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Polygenic Risk Score Analyses Workshop 2025

Day 1a: GWAS & relevant Statistics

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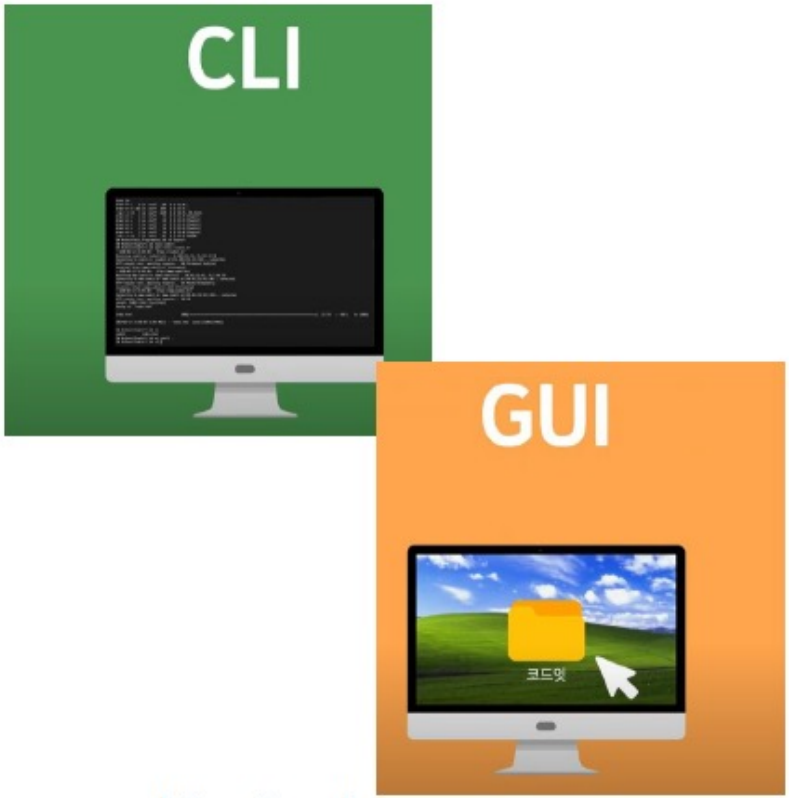
What is a Shell?

- A computer program that provides a command-line interface, accessible from the console or a terminal.
- Alternative to using a graphical user interface (GUI).
- Allows users to control the computer using commands entered with a keyboard.
- The user enters commands as text input, which is then interpreted and executed by the shell.
- Linux and macOS : robust command-line interfaces

Command Line Interface (CLI)



https://medium.com

How to access the shell ?

- On a Mac or Linux machine, you can access a shell through a program called “Terminal”, which is

already available on your computer.

- The Terminal is a window into which we will type commands.
- For scripting and automation, Linux commonly uses:
    - Bash (Bourne Again Shell) – Default scripting language for many Linux distributions.
    - Python – Widely used for automation and system administration.
    - Perl, Ruby, and Shell scripting languages – Used in various system tasks.

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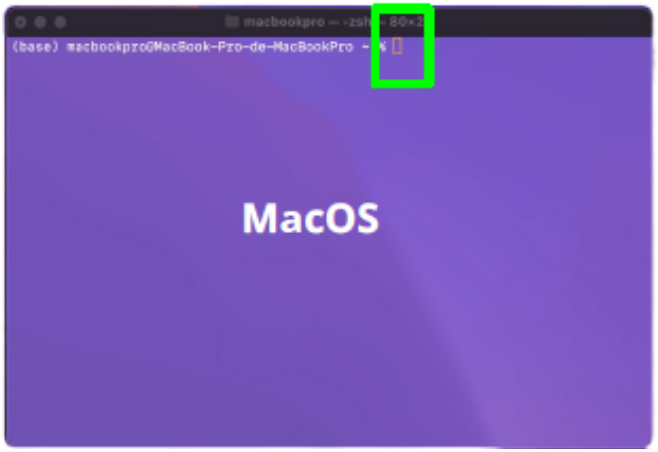
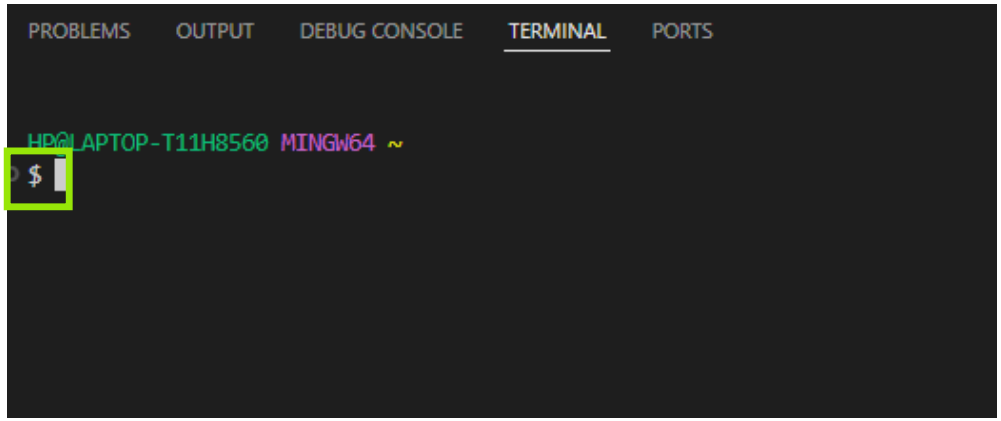
How to access the shell ?

- The dollar sign is a prompt,
- Prompt shows that the shell is waiting for input
- Your shell may use a different character as a prompt and may add information before the prompt.

When typing commands, either from these lessons or from other sources, do not type the prompt, only the commands that follow it !!!





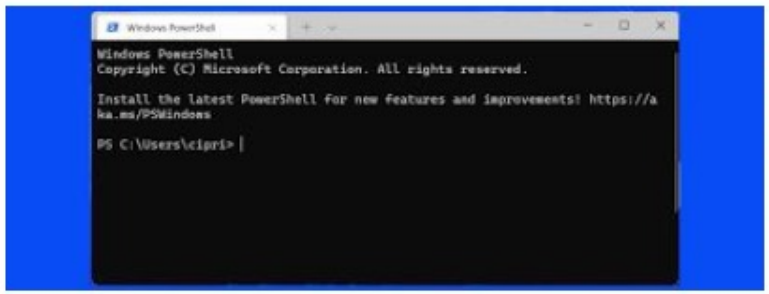
Shell for Windows users

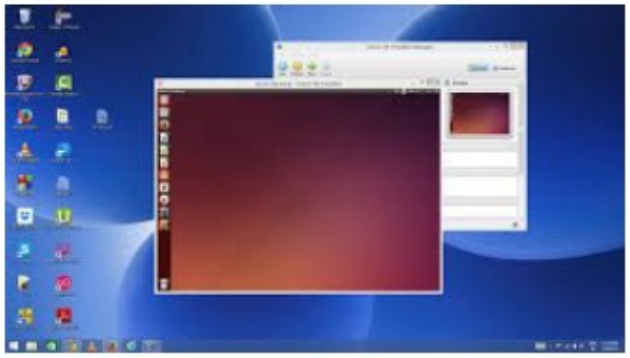
- If you're using Windows, you'll need to :

- download a separate program to access the shell.

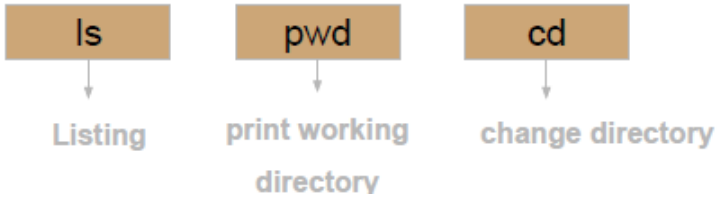
VirtualBox is a free and open-source virtualization software that allows you to run multiple operating systems (like Linux) inside Windows as a virtual machine (VM).

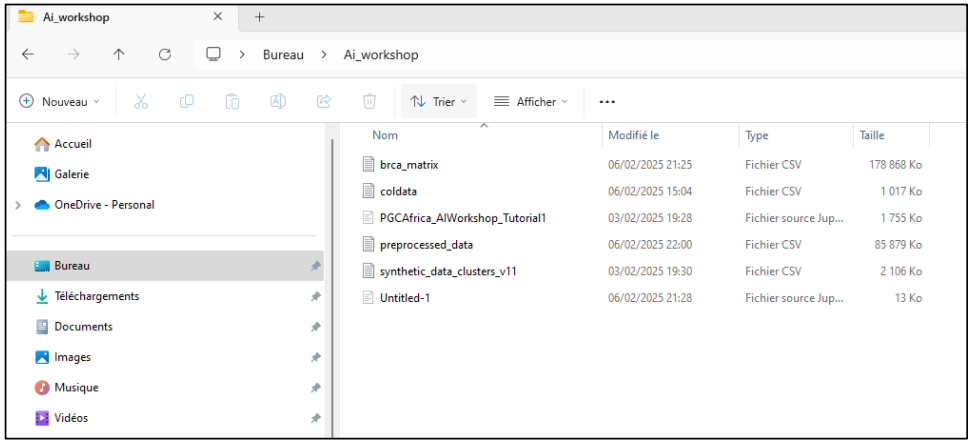
- Install Ubuntu





Navigating your file system





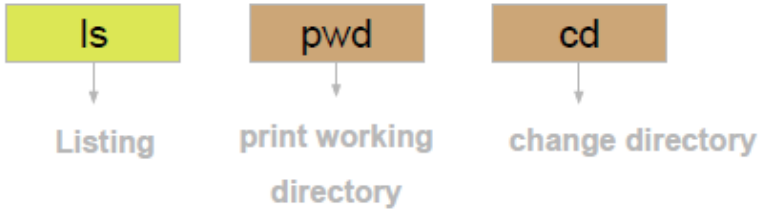
File system : The part of the operating system that manages files and directories.

It organizes our data into :

- Files ⇒ hold information
- Directories (also called “folders”) ==> hold files or other directories

```
PS C:\Users\HP\Desktop> tree
Structure du dossier pour le volume Windows
Le numéro de série du volume est 3EE3-3E01
C:.
├───Adobe professionnel
│   ├───Adobe Acrobat X
│   └───Crack-MPT[h33t][eSpNs]
├───Ai_workshop
└───Office 2016
    └───Office_2016_French_Language_Pack_-x64.MyEgy.Tv
        ├───access.fr-fr
        ├───catalog
        ├───dcf.fr-fr
        ├───excel.fr-fr
        ├───groove.fr-fr
        ├───hotfixes
        ├───infopath.fr-fr
        ├───lync.fr-fr
        ├───office.fr-fr
        ├───office32.fr-fr
        ├───omui.fr-fr
        ├───onenote.fr-fr
        ├───osm.fr-fr
        ├───osmux.fr-fr
        ├───outlook.fr-fr
        ├───pmui.fr-fr
        ├───powerpoint.fr-fr
        ├───project.fr-fr
        ├───proofing.fr-fr
```

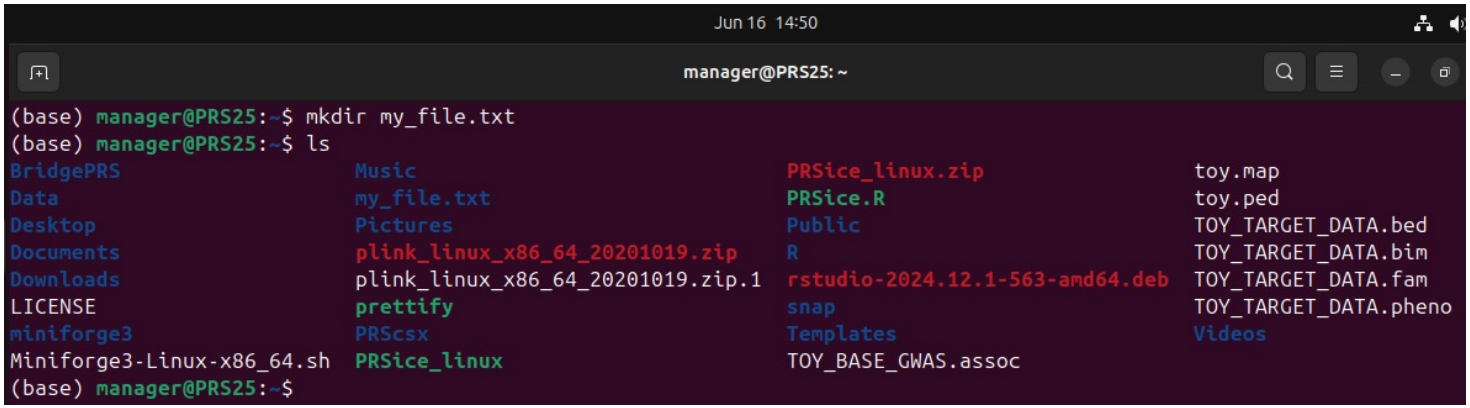
Navigating your file system



- ls : prints the names of the files and directories in the current directory in alphabetical order, arranged neatly

into columns.

- '*' option : Command displays the all file type.



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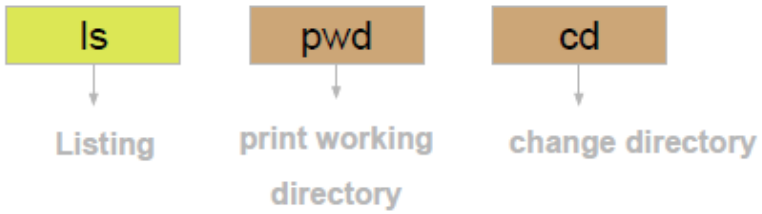
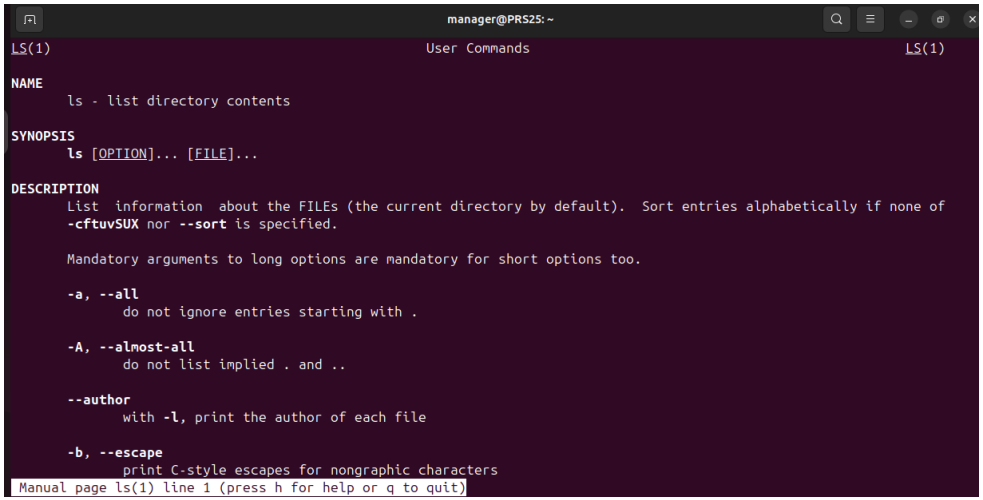


man (short for manual) displays detailed documentation (also referred as man page or man file) for bash commands.

It is a powerful resource to explore bash commands, understand their usage and flags. Some manual files are very long.

You can scroll through the file using your keyboard's down arrow or use the Space key to go forward one page and the b key to go backwards one page.

When you are done reading, hit q to quit.



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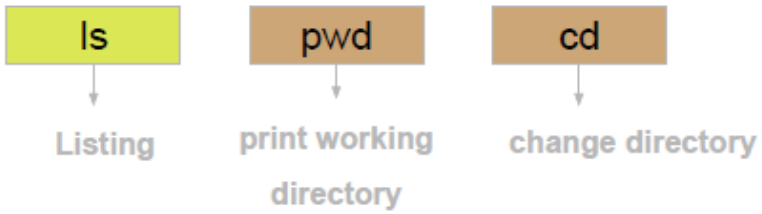

We can make the ls output more comprehensible by using the flag –F

- ls -F` command lists files and directories with indicators.
    - Directories are shown with a trailing slash (`/`).
    - Executable files are marked with an asterisk (`*`).
- Regular files have no special indicator.
- ls -a : see hidden directory/file that start with “.”.

The hidden files/folders are usually system-related data, configuration settings, or temporary files.

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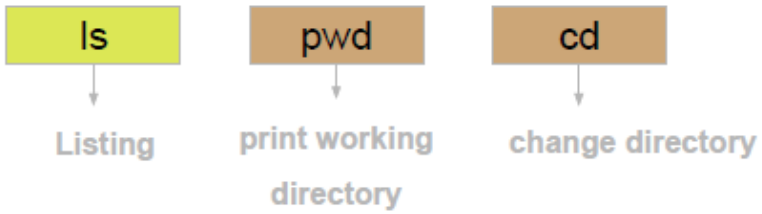

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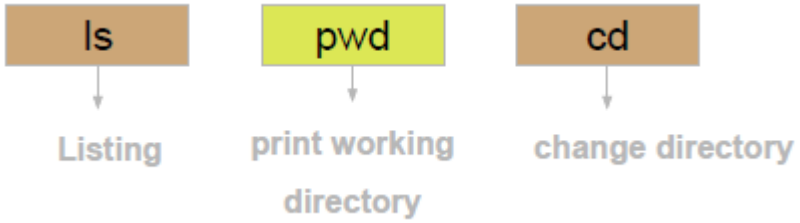
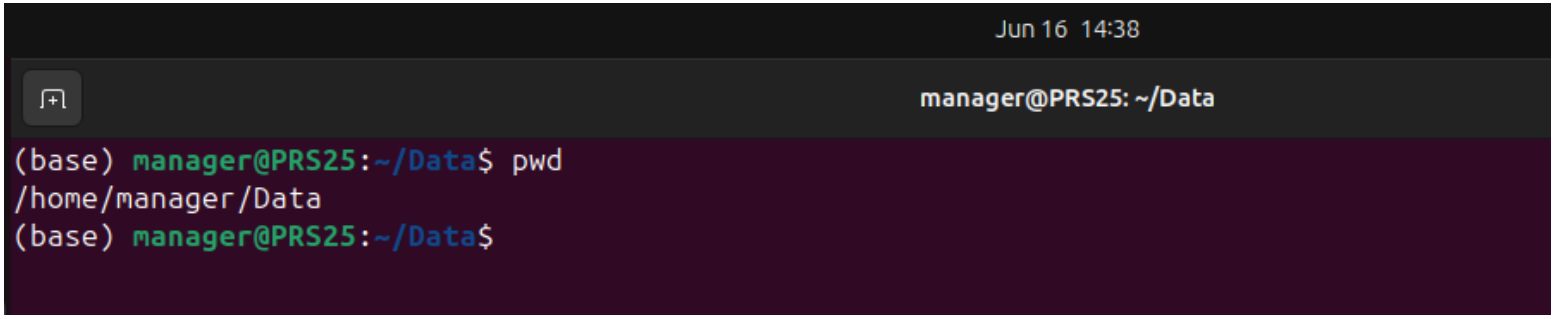


pwd : Prints the absolute path of the current working directory.



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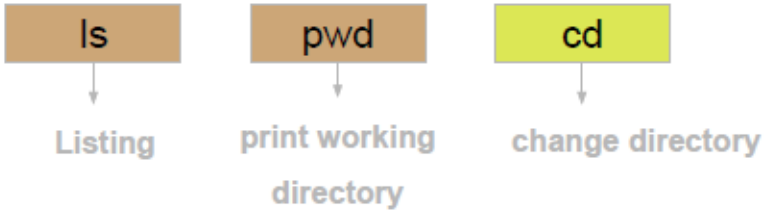
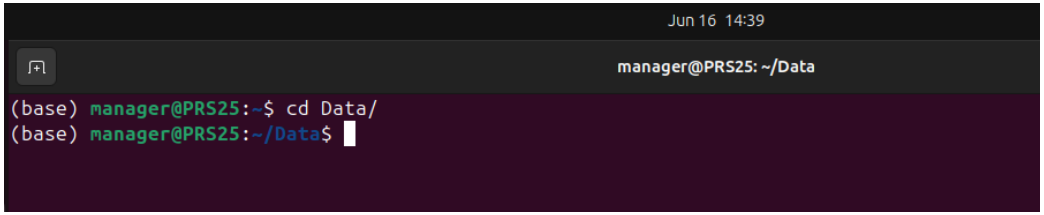


- The command to change locations in our file system is cd, followed by a directory name to change our working directory.

- Typing out file or directory names can waste a lot of time and it's easy to make typing mistakes.

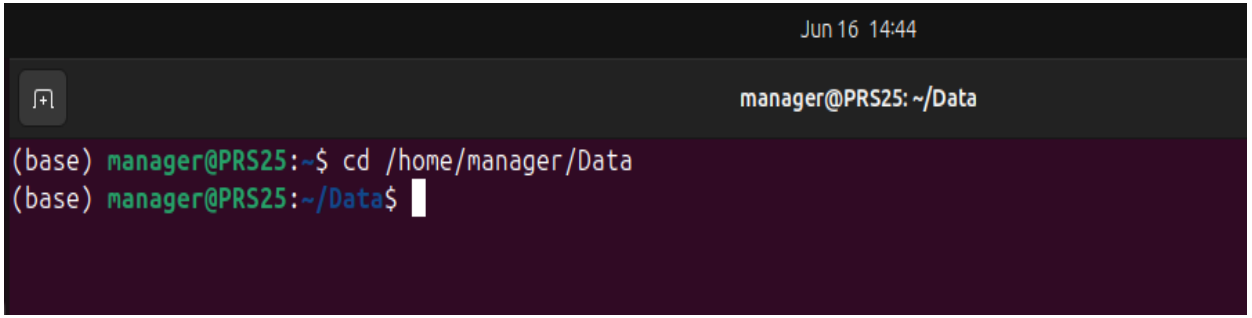
- Instead, we can use tab complete as a shortcut.

- When you start typing out the name of a directory or file, then hit the Tab key, the shell will try to fill in the rest of the directory or file name.



Full vs. Relative Paths

- The directories on the computer are arranged into a hierarchy.
- The cd command takes an argument which is a directory name.
- Directories can be specified using either a relative path or a full absolute path.



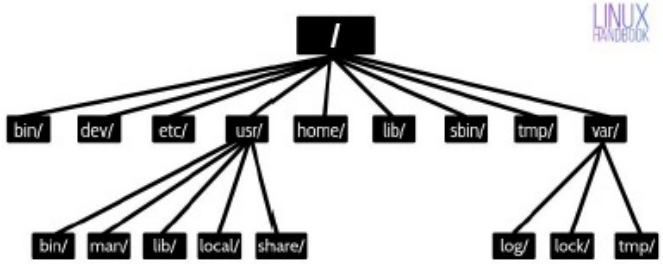
Absolute path, giving the full address from the home directory.

The very top of the hierarchy is a directory called / which is usually referred to as the root directory.

Relative path, giving only the address from the working directory.

Navigational Shortcuts : Root Directory

- The root directory (`/`) is the top-level directory of the file system.
- It contains critical system files and should not be directly modified.
- Avoid working in the root directory to prevent unintended system changes.
- The home directory is where users typically store their files.
- The tilde (`~`) is a shortcut that represents the user's home directory.
- The commands cd, and cd ~ are very useful for quickly navigating back to your home directory.

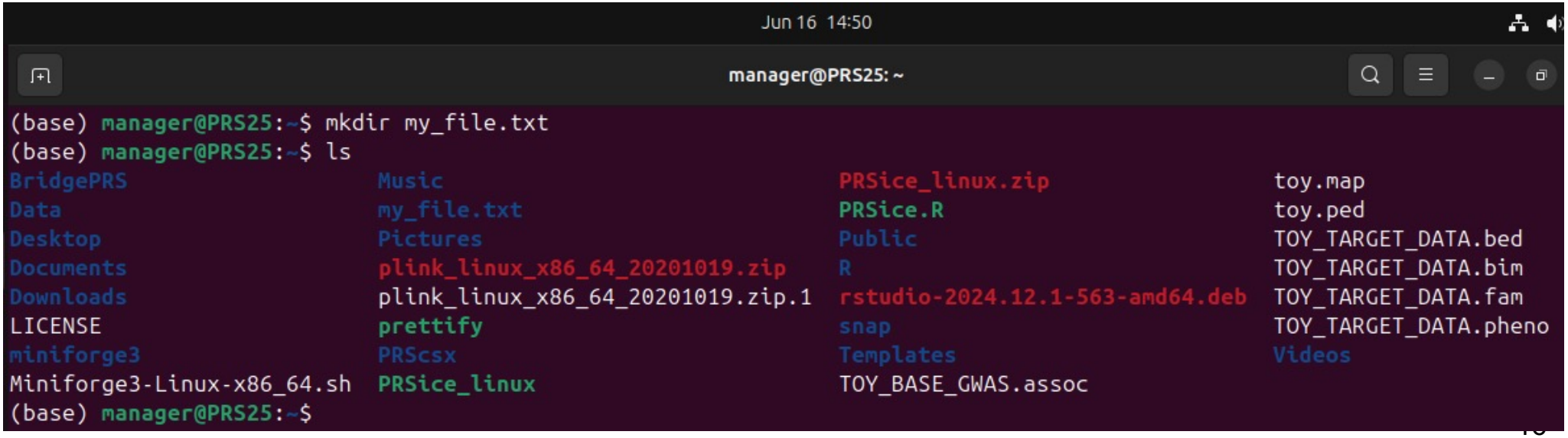


Working with Files and Directories : create a folder

mkdir | cp | mv | rm

The mkdir command is used to make a directory.

Enter mkdir followed by a space, then the directory name you want to create.

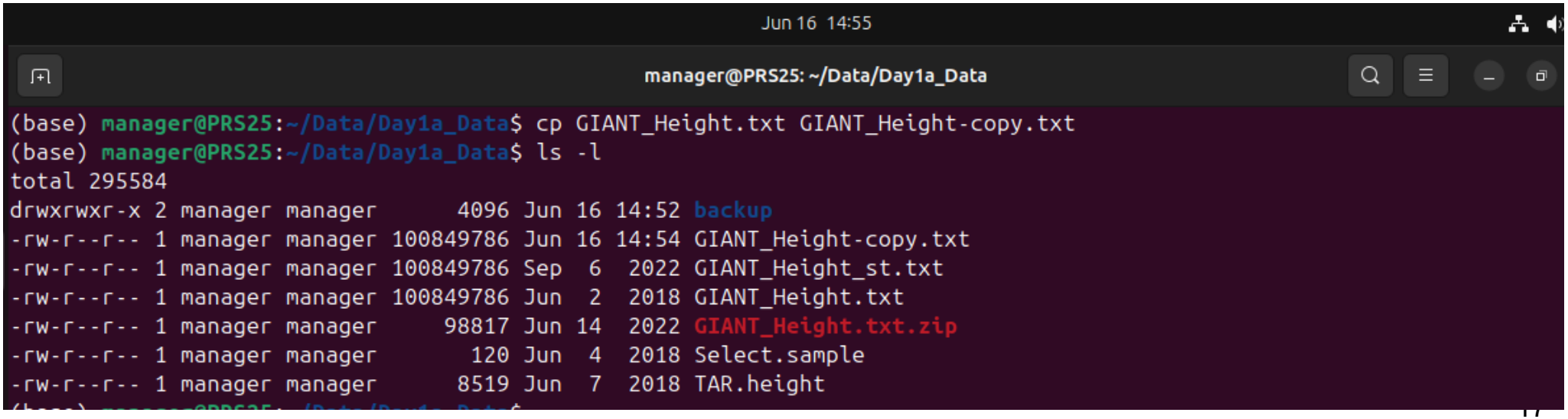


Working with Files and Directories : copy file/folder

mkdir | cp | mv | rm

Copying Files : When working with computational data, it's important to keep a safe copy of that data that can't be accidentally overwritten or deleted.

For this lesson, our raw data is our txt files. We don't want to accidentally change the original files, so we'll make a copy of them

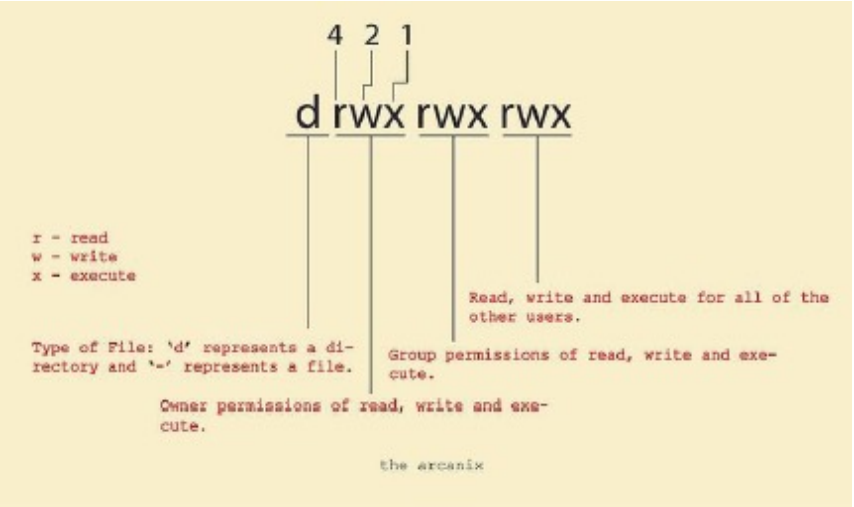


Working with Files and Directories : File permission

● Keep a safe copy of data that can't be accidentally overwritten or deleted.

● View the current permissions on a file using the -l (long) flag for the ls command.

● 10 slots in the permissions list: first character is file type, next 3 are owner permissions, next 3 are group permissions, last 3 are everyone else.



Working with Files and Directories : File permission

The "chmod" command modifies the read, write, and execute permissions of specified files and the search permissions of specified directories

```
chmod <operation> <file/dir>      # To change permissions of a file or
                                    directory.
chmod +r sample.sh                # Granting read permission to a file.
chmod +w sample.sh                # Granting write permission to a file.
chmod +x sample.sh                # Granting execute permission to a file.
```

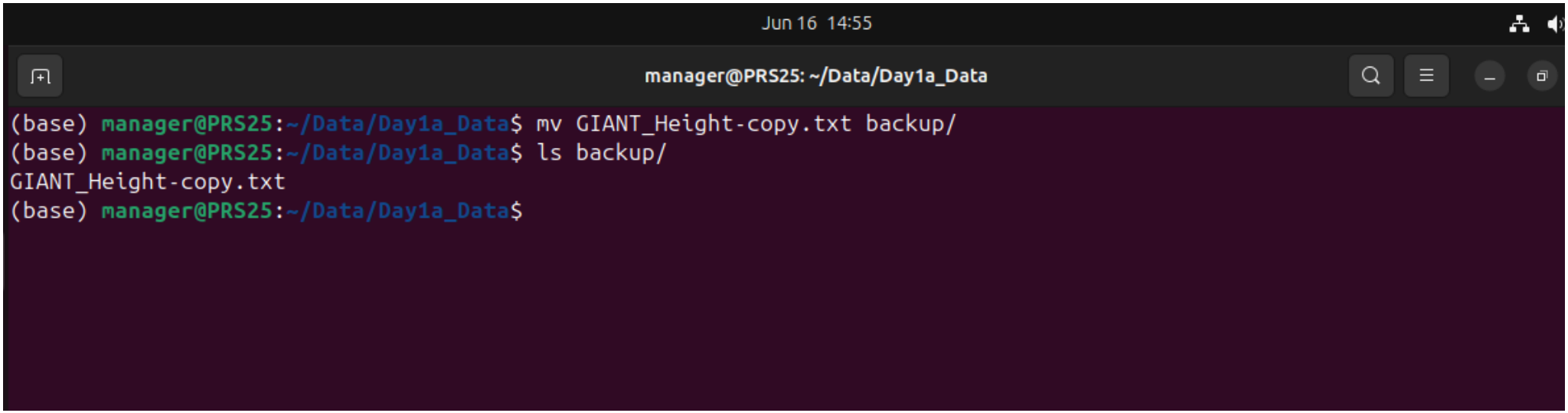
```
chmod -r sample.sh                # Revoking read permission from a file
chmod -w sample.txt               # Revoking write permission from a file
chmod -x sample.txt               # Revoking execute permission from a
                                    file.

sudo chmod -R -r directory        # Revoking read permission from a
                                    directory recursively.
```

Working with Files and Directories: Moving / Renaming

mkdir cp **mv** rm

- We can now move our backup file to another directory/file using the command mv
- The mv command is also how you rename files.



Working with Files and Directories: Moving / Renaming

The rm command permanently removes the file. Be careful with this command. It doesn't just nicely put the files in the Trash. They're really gone.

By default, rm will not delete directories. You can tell rm to delete a directory using the -r (recursive) option. Let's delete the backup directory we just made.

Examining Files

cat | less | head | tail | wc | grep

- One way to examine a file is to print out all of the contents using the program **cat**. This will print out all of the contents of the file to the screen.

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The program, **less**, is useful for this case. less opens the file as read only, and lets you navigate through it.

- The **wc** (word count) command is used to count the **numb**

- **grep** : Identifies sequences matching a specific pattern, We can use the -B argument for grep to return a specific number of lines before each match. The -A argument returns a specific number of lines after each matching line.

Filtering and Reshuffling Files

A very powerful feature of the terminal is the **awk** programming language, which allows us to extract subsets of a data file, filter data according to some criteria or perform arithmetic operations on the data. awk manipulates a data file by per- forming operations on its **columns** - this is extremely useful for scientific data sets because typically the columns features or variables of interest.

For example, we can use awk to produce a new results file that only contains SNP rsIDs (column 1), allele frequencies (column 4) and *P* -values (column 7) as follows:

```
awk '{ print $1,$4,$7}' GIANT_Height.txt > GIANT_Height_3cols.txt
```

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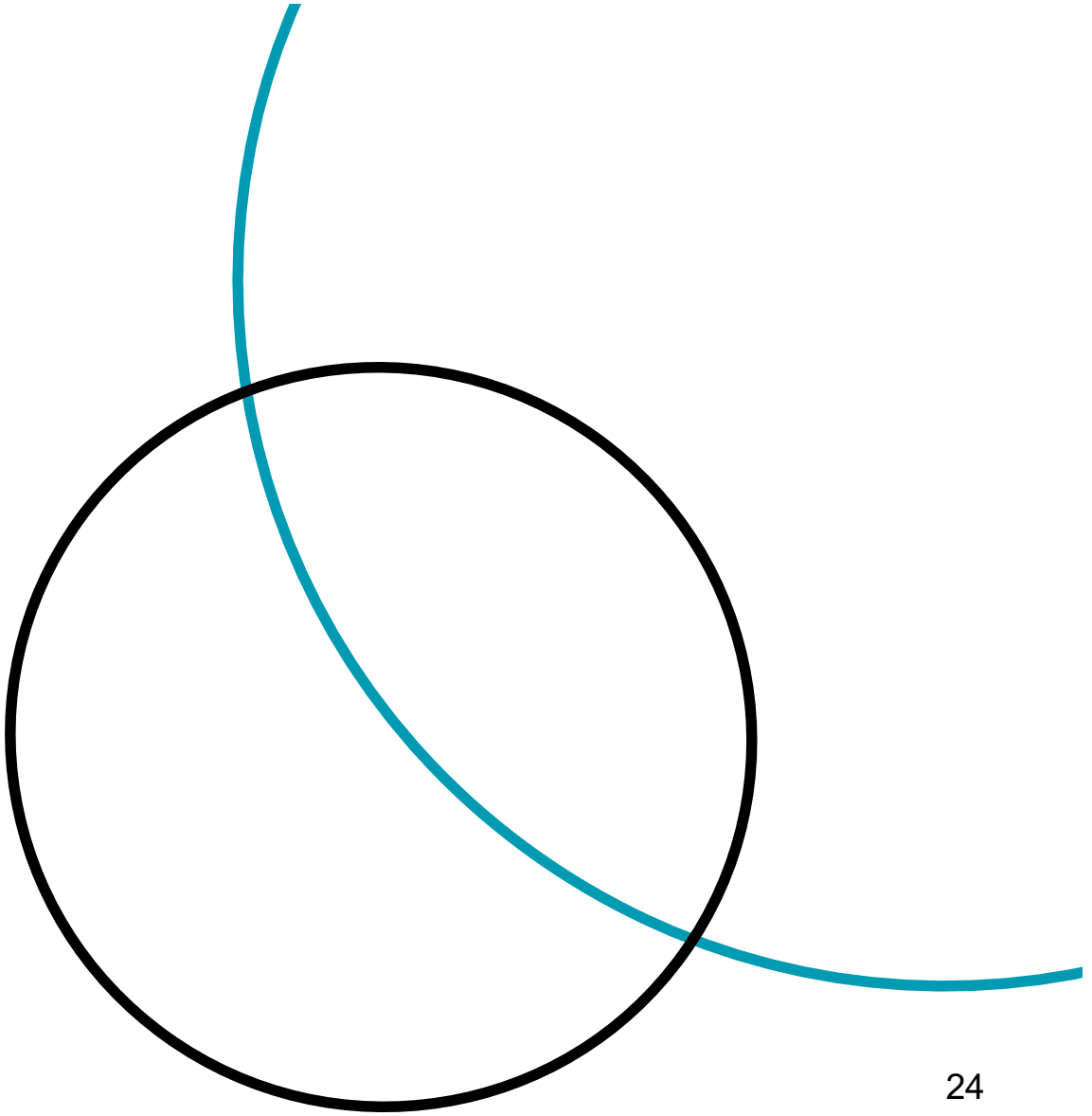
questions?



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thanks!