Introduction to Bioinformatics

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about me

Alexandros C. Dimopoulos, Ph.D.

- BSc Electrical and Computer Engineer, NTUA (2004)
- Ph.D. in Computer Science, NTUA (2009)
- Adjunct Lecturer, Harokopio University (HUA) (2010-2020)
- Post-Doc Researcher, BSRC Al. Fleming (2012 -)
- M.Sc. “Data Science and Information Technologies” DIT, UoA (2017 -)
- Lecturer, Hellenic Naval Academy (2020 - 2025)
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Course Overview

1. Tuesday, 14 October 2025: Introduction to GNU/Linux and to basic commands
2. Tuesday, 4 November 2025: Introduction to the R programming language and to RStudio utilization
3. Tuesday, 11 November 2025: More advanced programming in R and introduction to Bioconductor
4. Tuesday, 18 November 2025: Usage of CLI tools such as bedtools, vcftools, samtools etc.
5. Tuesday, 2 December 2025: SNP calling Pipelines

GNU/Linux

GNU/Linux

Linux is a Unix-like computer operating system assembled under the model of free and open-source software development and distribution. The defining component of Linux is the Linux kernel, an operating system kernel first released on September 17, 1991 by Linus Torvalds. The Free Software Foundation uses the name GNU/Linux to describe the operating system, which has led to some controversy.

Linus Torvalds & Richard Stallman

GNU/Linux distributions (distros)

GUI vs CLI

GUI

Graphical User Interface

CLI

Command-line interface

Shell

- The oldest way of communicating with the computer
- Not always very (user) friendly

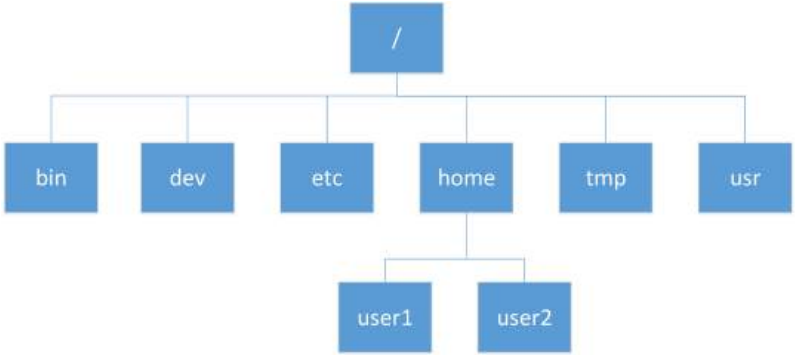
```
paste <(cat out_23Genes.txt | cut -f16-18 |awk '{
    print "chr"$1"\t"$2-1"\t"$2 }') <(cat out_23Genes.txt
    ) >out_23Genes.new.bed
```

- Very useful for combining existing commands/tools and redirection (pipes)
- Various different shells: `Bash`, `Tcsh/Csh`, `Ksh`, `Zsh`, `Fish`, ...

Bash (Bourne-again shell)

- `$ <user privilages>`
- `# <root privilages>`
- auto-completion while typing by pressing the Tab key
- program execution in the foreground
- program execution in the background (&)

File structure I



File structure II

```
ls -l /
total 120
drwxr-xr-x   4 root root 12288 Jul 24 11:30 bin
drwxr-xr-x   3 root root  4096 Aug  7 08:14 boot
drwxr-xr-x  20 root root  3500 Sep 13 11:41 dev
drwxr-xr-x 220 root root 16384 Sep 12 19:43 etc
lrwxrwxrwx   1 root root    15 Apr  4 14:42 home
....
```

- EVERYTHING is a file (files, directories, hard-drives, modems, keyboards, printers)
- home folder (~)
  - unlimited access/rights from the user
  - cd ~ or just cd

Change directory (folder)

```
$ cd /tmp
$ pwd
/tmp
$ cd ~
$ pwd
/home/alexdem
$ cd /tmp
$ cd
$ pwd
/home/alexdem
```

List directory contents

```
$ ls /etc/dhcp/
debug  dhclient.conf  dhclient-enter-hooks.d  dhclient-exit-hooks.d
$ ls /etc/dhcp/ -1 # number 1
debug
dhclient.conf
dhclient-enter-hooks.d
dhclient-exit-hooks.d
$ ls /etc/dhcp/ -l # smallcase L
total 16
-rw-r--r-- 1 root root 1426 Nov 26  2016 debug
-rw-r--r-- 1 root root 1735 Nov 26  2016 dhclient.conf
drwxr-xr-x 2 root root 4096 Jul 18 11:33 dhclient-enter-hooks.d
drwxr-xr-x 2 root root 4096 Jul 24 11:30 dhclient-exit-hooks.d
```

Help

```
$ man ls
LS(1)                    User Commands                    LS(1)

NAME
       ls - list directory contents

SYNOPSIS
       ls [OPTION]... [FILE]...

DESCRIPTION
       List information about the FILEs (the current directory by default).
       Sort entries alphabetically if none of -cftuvSUX nor --sort is
           specified.
       Mandatory arguments to long options are mandatory for short options
           too.
       -a, --all
              do not ignore entries starting with .
```

- google

Total and relevant Paths

- . → current directory
- .. → one level back

```
$ pwd
/tmp/directory1/directory2
$ cd .
$ pwd
/tmp/directory1/directory2
$ cd ..
$ pwd
/tmp/directory1/
$ cd /tmp/directory3
$ pwd
/tmp/directory3
$ cd ../directory2
$ pwd
/tmp/directory2
```

Creating and modifying directories

- create directory

  ```
  mkdir dirName
  ```

- rename directory

  ```
  mv oldDirName newDirName
  ```

- move directory

  ```
  mv oldDirName /tmp/newDirName
  ```

- remove (delete) directory

  `rmdir dirName` (if it is empty)

  `rm -r dirName` (even if not empty; any files are first deleted and then removed)

File permissions I

```
drwxr-xr-x 220 root root 16384 Sep 12 19:43 etc
```

3 access categories:

1. user : refers only to the user that owns the file
2. group : refers to all the users that belong to the specific group
3. other : refers to all the system users

Permission	Meaning for directory	Meaning for file
r	List the directory	Read contents
w	Create or remove files	Write contents
x	Access files and subdirectories	Execute

File permissions II

Value	Meaning
0	- - -
1	- -x
2	-w-
3	-wx
4	r- -
5	r-x
6	rw-
7	rwx

```
chmod 740 fname
```

Program execution

- `./a.out` (if the executable is in the current directory)
- `/<PATH_TO_FILE>/a.out`

Useful commands & programs I

cp - copy

```
cp source destination
```

mv - move

```
mv source destination
```

cat - concatenate files and print on the standard output

```
cat text_file
```

echo - display a line of text

```
$echo "hello world"
hello world
```

Useful commands & programs II

head - output the first part of files

`head text_file`

`head -n 30 text_file` (first 30 lines)

tail - output the last part of files

`tail text_file`

`tail -n 30 text_file` (last 30 lines)

Useful commands & programs III

```
$cat n.txt
101
102
103
104
105
106
...
199
200
```

```
$head -n 5 n.txt
101
102
103
104
105
```

```
$tail -n 3 n.txt
198
199
200
```

Useful commands & programs IV

more - file perusal filter for crt viewing

```
more text_file
cat text_file | more
```

less - opposite of more

```
less text_file
cat text_file| less
```

Redirection

- > : redirecting output (stdout) into a file - create/overwrite a new/existing file
  e.g. `ls > /tmp/out.txt`
- > > : redirecting output (stdout) into a file - append to an existing file
  e.g. `ls >> /tmp/out.txt`
- 2> : redirecting standard error (stderr) into a file
  e.g. `ls 2> /tmp/out_error.txt`
- &> : redirecting both stdout and stderr into a file
  e.g. `ls &> /tmp/out_stdout_error.txt`
- | : redirecting (stdout) to be used as input by another command
  e.g. `cat a.txt | less`

Additional useful command & programs I

grep - print lines matching a pattern

```
grep pattern text_file
cat text_file | grep pattern
```

```
$cat n.txt
1
3
5
7
9
11
13
```

```
$grep 1 n.txt
1
11
13
```

Additional useful command & programs II

grep options:

- -i: ignore case
- -v: invert match
- -n: line number
- -A NUM: print NUM lines after-context
- -B NUM: print NUM lines before-context
- . . .

Additional useful command & programs III

cut - remove sections from each line of files

```
cut text_file -f 10 -d ","
cat text_file | cut -f 10 -d ","
```

```
$cat n.txt
a,b,c,d,e
f,g,h,i,j
```

```
$cut -f 2,3 -d "," n.txt
b,c
g,h
```

Additional useful command & programs IV

tr - translate or delete characters

```
cat text_file | tr SET1 SET2
```

```
$cat n.txt
1,2,3,4,5
6,7,8,9,10
```

```
$cat n.txt| tr "," "_"
1_2_3_4_5
6_7_8_9_10
```

Additional useful command & programs V

gzip - compress files

```
gzip text_file
```

gunzip - expand files

```
gunzip file.gz
```

zcat - cat for compressed files

```
zcat file.gz
```

zless - less for compressed files

```
zless file.gz
```

Additional useful command & programs VI

tree - list contents of directories in a tree-like format

```
tree /usr/
tree /usr/ -d  (List directories only)
```

find - search for files in a directory hierarchy

```
find . -name filename.txt
find . |grep filename.txt
find . -iname filename.txt  (ignore case)
find . -type f -iname filename.txt  (find files only)
find . -type f -iname -perm 0777 filename.txt  (find files only with 777 permissions)
```

Additional useful command & programs VII

seq - print a sequence of numbers

```
seq 3
1
2
3
```

```
$seq 3
1
2
3
```

```
$seq 11 13
11
12
13
```

```
$seq -f alex%g 4 6
alex4
alex5
alex6
```

Additional useful command & programs VIII

sort - sort lines of text files

```
sort file
cat file|sort
```

```
$cat n.txt
10
5
4
3
2
1
```

```
$cat n.txt|sort -n
1
2
3
4
5
10
```

```
$cat n.txt|sort
1
10
2
3
4
5
```

Additional useful command & programs IX

wc - print newline, word, and byte counts for each file

```
$cat n.txt
1
2
3
4
5
$wc n.txt
5 5 10 n.txt
```

Variables

```
user@pc$ STR="Hello World!"
user@pc$ echo $STR
Hello World!
```

Variables

```
user@pc$ STR="Hello World!"
user@pc$ echo $STR
Hello World!
```

```
user@pc$ a=1
user@pc$ b=$((a + 1 ))
user@pc$ echo $a "+ 1 = " $b
1 + 1 = 2
```

Conditional

```
user@pc$ a=1
user@pc$ if [ $a = 1 ]; then
user@pc$ echo true
user@pc$ fi
true
```

Conditional

```
user@pc$ a=1
user@pc$ if [ $a = 1 ]; then
user@pc$ echo true
user@pc$ fi
true
```

```
user@pc$ a=2
user@pc$ if [ $a = 1 ]; then
user@pc$ echo true
user@pc$ else
user@pc$ echo false
user@pc$ fi
false
```

For loop I

```
user@pc$ for i in a b c d
user@pc$ do
user@pc$ echo $i
user@pc$ done
a
b
c
d
```

For loop I

```
user@pc$ for i in a b c d
user@pc$ do
user@pc$ echo $i
user@pc$ done
a
b
c
d
```

```
user@pc$ for i in `seq 1 5`
user@pc$ do
user@pc$ echo $i
user@pc$ done
1
2
3
4
5
```

For loop II

```
user@pc$ for i in `ls /usr/sbin`
user@pc$ do
user@pc$ echo $i
user@pc$ done
a2disconf
a2dismod
a2dissite
a2enconf
a2enmod
a2ensite
a2query
...
```

Executable scripts

```
user@pc$ cat runme.sh
#!/bin/bash
echo "Hello World"
```

Executable scripts

```
user@pc$ cat runme.sh
#!/bin/bash
echo "Hello World"

user@pc$ ./runme.sh
bash: ./runme.sh: Permission denied
```

Executable scripts

```
user@pc$ cat runme.sh
#!/bin/bash
echo "Hello World"

user@pc$ ./runme.sh
bash: ./runme.sh: Permission denied

user@pc$ chmod  +x ./runme.sh
user@pc$ ./runme.sh
Hello World
```

ssh (Secure Shell)

- Secure
- Encrypted
- Remote connection
  - And many MORE potentials

Two ways of authentication:

- By password
- By key usage

Connection example

```
ssh mypc.uoa.gr
```

```
The authenticity of host 'mypc.uoa.gr (10.100.52.11)' can't be established.
RSA key fingerprint is c8:03:20:79:18:0d:ea:1d:e3:1c:29:0d:0b:ce:a9:f4.
Are you sure you want to continue connecting (yes/no)?
```

Creating a pair of private & public keys

- Creating a pair of keys and storing them in ∼/.ssh/id_rsa (**private**) & ∼/.ssh/id_rsa.pub (**public**)

```
ssh-keygen -t rsa
```

Creating a pair of private & public keys

- Creating a pair of keys and storing them in ∼/.ssh/id_rsa (**private**) & ∼/.ssh/id_rsa.pub (**public**)

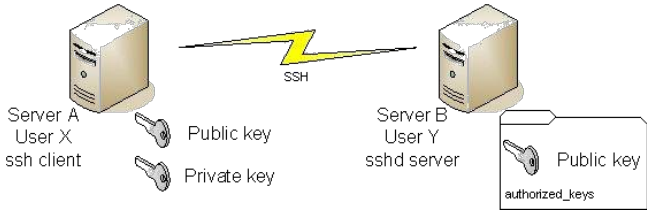
```
ssh-keygen -t rsa
```

```
Generating public/private rsa key pair.
Enter passphrase (empty for no passphrase):
Enter same passphrase again:
Your identification has been saved in ~/.ssh/id_rsa.
Your public key has been saved in ~/.ssh/id_rsa.pub.
The key fingerprint is:
ca:0f:15:49:09:2e:e9:d8:59:16:8b:8c:30:d2:b9:77 root@snf
The key's␣randomart␣image␣is:
+--[␣RSA␣2048]----+
|␣␣..o.␣␣␣␣++++␣␣␣|
|␣␣␣␣␣␣␣␣␣␣␣␣␣␣␣␣␣|
+-----------------+
```

Storing a “foreign” public key

```
cat ~/.ssh/id_rsa.pub  >> ~/.ssh/authorized_keys2
chmod 644 ~/.ssh/authorized_keys2
```

How it works?


SSH
Server A
User X
ssh client
Public key
Private key
Server B
User Y
sshd server
Public key
authorized_keys

VPS on Genomics Lab I

- Virtual private server (VPS)
  - 16 CPUs
  - 32 GB RAM
  - 100 + 2TB HDDs (with quotas)
  - IPv4
  - running Ubuntu 24.04
- suggested for executing lab exercises and final project
- connect using ssh key (~~or password ??~~)
- graphical interface via X2Go (`https://wiki.x2go.org/doku.php`)

Exercise 2 - Familiarizing with GNU/Linux CLI

- Create directory
- Rename directory
- Move directory
- Delete directory
- . . .

Submit via e-class assigment

`https://eclass.uoa.gr/modules/work/index.php?course=DI425&id=62479`

OR by email at alexdem@di.uoa.gr

`https://eclass.uoa.gr/modules/document/file.php/DI425/2025-26/exercises/ITBI2025-exercise2-ACD14102025.pdf`

DEADLINE 28/10/25

Questions?