:

```
$ biomojify fasta test.fasta
▶ SEQUENCE_1
🥑🍅🥑🍇🍅🌽🥑🍇🍅🥑🌽🍇🍅🥑🍇🍅🌽🍇🥑🍅🍇🌽🍅🥑🍇🌽🍅
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🍇🍅🥑🍇🌽🍇
```

Challenges in implementing the lesson

- FASTQE is a python package that needs to be installed (as well as its dependencies) — and python installations are ~~always~~ often a nightmare!
- Proposed implementation in publication: either a) local installation or b) CyVerse
- 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

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

Learning goals

1) Have a soft introduction to the command line

- ls
- mkdir
- cd
- pwd
- more
- less
- wc
- pip
- conda

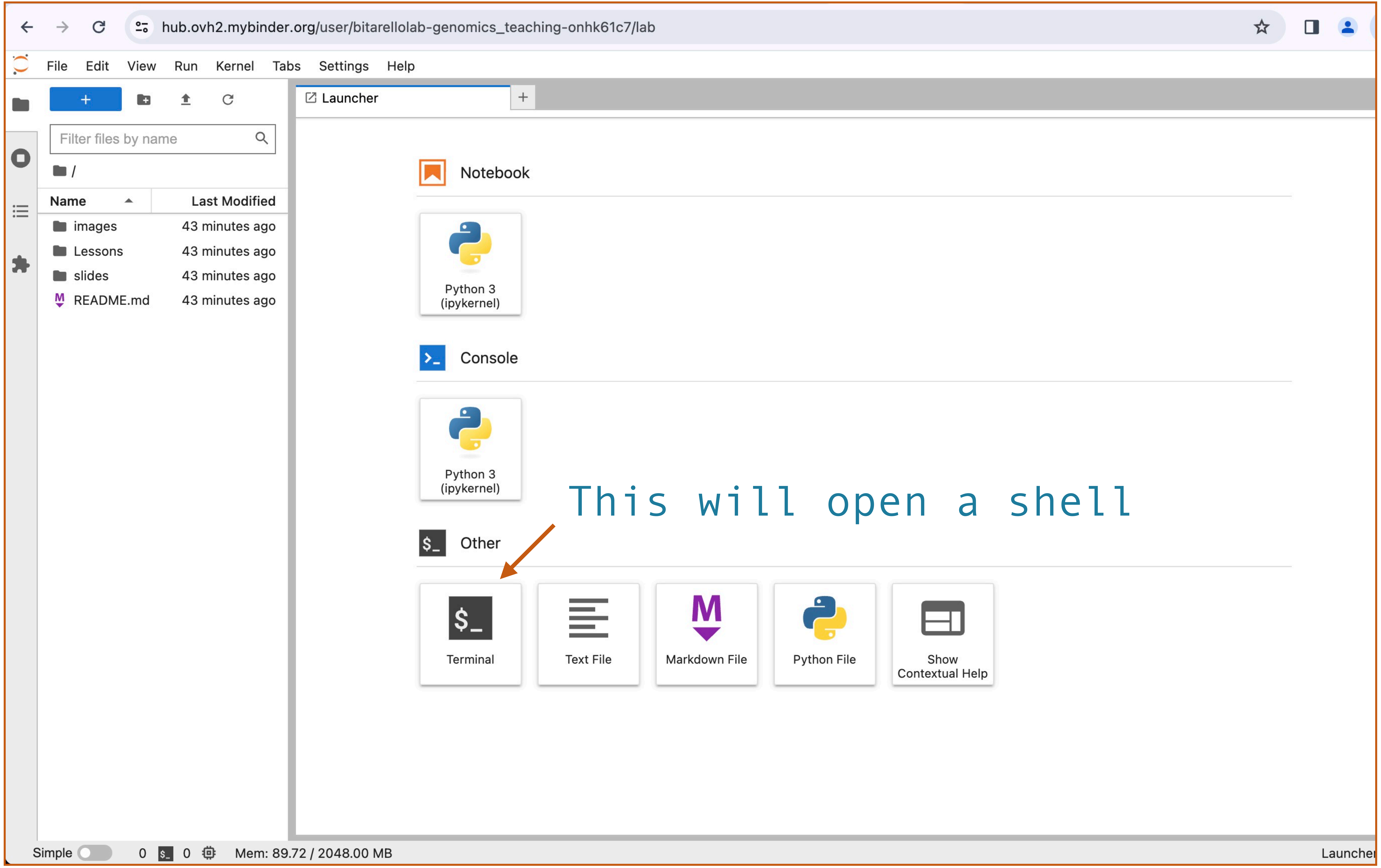
2) Have a soft introduction to FASTA and FASTQ files

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

Structure

- Students follow the two tutorials through the mybinder.org link on their browser in order:
  - 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

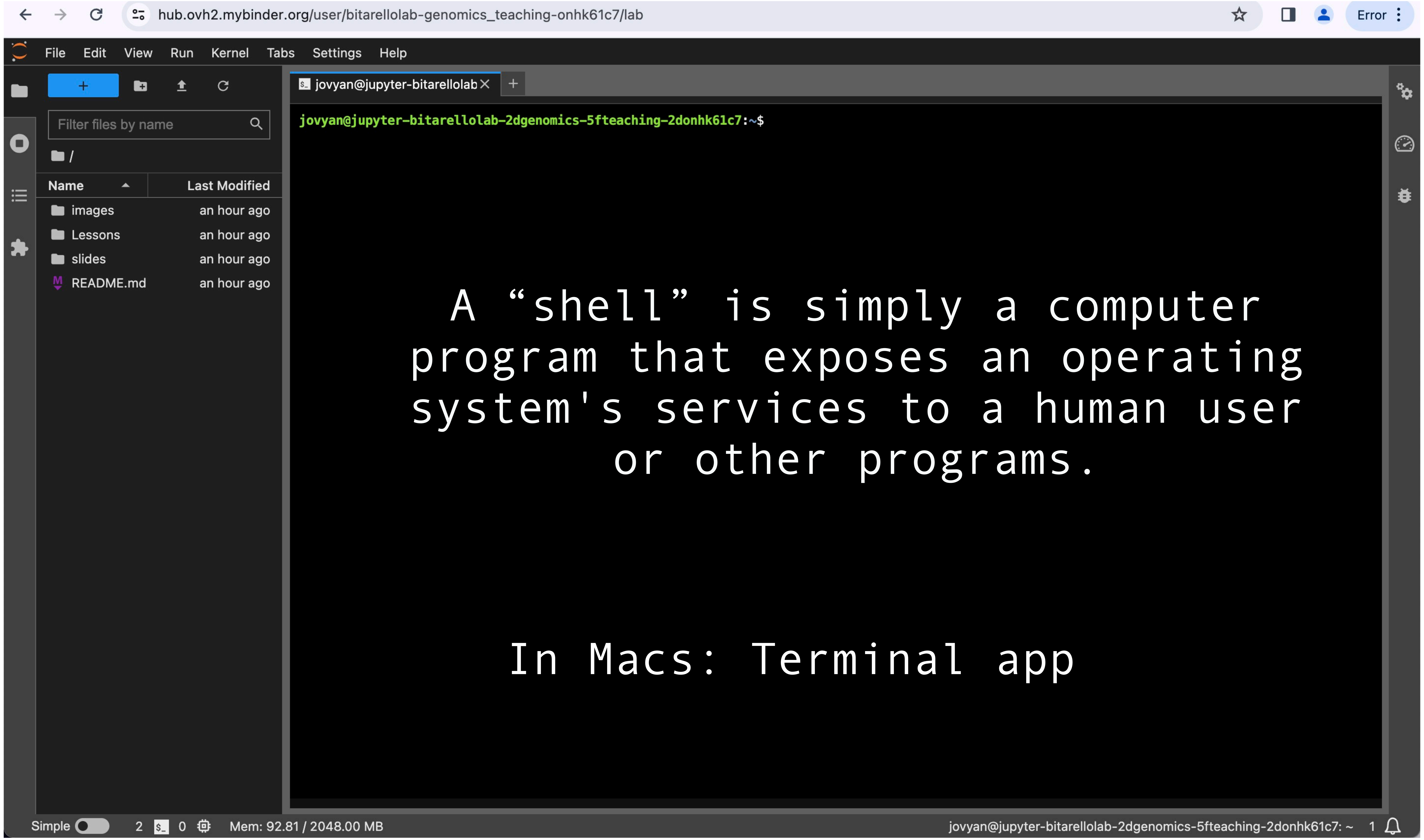
Landing page



Name	Last Modified
images	43 minutes ago
Lessons	43 minutes ago
slides	43 minutes ago
README.md	43 minutes ago

This will open a shell

A shell is now open


hub.ovh2.mybinder.org/user/bitarellolab-genomics_teaching-onhk61c7/lab
Error
File
Edit
View
Run
Kernel
Tabs
Settings
Help
+
Filter files by name
/
jovyan@jupyter-bitarellolab
Simple

Name	Last Modified
images	an hour ago
Lessons	an hour ago
slides	an hour ago
README.md	an hour ago

```
jovyan@jupyter-bitarellolab-2dgenomics-5fteaching-2donhk61c7:~$
```

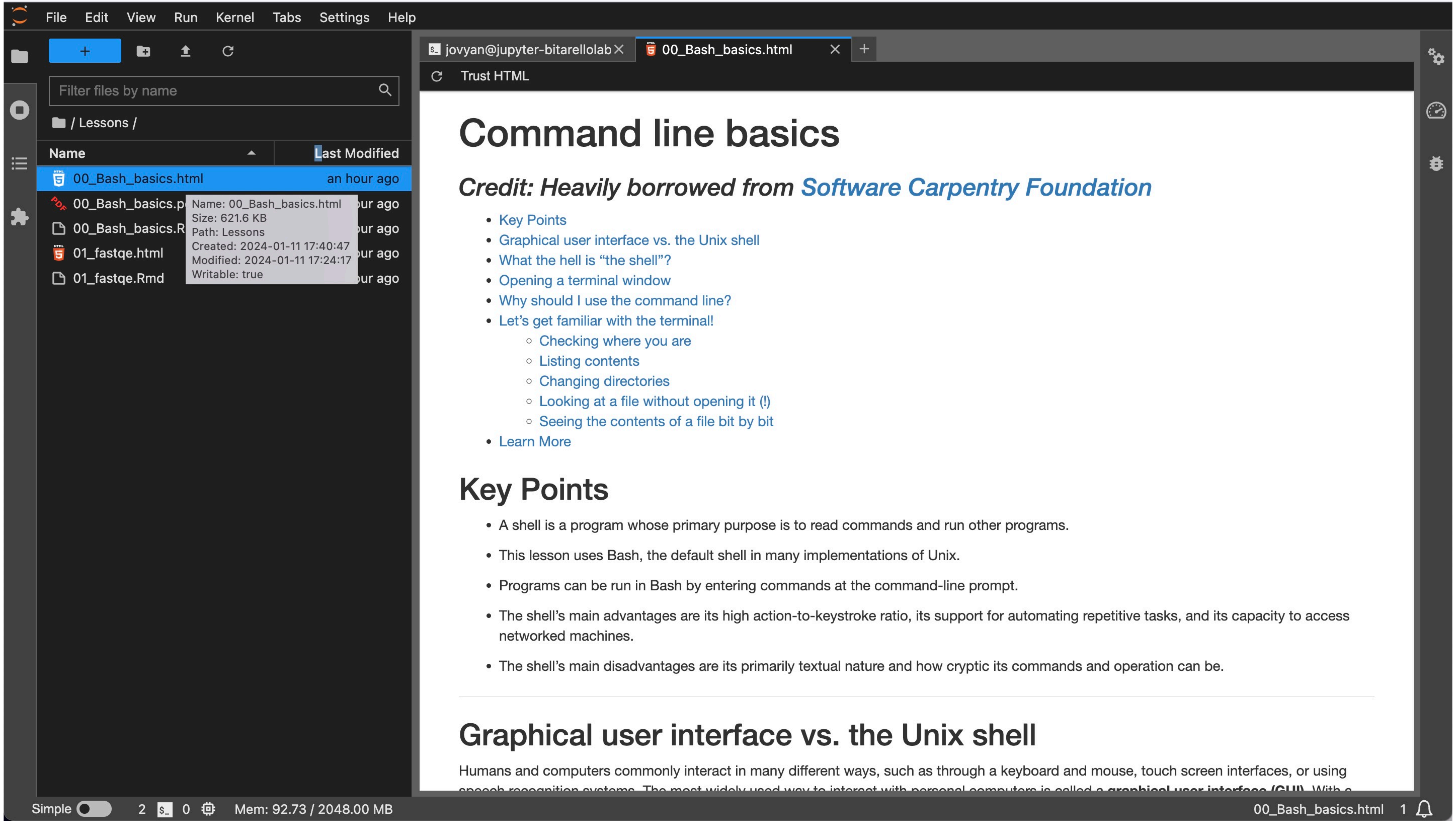
2 0 Mem: 92.81 / 2048.00 MB

jovyan@jupyter-bitarellolab-2dgenomics-5fteaching-2donhk61c7: ~ 1

A “shell” is simply a computer program that exposes an operating system's services to a human user or other programs.

In Macs: Terminal app

Soft introduction to the command line



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

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

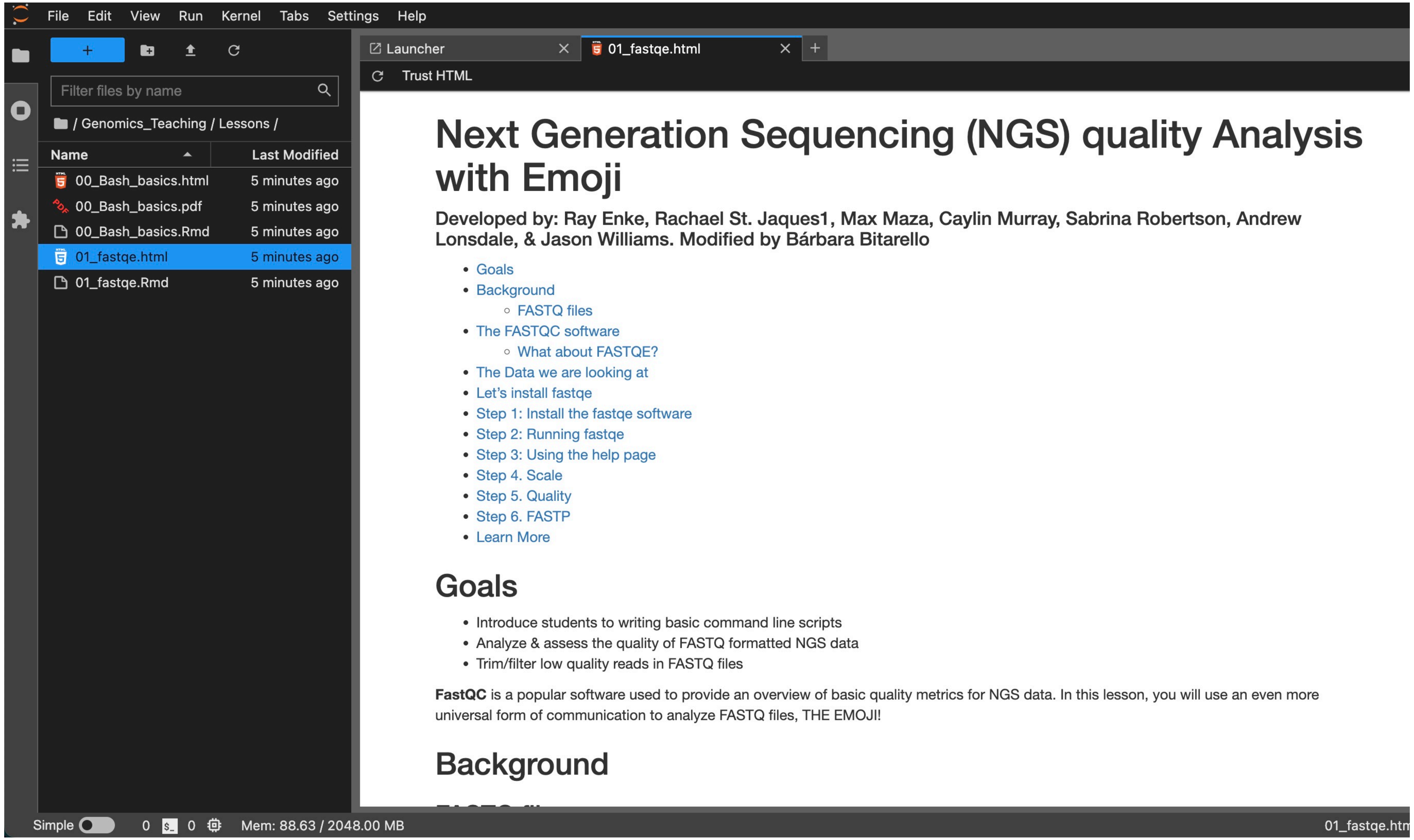
Key Points

- 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



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

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

- 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

Spread the love

- In its current implementation I have had zero issues with folks accessing it
- Instructors with coding experience/teaching goals: can use this as a template for other activities
- Instructors without coding experience/teaching goals: can access and follow the activity seamlessly through my github

https://github.com/bitarellolab/Genomics_Teaching

Where we're at

- Possibility: publish on QUBES/CourseSource to increase visibility
- Currently expanding/modifying the intro to command-line portion

Tl;dr

- Adapts St. Jacques et al. (2021) materials so that everything can be installed and run from a browser
- Preserves the learning process of installing the packages while providing a uniform environment for all students
- Hopefully useful to instructors with different degrees of programming experience
- Upcoming: updates on both portions of the tutorial

Project 2: The Genomics Education partnership (GEP)

The Genomics Education Partnership (GEP)

- Active member since July 2021.
- Integrates **active learning** into the undergraduate curriculum through CURES centered in **bioinformatics** and **genomics.**
- Use of GEP materials depends on course structure and student background experience
  - incorporating short lessons into an existing course (e.g., using a genome browser to investigate eukaryotic gene structure)
  - 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

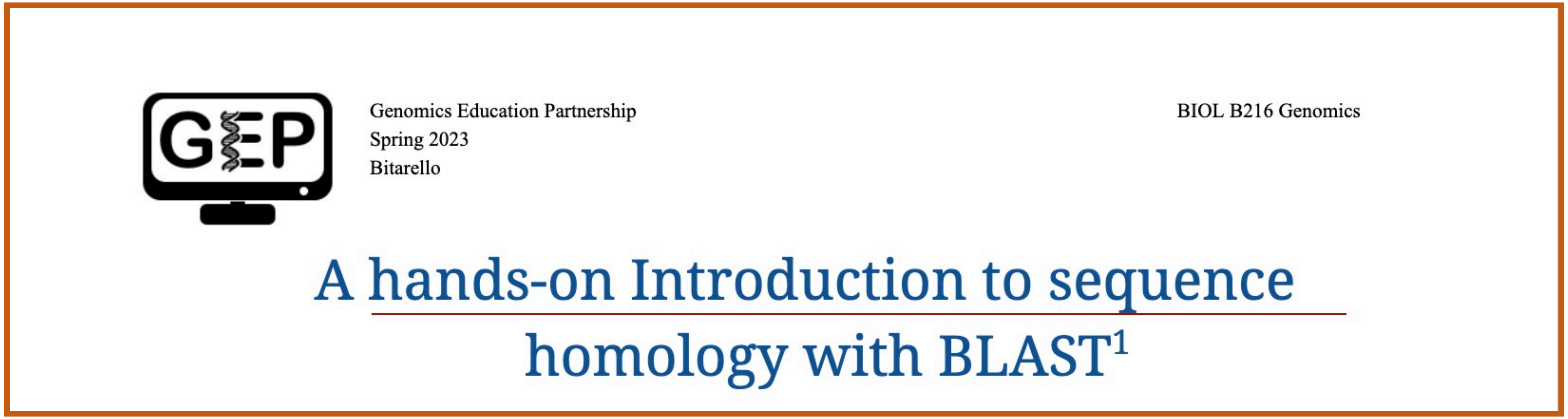
My experience with UEG materials

- 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
- Very helpful when designing new activities without having to start from scratch
- Particularly awesome for junior faculty (my opinion)
- Lots of space to offer suggestions and contributions
- Conferences

Example 1: Mixing and adding



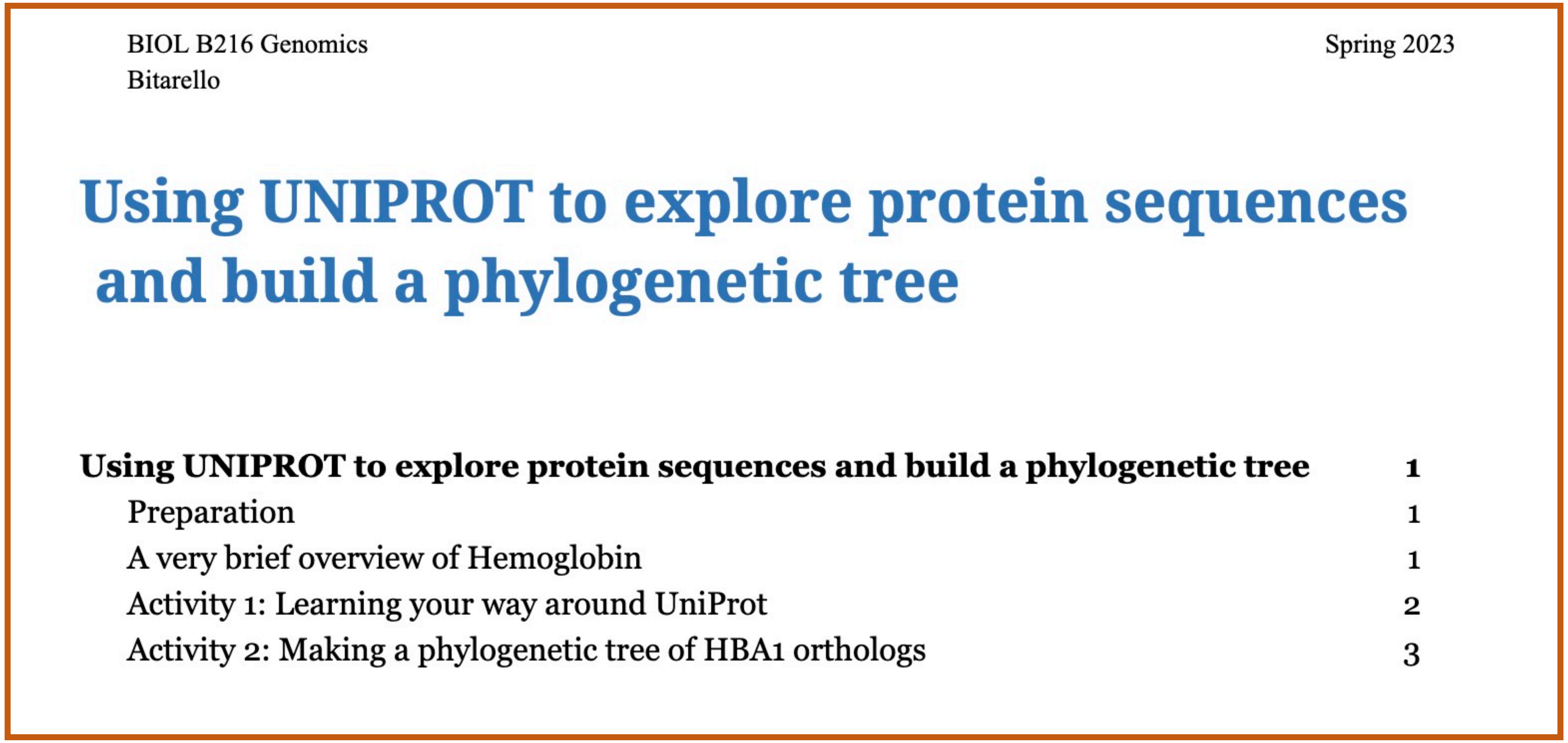
Genomics Education Partnership
Spring 2023
Bitarello

BIOL B216 Genomics

A hands-on Introduction to sequence homology with BLAST[1]

- 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
Bitarello

Spring 2023

Using UNIPROT to explore protein sequences and build a phylogenetic tree

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/

- My own, but using the GEP "format" as inspiration saved me a lot of time
- Optional: submit this for evaluation by GEP to become part of curated curriculum

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/index.html (Accessed March 22, 2023)

- https://thegep.org/

GEP

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Thank you!

Questions?

Email: bbitarello@brynmawr.edu

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

GitHub: https://github.com/bitarellolab

Twitter (X): @dudutchy

Thank you!

Questions?

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

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