



Summer Institute in Statistical Genetics

Module 3: Computing for Statistical Genetics

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Seattle, June 2008

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.

A calculator

```
> 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

case	id	gender	deg	yrdeg	field	startyr	year	rank	admin
1	1	F	Other	92	Other	95	95	Assist	0
2	2	M	Other	91	Other	94	94	Assist	0
3	2	M	Other	91	Other	94	95	Assist	0
4	4	M	PhD	96	Other	95	95	Assist	0

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

```
> 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.

Reading text data

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 (`sep` wasn't used the first time). Use `help(read.table)` for a complete description of the function and all the arguments.
- There's more than one way to do it.
- `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 `temp==NA` (Is temperature equal to some number I don't know?), so there is a function `is.na()`.

Reading text data

Sometime the variable names aren't included

1	0.2	115	90	1	3	68	42	yes
2	0.7	193	90	3	1	61	48	yes
3	0.2	58	90	1	3	63	40	yes
4	0.2	5	80	2	3	65	75	yes
5	0.2	8.5	90	1	2	64	30	yes

and you have to supply them

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

or

```
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.

Other statistical packages

```
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>.

The web

Files for `read.table` can live on the web

```
f12000<-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`.

```
## 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

```
## 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

```
## 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 `==`)

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

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

```
> 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)