Welcome to this Training Session with Theiagen Genomics

We will soon be getting started

Software Development Practices for Public Health Bioinformatics

Week 01: *Design Documents & Development Environments*

A Northeastern Bioinformatics Regional Resource Offering Provided by the Massachusetts Department of Public Health in Collaboration with Theiagen Genomics

Course Introduction

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Training Workshop Resources

Training Information, Communication, and Support

- **GitHub Repo** created to host training resources and information:
  - https://github.com/theiagen/Northeast-SDP4PHB-2025
- **Support contact**:
  - support@theiagen.com

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Training Objectives

Software Development Practices for Public Health Bioinformatics

- Knowledge of **best software development practices applied to public health bioinformatics** such as **design documents** and the use of **integrated development environments**
- Proficiency in using **version control systems** for collaborative development
- Ability to deploy **continuous integration** and acceptance testing
- Understanding **workflow managers** and how to construct an analysis pipeline

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Training Workshop Instructors

Andrew Lang, PhD

- Development Team Lead
- PhD Genetics

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Public Health Bioinformatics

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Public Health Bioinformatics

Young Discipline

- US public health systems adopted NGS and bioinformatics technologies **~10 years ago**
  - **Next-to-zero bioinformatics capabilities** across the US public health system in 2015
- First practitioners had **limited software experience**
  - Most were *wet-lab-turned-dry-lab* scientists

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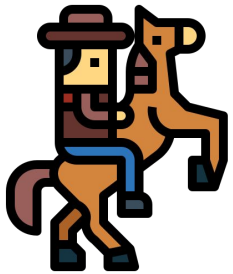
Public Health Bioinformatics

The Wild West

- Public health bioinformatics was often **chaotic and unstructured**
  - Created **major challenges** in accessible, reproducible, and interoperability software solutions



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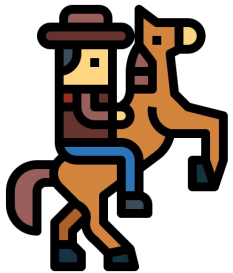
How to get a bioinformatics headache

1. See tweet about new published tool
2. Read abstract - sounds awesome!
3. Fail to find link to source code - eventually Google it
4. Attempt to compile and install it
5. Google for 30 min for fixes
6. Finally get it built
7. Run it on tiny data set
8. Get a vague error
9. Delete and never revisit it again



Slide from: T. Seemann, ASMNGS18
https://www.slideshare.net/torstenseemann/how-to-write-bioinformatics-software-no-one-will-use/11

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Public Health Bioinformatics

Huge Leaps Forward

- Major progress was made in our field with the adoption of specific technologies:
  - **Software containerization**
  - **Workflow managers**
  - **Cloud infrastructure**
  - **Graphic User Interfaces (GUIs)**

*Adoption of these technologies led to an emergence of **more mature software development practices***

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Public Health Bioinformatics

Maturing Software Development Practices

- Accessibility, reproducibility, and interoperability became a **major goal across the field**; forcing function for:
  - Adopting of **standardized development practices**
  - **Enhanced collaboration** across interdisciplinary teams
  - Development of software meant for **wide distribution** across the public health community

*These practices have become the **new status quo** in public health bioinformatics*

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Software Development Practices

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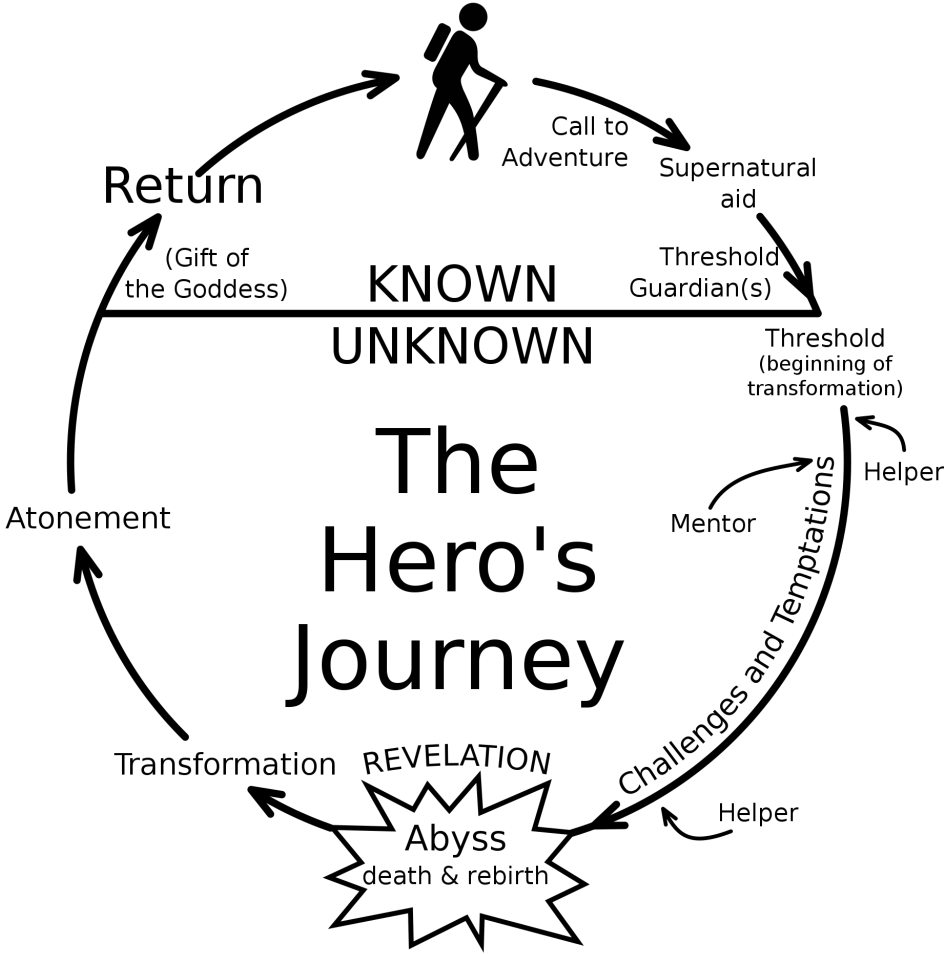
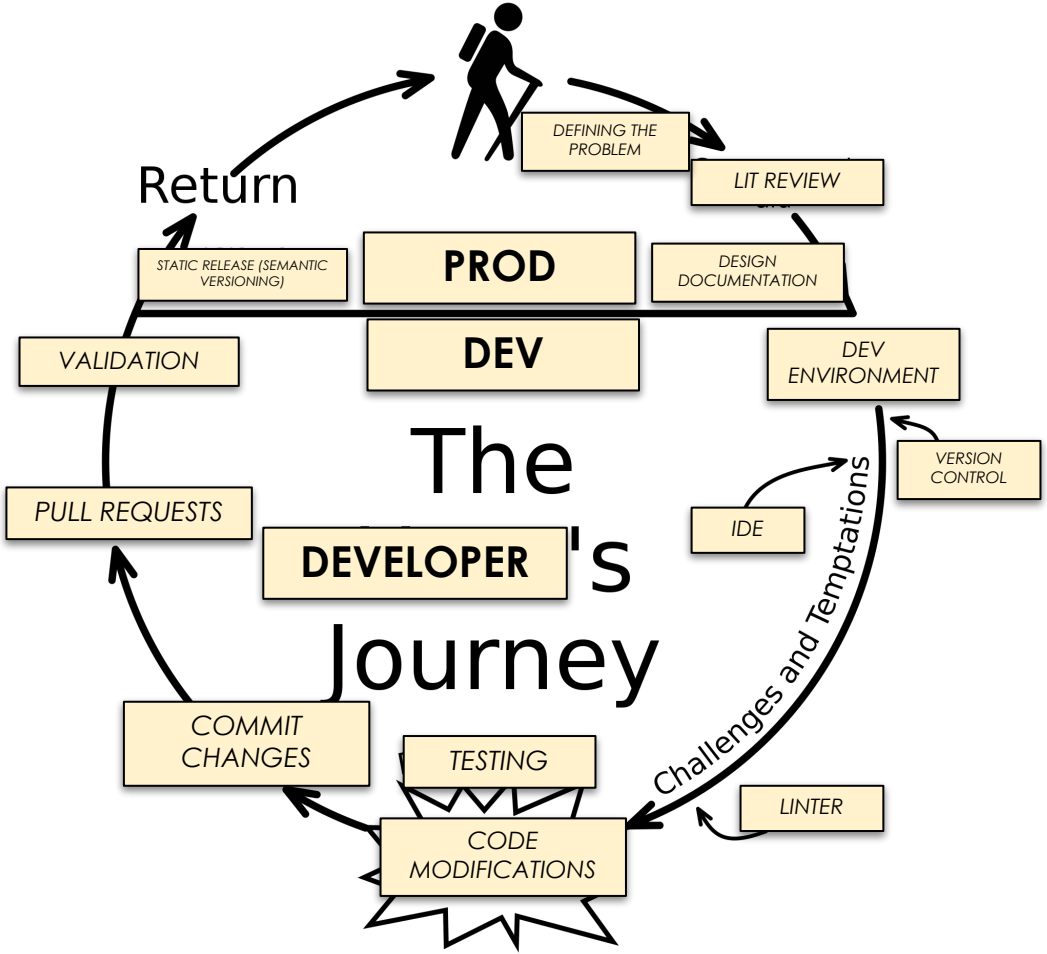


The Hero's Journey

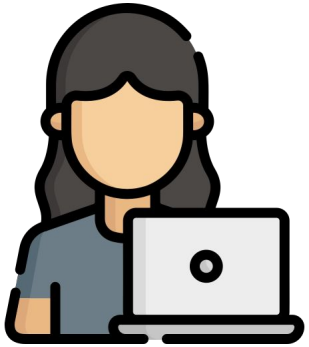
Framework where a protagonist **enters into the unknown**, faces challenges, gains new wisdom, and **returns transformed**.

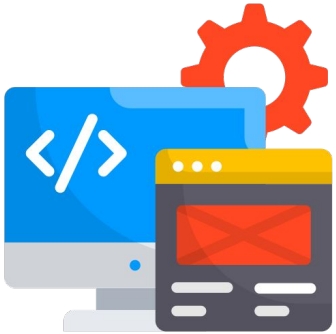
Byrne, C. (2017). The Hero's Journey. Wikipedia https://en.wikipedia.org/wiki/File:Heroesjourney.svg



The Developer's Journey

Framework where a protagonist **enters into their dev environment**, faces challenges, gains new wisdom, and **brings changes into production**.





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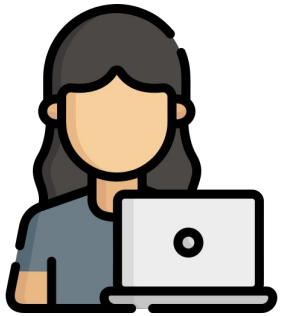
Software Development Practices

Developer's Journey

Week 1 Focus

1. **Design Document**
   a. Clearly defining the problem and the proposed solution
2. **Development Environment**
   a. Separate from production
   b. Text editors and IDE's
3. **Making Source Code Modifications**
   a. Small interactive changes (version control)
4. **Peer Review**
   a. Collaborative development teams
5. **Bringing Changes into Production**
   a. Final testing
   b. Static version releases



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1. Design Document

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Design Document

Defining the Problem and Proposed Solution

- Design docs are written plans that **outline the problem, objectives, proposed solutions, and implementation strategy** for a software project
  - Static if small scope
  - "Living" if larger scope
- Ensure everyone has a clear understanding of the project scope and requirements (**as well as what is out of scope**)

Living document *that serves as a* ***reference*** *throughout the development process*

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Design Document

Major Components

- Problem Statement: Clearly define the problem that needs to be solved
- Objectives*: Outline the goals and what success looks like.
- Proposed Solution: Describe the approach to solving the problem.
- Implementation Plan: Detail the steps, timeline, resources required, and integration with existing systems

*Should be informed by **literature review** and **community feedback***

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Design Document

Literature Review and Community Feedback

- Conduct a review of existing publications and solutions related to the problem
  - Look specifically for **public health applications**
- Reach out to the **wider technical community**
  - MicroBinfie, StaPH-B, etc.

*Helps to **identify open-source tools** that you will **incorporate in your pipeline***

*May identify a dev solution that **already exists** to address your problem*

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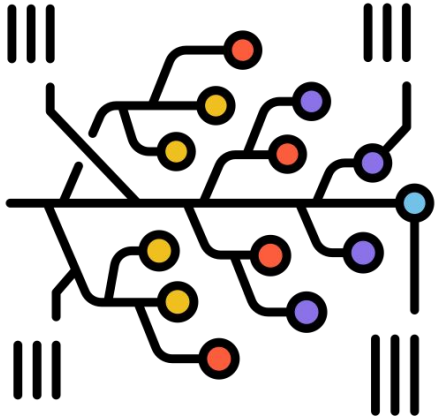
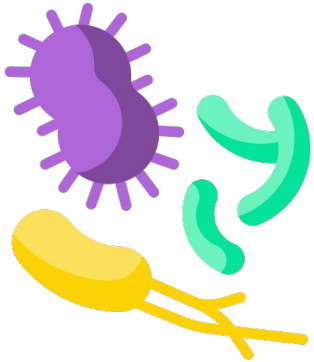
Selecting the Appropriate Tool for a Pipeline

Define the problem you are hoping to solve; highlight how your software will **help to inform public health decision-making**

- Be as **specific** as you can!
- Define how you will **measure the fit** of a software solution





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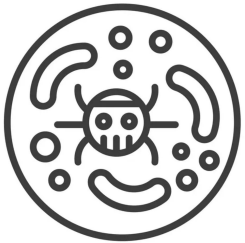
Selecting the Appropriate Tool for a Pipeline

Read the literature to **define your options**

- Helpful to identify evidence of **public health application**

Seek **community feedback**

- StaPH-B, microBinfie, OAMD BRR/WFD
- **You are not the only one** facing this challenge



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Design Document

Organization Specific Considerations

- Tailor your design document to your organization's processes
  - May be **additional details** that are helpful for your colleagues, e.g. state IT personnel may need to be involved to support some dev initiatives
- Quality Management Systems
  - **Defined testing and validation datasets** may be defined if you're updating an existing, validated pipeline

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Design Document

Summary

- The design document is a vital tool that **defines the problem and the proposed solution**, informed by literature review and community feedback.
  - It ensures **clear communication** and alignment among stakeholders
- In writing a design document, you should be assessing other open-source solutions
  - Seek tools that **align** with your project goals
  - **Read the literature** and **engage the community**

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2. Development Environment

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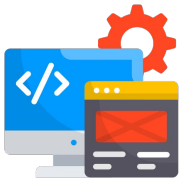
Development Environment

Separating Prod from Dev Resources

- Prevents **untested code** from affecting live production systems.
- Allows for **safer experimentation and testing.**
- Risk mitigation:
  - Isolates development activities, ensuring that any bugs or issues are **contained within the development environment**.



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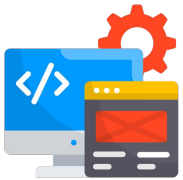
Development Environment

Maintaining Prod and Dev Environments

- Version Control Systems like Git help manage changes to source code over time
  - Can create **development copies** of the source code without modifying production source code
    - Developing in **forks or branches** of the production codebase



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A Note on Version Control Systems

Version Control Systems (VCS)

- Essential development tools that help manage changes to source code over time
  - Track (and save) modifications to the code

Git and GitHub

- Git is a VCS software for managing code in repositories
- GitHub is a platform to host Git repositories

*Git repositories can be hosted on other platforms such as **BitBucket and GitLab***

*More on **Git next week**!*

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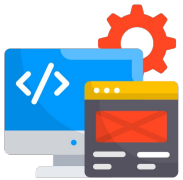
Development Environment

Maintaining Prod and Dev Environments

- Mimicking your production environment
  - VMs can be based off of production **images**
  - Readily deployable in cloud environments, can also be setup on local systems with software like **VirtualBox**

*In this context, "**images**" refer to a snapshot or template of a system's state at a **particular point in time***



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Development Environment

Example Configuration:

- **Dev**: Dedicated VM without access to production resources, local (mutable) checkout of development branch of code
- **Staging:** Dedicated VM without access to production resources, but a mirror of all production resources & service account permissions & local checkout of code (immutable)
- **Prod**: Dedicated VM with access to production resources, local checkout of stable version of code (immutable)

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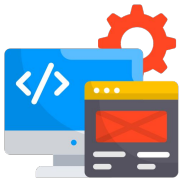
Development Environment

IDE v. Text Editors

- **Text Editor** - software application used for **editing plain text**
  - Example software: VIM, EMACS
- **IDEs** (Interactive Development Environments): software suite that offer a **GUI to various tools** that facilitate software development such as:
  - Code editing, debugging, and version control within a **single application**
  - Example software: Visual Studio and Atom



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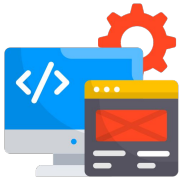
Development Environment

Advantages of IDE

- **Code navigation:** Enable developers to move through and understand structure of a large codebase efficiently
  - Particularly important with complex file systems
- **Error reduction:** Provide real-time syntax checking and debugging tools to catch errors early in development cycle
- **Resolving merge conflicts:** Provide visual comparisons and interactive editing to assist in resolving merge conflicts



Note: *These features mean that IDEs tend to be* ***more resource-intensive*** *relative lightweight text editors like VIM*

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IDE Example - Error reduction

```
output {
  # Version Capture
  String freyja_fastq_wf_version = version_capture.phb_version
  String freyja_fastq_wf_analysis_date = version_capture.date
  # Read QC - fastq_scan outputs
  Int fastq_scan_num_reads_raw1 = read_QC_trim.fastq_scan_raw1
  Int? fastq_scan_num_reads_raw2 = read_QC_trim.fastq_scan_raw2
  String? fastq_scan_num_reads_raw_pairs = read_QC_trim.fastq_scan_raw_pairs
  String? fastq_scan_version = read_QC_trim.fastq_scan_version
  Int? fastq_scan_num_reads_clean1 = read_QC_trim.fastq_scan_clean1
  Int? fastq_scan_num_reads_clean2 = read_QC_trim.fastq_scan_clean2
  String? fastq_scan_num_reads_clean_pairs = read_QC_trim.fastq_scan_clean_pairs
```

*Catching errors with VIM – **manual review**; post-hoc testing*

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IDE Example - Error reduction

```
50  output {
51      # Version Capture
52      String freyja_fastq_wf_version = version_capture.phb_version
53      String freyja_fastq_wf_analysis_date = version_capture.date
54      # Read QC - fastq_scan outputs
55      Int fastq_scan_num_reads_raw1 = read_QC_trim.fastq_scan_raw1
56      Int? fastq_scan_num_reads_raw2 = read_QC_trim.fastq_scan_raw2
57      String? fastq_scan_num_reads_raw_pairs = read_QC_trim.fastq_scan_raw_pairs
58      String? fastq_scan_version = read_QC_trim.fastq_scan_version
59      Int? fastq_scan_num_reads_clean1 = read_QC_trim.fastq_scan_clean1
60      Int? fastq_scan_num_reads_clean2 = read_QC_trim.fastq_scan_clean2
61      String? fastq_scan_num_reads_clean_pairs = read_QC_trim.fastq_scan_clean_pairs
```

*Catching errors with VSCode – **Error highlighting**; suggested fixes*

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IDE Example - Error reduction

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55      Int fastq_scan_num_reads_raw1 = read_QC_trim.fastq_scan_raw1
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57      String? fastq_scan_num_reads_raw_pairs = read_QC_trim.fastq_scan_raw_pairs
58      String? fastq_scan_version = read_QC_trim.fastq_scan_version
59      Int? fastq_scan_num_reads_clean1 = read_QC_trim.fastq_scan_clean1
60      Int? fastq_scan_num_reads_clean2 = read_QC_trim.fastq_scan_clean2
61      String? fastq_scan_num_reads_clean_pairs = read_QC_trim.fastq_scan_clean_pairs
```

```
Expected Int instead of Int? -- to coerce T? X into T, try
select_first([X,defaultValue]) or select_first([X]) (which might fail at runtime); to
coerce Array[T?] X into Array[T], try select_all(X)

View Problem (Alt+F8)    Quick Fix... (Ctrl+.)
```

Catching errors with VSCode – Error highlighting; ***suggested fixes***

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Development Environment

Summary

- Separating development and production environments is crucial to **mitigate risks**
  - Strategies such as using **separate compute environments**, **version control systems**, and **mimicking prod environment configurations** help achieve this separation effectively.
- IDEs **can enhance development productivity** with features like code navigation, active error catching, and version control integration

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Hands-On Exercise

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Software Development Practices

Developer's Journey

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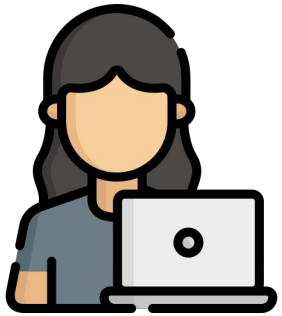

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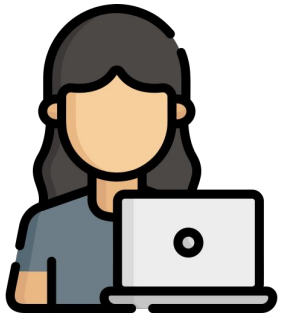
Exercise 01: Design Doc, Dev Environment, and Scripting with VSCode

Exercise Goal

1. Review a design document for a development initiative
2. Access a development environment via GitPod
3. Use VSCode IDE to test code and script solution



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