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GCATG LAT
ATGCATC
TGCATG
CATGCALGCATGCATGCATGC
CATGCATGCATCGCATGCATCGCA
CATGCATGCATGCATGCATGCATG
SCATGCATGCATGCATGCATGCATG
GCATGCATGCATGCATGCATGCATG
GCATGCATGCATGCATGCATGCAT
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GCATGCAT"
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Genetic Diversity: Analysis TEPPENIRCI / COBABACINCI Line Tuesday, June 17, 2025





Learning Objectives

Main

Our Understand the basic principles and differences between massive-parallel sequencing (MPS) platforms.

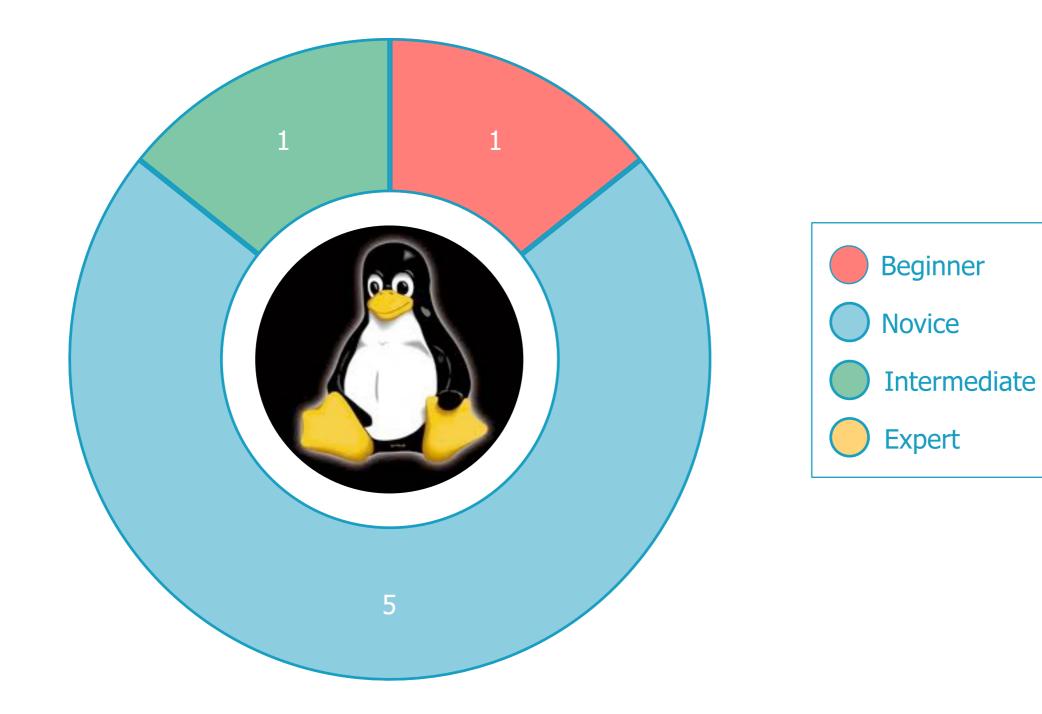
Our Understand sample indexing and sequence (read) types.

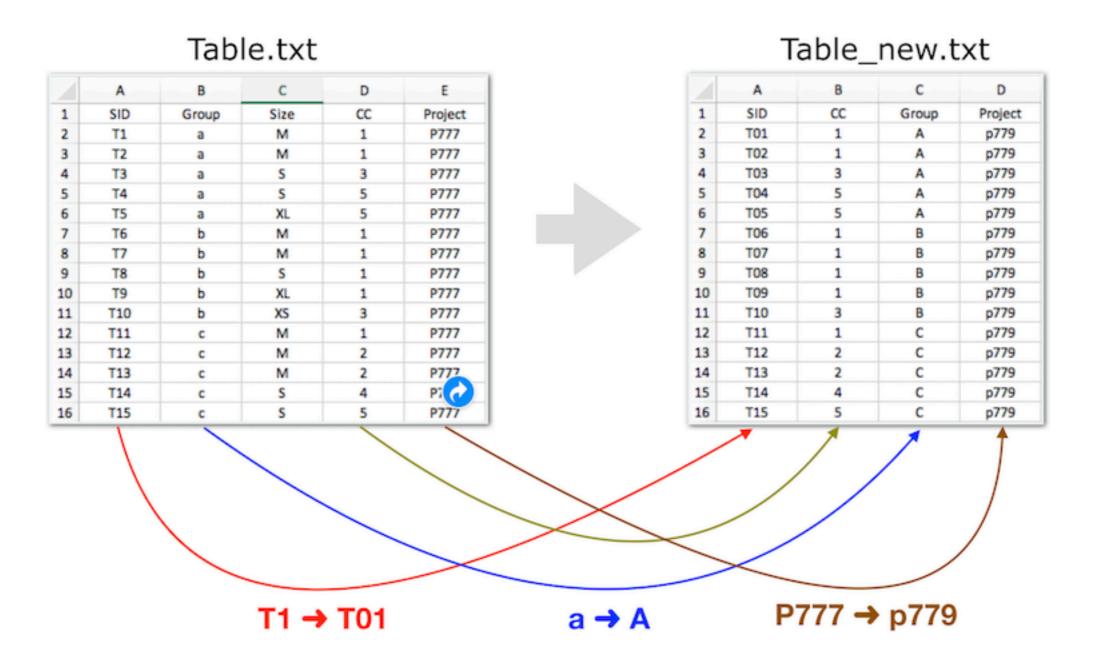
Minor

Our Understand the importance of a read data archive and be able to access such data.

Our Understand the advantages, limitations and applications of MPS in e.g. genomics, transcriptomics or metabarcoding.

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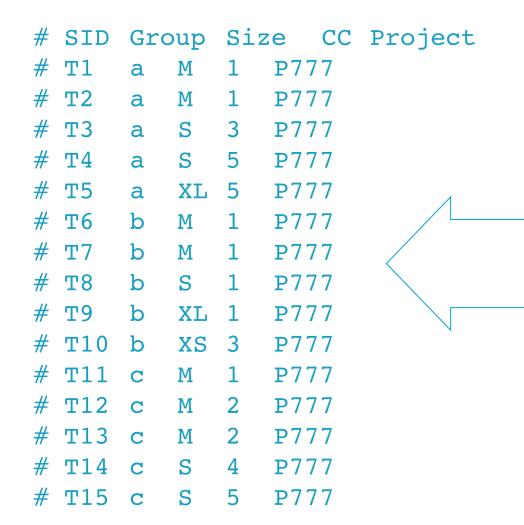
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Get Table
curl -0 https://www.gdc-docs.ethz.ch/GeneticDiversityAnalysis/GDA/data/Table.txt

Show Table
cat Table.txt





	A	В	С	D	E
1	SID	Group	Size	CC	Project
2	T1	а	м	1	P777
3	T2	а	M	1	P777
4	T3	а	S	3	P777
5	T4	а	S	5	P777
6	T5	а	XL	5	P777
7	T6	b	M	1	P777
8	T7	b	M	1	P777
9	T8	b	S	1	P777
10	T9	b	XL	1	P777
11	T10	ь	XS	3	P777
12	T11	с	M	1	P777
13	T12	c	M	2	P777
14	T13	с	M	2	P777
15	T14	с	S	4	PIC
16	T15	c	S	5	P777

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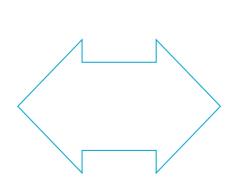
```
## Transform Table
awk '{
  if(NR==1) print $1,$4,$2,$5;
  else if(length($1)>2) print $1,$4,toupper($2),"p779"; # change project number
  else print "T0"substr($1,2,3),$4,toupper($2),"p779" # rename sample < 10</pre>
}' Table.txt > Table_new.txt
```

```
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```

exclude header line

Show NEW Table cat Table new.txt

```
# SID CC Group Project
# T01 1 A p779
# T02 1 A p779
# T03 3 A p779
# T04 5 A p779
# T05 5 A p779
# T06 1 B p779
# T07 1 B p779
# T08 1 B p779
# T09 1 B p779
# T10 3 B p779
# T11 1 C p779
# T12 2 C p779
# T13 2 C p779
# T14 4 C p779
# T15 5 C p779
```



Table_new.txt

	Α	В	С	D
1	SID	CC	Group	Project
2	T01	1	A	p779
3	T02	1	A	p779
4	T03	3	A	p779
5	T04	5	A	p779
6	T05	5	A	p779
7	T06	1	В	p779
8	T07	1	В	p779
9	T08	1	В	p779
10	T09	1	В	p779
11	T10	3	B	p779
12	T11	1	C	p779
13	T12	2	C	p779
14	T13	2	C	p779
15	T14	4	C	p779
16	T15	5	С	p779

Alternative solution providing a bash script:

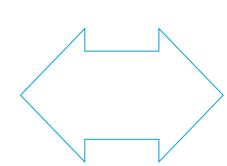
```
## Get Table
curl --verbose -0 https://gdc-docs.ethz.ch/GeneticDiversityAnalysis/GDA/scripts/reformat_table.sh
```

Make the script executable
chmod a+x reformat_table.sh

run the script but make sure the Table.txt file is in the same directory
./reformat_table.sh Table.txt

Show NEW Table

```
cat Table new.txt
# SID CC Group Project
# T01 1 A p779
# T02 1 A p779
# T03 3 A p779
# T04 5 A p779
# T05 5 A p779
# T06 1 B p779
# T07 1 B p779
# T08 1 B p779
# T09 1 B p779
# T10 3 B p779
# T11 1 C p779
# T12 2 C p779
# T13 2 C p779
# T14 4 C p779
# T15 5 C p779
```



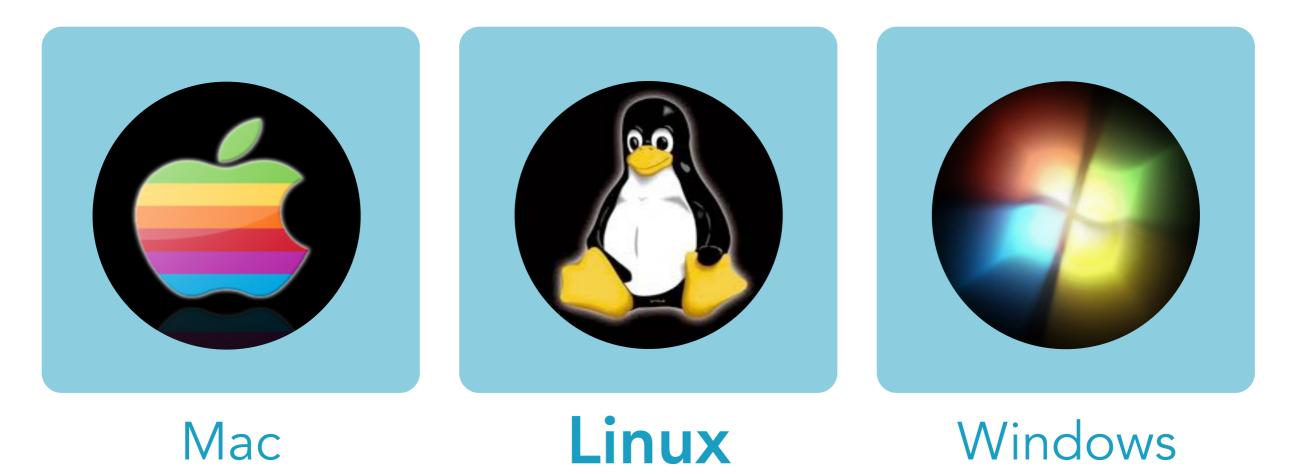
Table_new.txt

	Α	В	С	D
1	SID	CC	Group	Project
2	T01	1	A	p779
3	T02	1	A	p779
4	T03	3	A	p779
5	T04	5	A	p779
6	T05	5	A	p779
7	T06	1	В	p779
8	T07	1	В	p779
9	T08	1	В	p779
10	T09	1	В	p779
11	T10	3	В	p779
12	T11	1	С	p779
13	T12	2	С	p779
14	T13	2	C	p779
15	T14	4	С	p779
16	T15	5	С	p779
-	-	4		





Different OSs and Versions

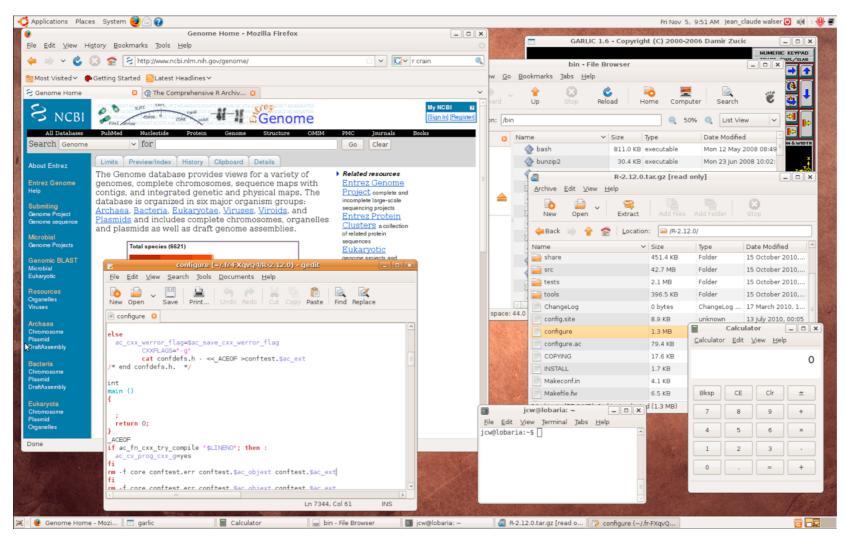






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A graphical user interface (GUI) - often pronounced gooey - an interface that allows the user (you) to interact with programs in more ways than typing.



GUIs are nice but limited. Don't be afraid to use the terminal!

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"Where there is a shell, there is a way,"

GUIs were introduced in reaction to the steep learning curve of **commandline interfaces (CLI)**, which require commands to be typed on the keyboard. Since the commands available in command line interfaces can be numerous, complicated operations can be completed using a short sequence of words and symbols. **This allows for greater efficiency**, **productivity once many commands are learned**, **and better reproducibility**.

Once upon a time, computers spoke only in cryptic spells typed by brave command-line wizards. These early users, armed with nothing but a blinking cursor and pure willpower, tamed machines by uttering arcane incantations like grep, awk, and chmod 775. It was powerful—but only if you had the patience of a monk and the memory of an elephant.

Then came the Graphical User Interface (GUI), a shiny new toy meant to rescue humanity from mistyping rm -rf / and accidentally deleting the universe. GUIs introduced buttons, windows, and icons - tools for the rest of us, who prefer dragging and clicking over memorizing which flag does what in a 12-word command.

Yet, ironically, while GUIs made computers more approachable, the command line didn't go away. Why? Because once you master its secret language, you can perform sorcery at lightning speed: batchrenaming files, deploying software, or bending entire servers to your will - often in just a single line. It's like riding a bicycle that can also fly, once you stop falling off.

So yes, GUIs are friendly. But CLIs? They're fast, efficient, and reproducible. Just... maybe keep a cheat sheet handy.

X

Shell - Terminal

Shell is a UNIX term for the interactive user interface with an operating system. The shell is the layer of programming that understands and executes the commands a user enters.

Bourne-Shell (sh) Korn-Shell (ksh) C-Shell (csh)

TC-Shell (tcsh)

Bourne-Again-Shell (bash)

Debian Almquist Shell (dash)

Z-Shell (zsh)

A-Shell (ash) PowerShell / cmd.exe What do I have? \$> echo \${SHELL}

Bash > zcat file.gz

Zsh > zcat < file.gz

Bash > sed -i 's/d/D/2' file.txt

Zsh > sed -i"" "s/d/D/2" file.txt



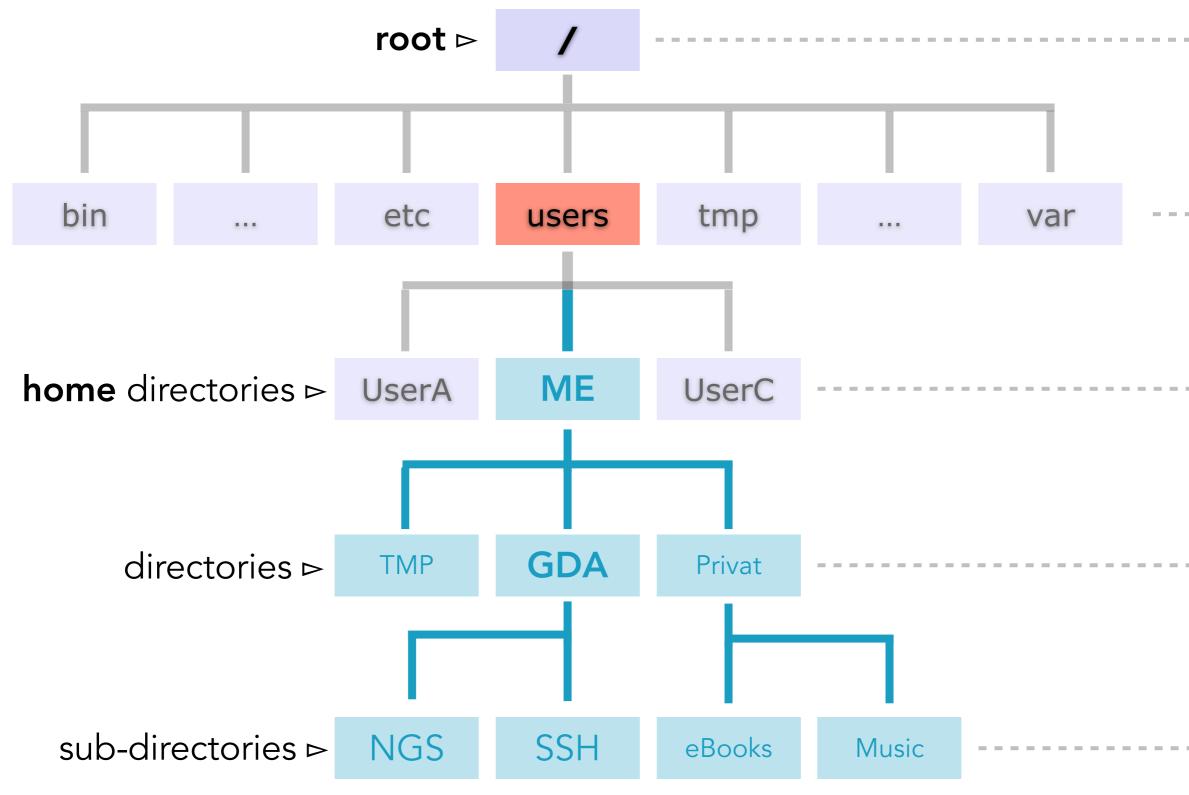
Change Shell

echo \${SHELL}
#which zsh
#which bash

Bash > Zsh
chsh -s /bin/bash

Zsh > Bash
chsh -s /bin/zsh

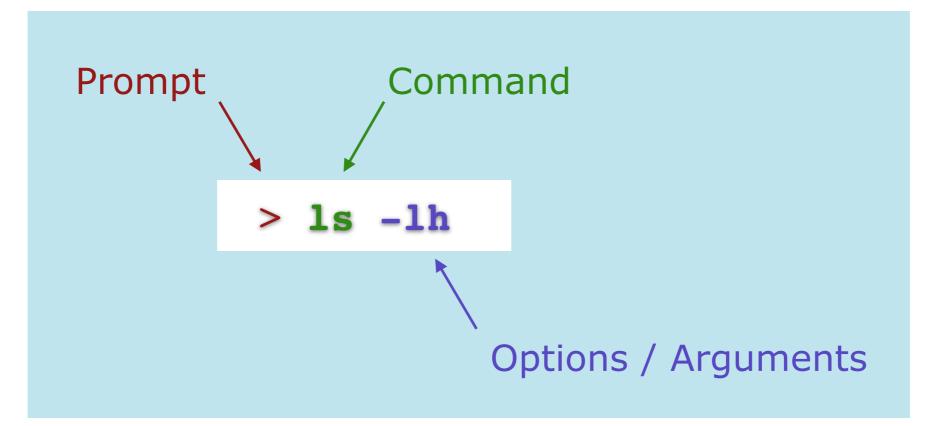
Which one is better? Both shells will get the job done. The bash is a bit outdated (version 3.2 -2006). Many of the conveniences provided by zsh can be made available in bash. It seems zsh is more helpful to newer shell user.



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Command - Line



Built-in Help

> info <command>

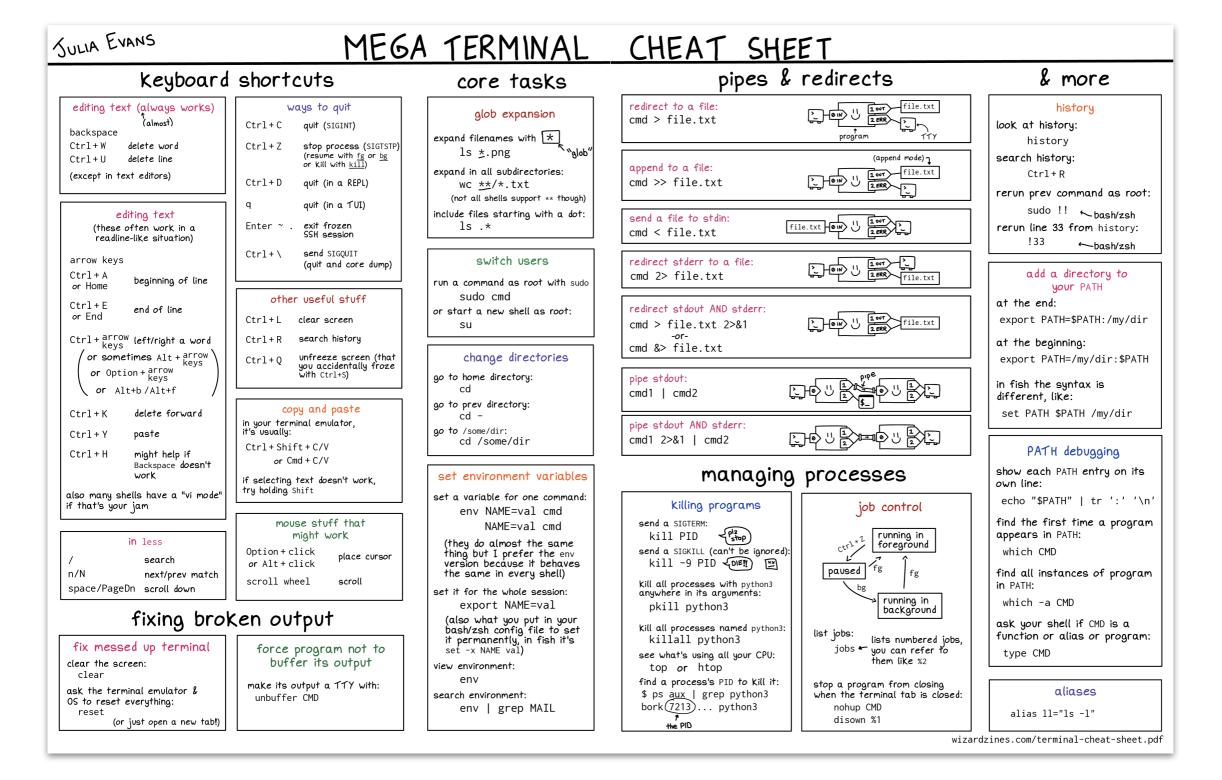
> info ls

> man <command>

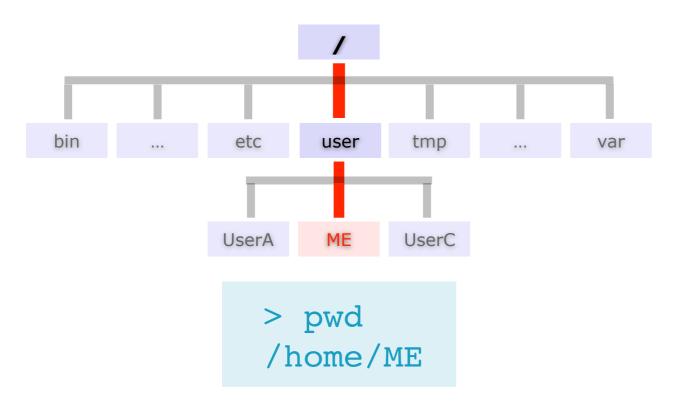
> man ls

* press Q to leave info or manual

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pwd - print name of current/working directory



> pwd
/home/ME
> cd \${HOME}
> pwd
/home/ME
> cd ~
> pwd
/home/ME
> cd
/home/ME
> cd
/home/ME

GS

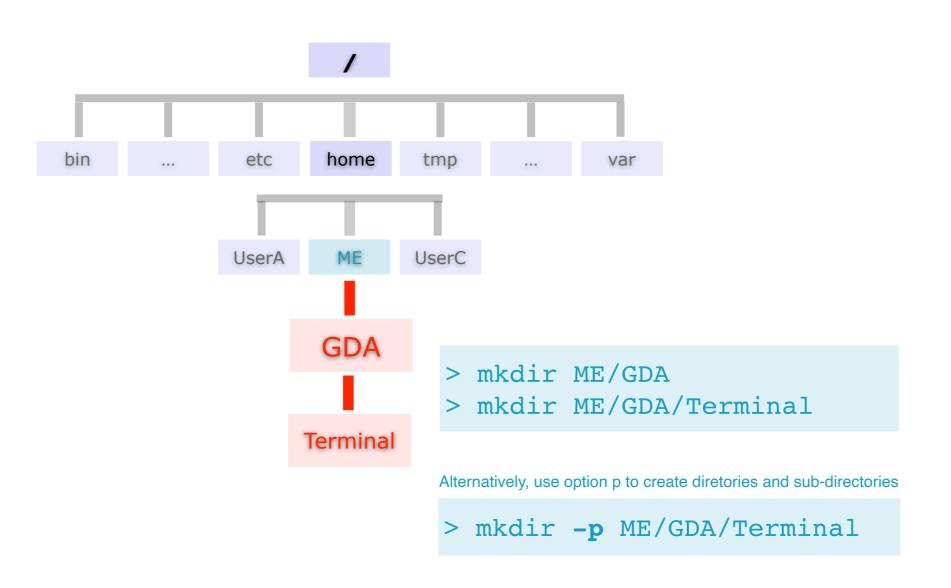
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mkdir - creating/making directories



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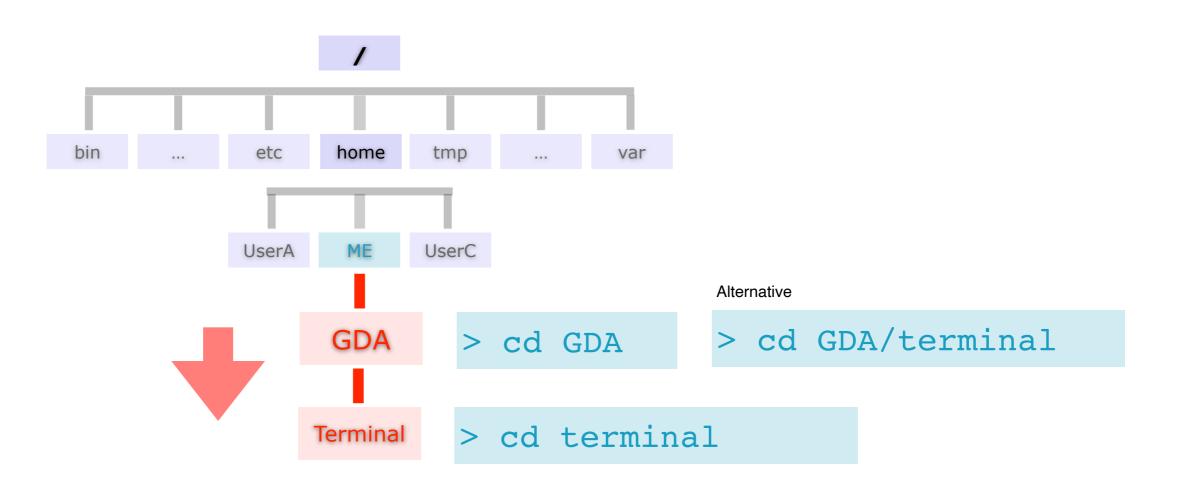
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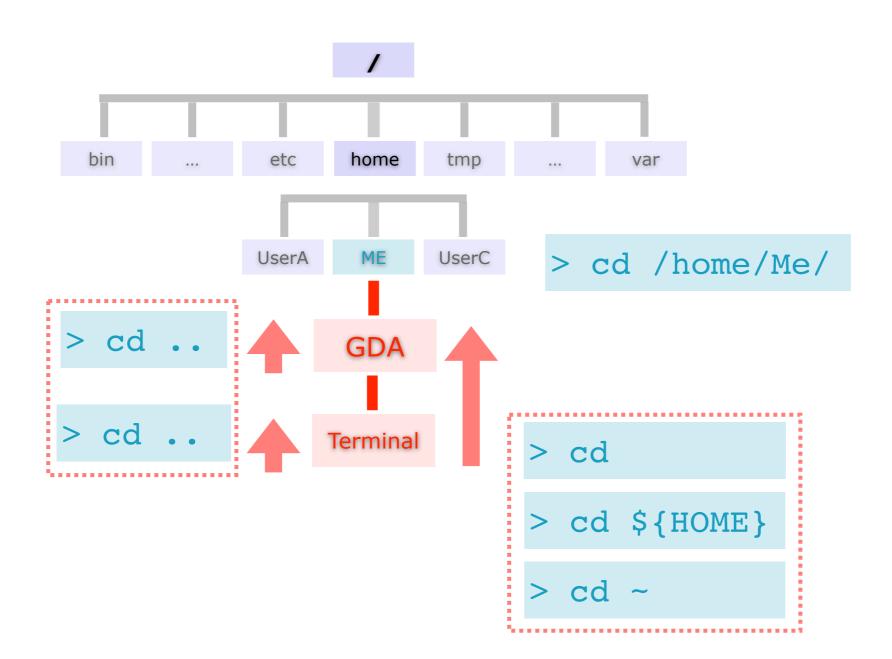
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cd - change directory







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Local Software Management

Software Dependencies

Parallel Versions GD

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Package, **dependency and environment management** for any language— Python, R, Ruby, Lua, Scala, Java, JavaScript, C/C++, FORTRAN. Conda as a package manager helps you find and install packages. If you need a package that requires a different version of Python, you do not need to switch to a different environment manager, because conda is also an environment manager. With just a few commands, you can set up a totally separate environment to run that different version of Python, while continuing to run your usual version of Python in your normal environment.



BINCONDA®

Bioconda is a **channel for the conda package manager** specialising in bioinformatics software. The conda package manager makes installing software a vastly more streamlined process. Conda is a combination of other package managers you may have encountered, such as pip, CPAN, CRAN, Bioconductor, apt-get, and homebrew. Conda is both language- and OSagnostic, and can be used to install C/C++, Fortran, Go, R, Python, Java etc programs on Linux, Mac OSX, and Windows.

```
python --version
# Python 2.7.15
bwa
# -bash: bwa: command not found
blast -help
# -bash: blast: command not found
```

```
conda info --envs
source activate aligners
conda info --envs
python --version
# Python 3.6.7
bwa
blastn -help
```



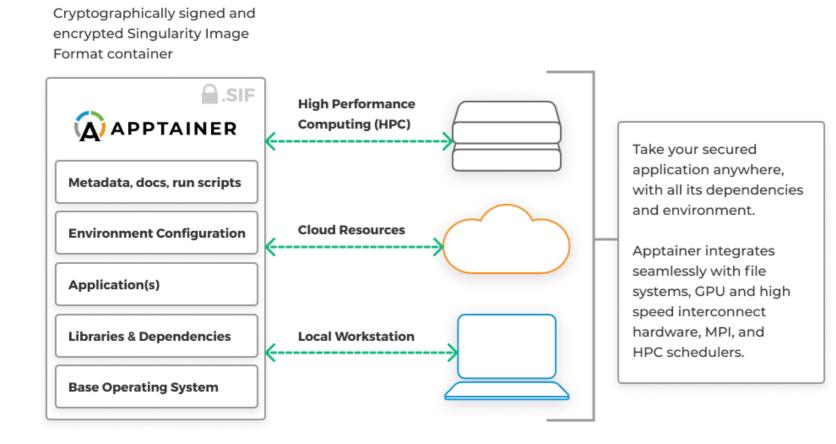


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- 1. **Portability**: Containers are portable and can be deployed across different systems without modifications. They encapsulate the application and its dependencies, ensuring consistent behaviour regardless of the underlying infrastructure.
- 2. **Scalability**: Containers enable applications to be easily scaled horizontally by running multiple instances of the same container across a cluster of machines. This allows for efficient resource utilisation and the ability to handle increased workload demands.
- 3. **Efficiency**: Containers share the host operating system's kernel and only require the necessary dependencies, making them lightweight and resource-efficient compared to traditional virtual machines.
- 4. **Isolation**: Containers provide a level of isolation between the application and the host operating system, as well as between different containers. This isolation helps prevent conflicts between applications and improves security by limiting the impact of any potential vulnerabilities.
- 5. **Rapid deployment**: Containers simplify the deployment process by providing a consistent environment. They can be quickly started, stopped, and updated, allowing for fast and efficient application deployment and rollbacks.

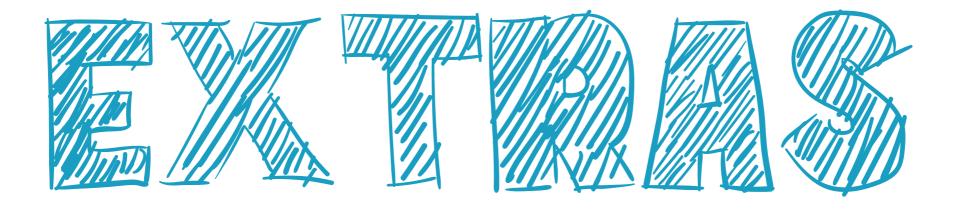
Containerisation is a technique used in software development and deployment that **allows applications and their dependencies to be packaged together into self-contained units called containers**. Containers provide a lightweight and isolated runtime environment that can run consistently across different computing environments, such as laptops, servers, or cloud platforms.

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- 1. **Reproducibility**: Singularity enables researchers to create and distribute containers that encapsulate their entire software stack, including the application, libraries, and dependencies. This ensures that the environment remains consistent and reproducible across different systems.
- 2. **Compatibility** with HPC: Singularity is designed to seamlessly integrate with HPC clusters and resource managers commonly used in scientific computing. It allows users to leverage specialised hardware resources, such as GPUs and high-performance interconnects, while still providing isolation and security.
- 3. **Security** and user isolation: Singularity employs a security model that focuses on user isolation and containment. It ensures that users can run containers securely without requiring escalated privileges, making it suitable for multi-user HPC environments.
- 4. **Mobility**: Singularity containers are portable and can be easily shared, distributed, and run on different systems, including local workstations, clusters, and cloud platforms.
- 5. **Interoperability**: Singularity supports a wide range of container image formats, including Docker, making it possible to leverage existing container images and workflows.

Singularity (Apptainer) is an open-source **containerisation platform** that focuses on providing secure and reproducible environments for scientific and **high-performance computing** (HPC) workloads. It was developed specifically to address the challenges faced by researchers and scientists when working with complex software stacks and specialized computing resources.



```
#!/usr/bin/perl -w
use strict;
my $input fasta=$ARGV[0];
open(IN, "<$input fasta") || die ("Error opening $input fasta $!");</pre>
my $line = <IN>;
print $line;
while ($line = <IN>)
{
chomp $line;
 if ($line=~m/^>gi/) { print "\n",$line,"\n"; }
else { print $line; }
}
print "\n";
perl fa2one.pl multiple.fa > one.fa
```



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```
#!/usr/bin/perl -w
use strict;
# Perl script to reformat fasta files
# Usage: perl fa2one.pl multiple.fa > one.fa
my $input fasta=$ARGV[0];
open(IN, "<$input fasta") || die ("Error opening $input fasta $!");</pre>
my $line = <IN>;
print $line;
while ($line = <IN>)
{
                 ✓ limiting factor
 chomp $line;
if ($line=~m/^>gi/) { print "\n",$line,"\n"; }
 else { print $line; }
}
print "\n";
```

perl fa2one.pl multiple.fa > one.fa

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