Summer Institute in Statistical Genetics
Module 3: Computing for Statistical Genetics

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UW Biostatistics

Liége, September 2007
Introduction: Course Aims

• Introducing R
  – Manipulating datasets
  – Doing common (genetic) analyses

• Using R for sophisticated analyses
  – Doing some popular (genetic) examples
  – Knowing where to start when you need to do more
Introduction: About Prof Lumley

- Associate Prof, UW Biostat
- R Core developer
- Genetic/Genomic research in Cardiovascular Epidemiology
- Sings bass
Introduction: About Prof Rice

- Assistant Prof, UW Biostat
- Not an author, but a user (and a teacher)
- Genetic/Genomic research in Cardiovascular Epidemiology
- Sings bass

... and you?

(who are you, what area of genetics, what are you looking for from the course)
Introduction: Course structure

- 10 sessions over 2.5 days
- Day 1; Basics of R
- Day 2; Commonly-used genetic examples
- Day 2.5; More advanced topics
Introduction: Session structure

- 45 mins teaching (questions welcome)
- 30 mins hands-on
- 15 mins summary, discussion

There will also be one take-home exercise; final session will include in-depth discussion/evaluation
Introduction to R:
First steps

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What is R

R is a ‘programming environment for statistics and graphics’

The base R has fewer prepackaged statistics procedure than SPSS or SAS, but it is much easier to extend with new procedures.

There are about 1500 published extension packages for R, many aimed at genetics and genomics research.
Using R

R is a free implementation of S, for which John Chambers won the ACM Software Systems award.

For the S system, which has forever altered how people analyze, visualize, and manipulate data.

The downside is that using R effectively may require changing how you analyze, visualize, and manipulate data.

R is a command-line system, not a point-and-click system.
> 2+2
[1] 4
> 3^2
[1] 9
> 1536/317000
[1] 0.004845426
> exp(pi)-pi
[1] 19.9991
> round(qnorm(0.05/2), 2)
[1] -1.96
Reading data

• Text files

• Other statistics packages datasets

• Web pages

Much more information is in the Data Import/Export manual.
Reading text data

The easiest format has variable names in the first row

<table>
<thead>
<tr>
<th>case</th>
<th>id</th>
<th>gender</th>
<th>deg</th>
<th>yrdeg</th>
<th>field</th>
<th>startyr</th>
<th>year</th>
<th>rank</th>
<th>admin</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>1</td>
<td>F</td>
<td>Other</td>
<td>92</td>
<td>Other</td>
<td>95</td>
<td>95</td>
<td>Assist</td>
<td>0</td>
</tr>
<tr>
<td>2</td>
<td>2</td>
<td>M</td>
<td>Other</td>
<td>91</td>
<td>Other</td>
<td>94</td>
<td>94</td>
<td>Assist</td>
<td>0</td>
</tr>
<tr>
<td>3</td>
<td>2</td>
<td>M</td>
<td>Other</td>
<td>91</td>
<td>Other</td>
<td>94</td>
<td>95</td>
<td>Assist</td>
<td>0</td>
</tr>
<tr>
<td>4</td>
<td>4</td>
<td>M</td>
<td>PhD</td>
<td>96</td>
<td>Other</td>
<td>95</td>
<td>95</td>
<td>Assist</td>
<td>0</td>
</tr>
</tbody>
</table>

and fields separated by spaces. In R, use

```
salary <- read.table("salary.txt", header=TRUE)
```

to read the data from the file `salary.txt` into the data frame `salary`.  

Syntax notes

- Spaces in commands don’t matter (except for readability), but Capitalisation Does Matter.

- **TRUE** (and **FALSE**) are logical constants

- Unlike many systems, R does not distinguish between commands that do something and commands that compute a value. Everything is a function: ie returns a value.

- Arguments to functions can be named (**header=TRUE**) or unnamed (**"salary.txt"**)

- A whole data set (called a **data frame**) is stored in a variable (**salary**), so more than one dataset can be available at the same time.
Did it work?

The `head()` function shows the first few lines of the data frame

```r
> head(salary)
case id gender deg yrdeg field startyr year rank admin salary
1 1 1 F Other 92 Other 95 95 Assist 0 6684
2 2 2 M Other 91 Other 94 94 Assist 0 4743
3 3 2 M Other 91 Other 94 95 Assist 0 4881
4 4 4 M PhD 96 Other 95 95 Assist 0 4231
5 5 6 M PhD 66 Other 91 91 Full 1 11182
6 6 6 M PhD 66 Other 91 92 Full 1 11507
```

which is what we expected, so it worked.
Sometimes columns are separated by commas (or tabs)

Ozone,Solar.R,Wind,Temp,Month,Day
41,190,7.4,67,5,1
36,118,8,72,5,2
12,149,12.6,74,5,3
18,313,11.5,62,5,4
NA,NA,14.3,56,5,5

Use

ozone <- read.table("ozone.csv", header=TRUE, sep="",")

or

ozone <- read.csv("ozone.csv")
Syntax notes

- Functions can have optional arguments (\texttt{sep} wasn’t used the first time). Use \texttt{help(read.table)} for a complete description of the function and all the arguments.

- There’s more than one way to do it.

- \texttt{NA} is the code for missing data. Think of it as “Don’t Know”. R handles it sensibly in computations: eg 1+NA, NA & FALSE, NA & TRUE. You cannot test \texttt{temp==NA} (Is temperature equal to some number I don’t know?), so there is a function \texttt{is.na()}. 
Sometime the variable names aren’t included

<table>
<thead>
<tr>
<th></th>
<th>0.2</th>
<th>115</th>
<th>90</th>
<th>1</th>
<th>3</th>
<th>68</th>
<th>42</th>
<th>yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>0.7</td>
<td>193</td>
<td>90</td>
<td>3</td>
<td>1</td>
<td>61</td>
<td>48</td>
<td>yes</td>
</tr>
<tr>
<td>3</td>
<td>0.2</td>
<td>58</td>
<td>90</td>
<td>1</td>
<td>3</td>
<td>63</td>
<td>40</td>
<td>yes</td>
</tr>
<tr>
<td>4</td>
<td>0.2</td>
<td>5</td>
<td>80</td>
<td>2</td>
<td>3</td>
<td>65</td>
<td>75</td>
<td>yes</td>
</tr>
<tr>
<td>5</td>
<td>0.2</td>
<td>8.5</td>
<td>90</td>
<td>1</td>
<td>2</td>
<td>64</td>
<td>30</td>
<td>yes</td>
</tr>
</tbody>
</table>

and you have to supply them

```r
psa <- read.table("psa.txt", col.names=c("ptid","nadirpsa", "pretxpsa", "ps","bss","grade","age", "obstime","inrem"))
```

or

```r
psa <- read.table("psa.txt")
names(psa) <- c("ptid","nadirpsa","pretxpsa", "ps", "bss","grade","age","obstime","inrem")
```
Syntax notes

- Assigning a single vector (or anything else) to a variable uses the same syntax as assigning a whole data frame.

- `c()` is a function that makes a single vector from its arguments.

- `names` is a function that accesses the variable names of a data frame

- Some functions (such as `names`) can be used on the LHS of an assignment.
library(foreign)
stata <- read.dta("salary.dta")
spss <- read.spss("salary.sav", to.data.frame=TRUE)
sasxport <- read.xport("salary.xpt")
epiinfo <- read.epiinfo("salary.rec")

Notes:

- Many functions in R live in optional packages. The library() function lists packages, shows help, or loads packages from the package library.

- The foreign package is in the standard distribution. It handles import and export of data. Hundreds of extra packages are available at http://cran.r-project.org.
Files for `read.table` can live on the web

```r
fl2000<-read.table("http://faculty.washington.edu/tlumley/data/FLvote.dat", header=TRUE)
```

It’s also possible to read from more complex web services (such as the genome databases)
Operating on data

As R can have more than one data frame available you need to specify where to find a variable. The syntax `antibiotics$duration` means the variable `duration` in the data frame `antibiotics`.

```r
## This is a comment
## Convert temperature to real degrees
antibiotics$tempC <- (antibiotics$temp-32)*5/9
## display mean, quartiles of all variables
summary(antibiotics)
```
Subsets

Everything in R is a vector (but some have only one element). Use [] to extract subsets

```r
# First element
antibiotics$temp[1]
# All but first element
antibiotics$temp[-1]
# Elements 5 through 10
antibiotics$temp[5:10]
# Elements 5 and 7
antibiotics$temp[c(5,7)]
# People who received antibiotics (note ==)
antibiotics$temp[ antibiotics$antib == 1 ]
# or
with(antibiotics, temp[antib==1])
```
Notes

- Positive indices select elements, negative indices drop elements

- **5:10** is the sequence from 5 to 10

- You need `==` to test equality, not just `=`

- `with()` temporarily sets up a data frame as the default place to look up variables.
More subsets

For data frames you need two indices

```r
## First row
antibiotics[1,]
## Second column
antibiotics[,]2
## Some rows and columns
antibiotics[3:7, 2:4]
## Columns by name
antibiotics[, c("id","temp","wbc")]
## People who received antibiotics
antibiotics[antibiotics$antib==1, ]
## Put this subset into a new data frame
yes <- antibiotics[antibiotics$antib==1,]
```
Computations

mean(antibiotics$temp)
median(antibiotics$temp)
var(antibiotics$temp)
sd(antibiotics$temp)
mean(yes$temp)
mean(antibiotics$temp[antibiotics$antib==1])
with(antibiotics, mean(temp[sex==2]))
toohot <- with(antibiotics, temp>99)
mean(toohot)
Factors

Factors represent categorical variables. You can't do mathematical operations on them (except for `==`)

```r
> table(salary$rank, salary$field)

          Arts Other Prof
    Assist  668  2626    754
    Assoc   1229  4229   1071
   Full     942  6285   1984
```

```r
> antibiotics$antib <- factor(antibiotics$antib, 
    labels = c("Yes", "No"))
> antibiotics$agegp <- cut(antibiotics$age, c(0, 18, 65, 100))
> table(antibiotics$agegp)

          (0,18] (18,65] (65,100]
        2       19       4
```
Help

- `?fn` or `help(fn)` for help on `fn`

- `help.search("topic")` for help pages related to "topic"

- `apropos("tab")` for functions whose names contain "tab"

- `RSiteSearch("FDR")` to search the R Project website (requires internet access)