HuGen2071 book

Daniel E. Weeks

July 28, 2023

Table of contents

Pr	eface		9						
1	Pre	paration	10						
	1.1	Basic programming ideas	10						
		1.1.1 Introduction to Coding	10						
	1.2	R	10						
		1.2.1 PhD Training Workshop: Statistics in R	10						
	1.3	R and RStudio	10						
		1.3.1 R for the Rest of Us	10						
	1.4	GitHub	11						
	1.5	R Markdown	11						
	1.6	Unix	11						
2	Intro	oduction	12						
_									
3	_	istics	13						
	3.1	GitHub: Set up an account	13						
	3.2	GitHub Classroom	13						
4	Active Learning and Readings								
	4.1	Introduction and Overview	14						
		4.1.1 Learning Objectives	14						
		4.1.2 Readings	14						
	4.2	GitHub	14						
		4.2.1 Learning Objectives	14						
		4.2.2 Readings	14						
	4.3	R: Basics	15						
		4.3.1 Learning Objectives	15						
		4.3.2 Active Learning:	15						
		4.3.3 Readings	15						
	4.4	R: Factors, Dates, Subscripting	15						
		4.4.1 Learning Objectives	15						
		4.4.2 Active Learning:	15						
		4.4.3 Readings	16						
	4.5	R: Character Manipulation	16						
		4.5.1 Learning Objectives	16						

	4.5.2	Readings						 		16
4.6	R: Loc	ops and Flow Control						 		17
	4.6.1	Learning Objectives						 		17
	4.6.2	Active Learning:						 		17
4.7	R: Fun	and Packages, Debugging R $$.						 		17
	4.7.1	Learning Objectives						 		17
	4.7.2	Active Learning:						 		17
	4.7.3	Readings						 		17
4.8	R: Tid	$lyverse \dots \dots \dots \dots \dots$						 		17
	4.8.1	Learning Objectives						 		17
	4.8.2	Active Learning:						 		18
	4.8.3	Readings						 		18
4.9	R: Rec	coding and Reshaping Data								18
	4.9.1	Learning Objectives						 		18
	4.9.2	Active Learning:						 		18
	4.9.3	Readings						 		18
4.10	Mergin	ng Data						 		18
	4.10.1	Learning Objectives						 		18
	4.10.2	Active Learning:						 		19
	4.10.3	Readings						 		19
4.11		ditional Graphics & Advanced Graph								19
	4.11.1	Learning Objectives						 		19
	4.11.2	Active Learning:						 		19
	4.11.3	Readings						 		19
4.12	R: Exp	ploratory Data Analysis						 		19
	4.12.1	Learning Objectives						 		19
	4.12.2	Readings						 		20
4.13	R: Inte	eractive and Dynamic Graphics						 		20
	4.13.1	Learning Objectives						 		20
	4.13.2	Readings						 		20
4.14	Review	v and Help with Project						 		20
4.15	Data (Quality Checking and Filters						 		20
		Learning Objectives								20
	4.15.2	Readings						 		21
4.16	Unix:	Basics, Streams, Redirection, & Pipe						 		21
	4.16.1	Learning Objectives						 		21
		Readings								21
4.17	PLINE	K & PLINK Format						 		21
	4.17.1	Learning Objectives						 		21
		Readings								22
4.18		K Advanced								22
		Learning Objectives								22

	4.19	PLINK Computer Lab	22
			22
	4.20	Unix: Interacting with Processes, Cluster Jobs, Shell Scripting	22
		4.20.1 Learning Objectives	22
		4.20.2 Active Learning:	22
		4.20.3 Readings	23
	4.21	Unix: Data Manipulation	23
		4.21.1 Learning Objectives	23
		4.21.2 Readings	23
	4.22	Unix: Pipes & Parallelization	23
		4.22.1 Learning Objectives	23
		4.22.2 Active Learning:	23
		4.22.3 Readings	23
	4.23	Unix: Scripting, Control Structures and Variables	24
		4.23.1 Learning Objectives	24
		4.23.2 Active Learning:	24
		4.23.3 Readings	24
	4.24	Genetic Data Structures	24
		4.24.1 Learning Objectives	24
		4.24.2 Readings	24
	4.25	VCF, bcftools, vcftools	25
		4.25.1 Learning Objectives	25
	4.26	SAM & samtools	25
		4.26.1 Learning Objectives	25
		4.26.2 Readings	25
	4.27	Genetic Data in R, GDS	25
		4.27.1 Learning Objectives	25
		4.27.2 Active Learning:	25
		4.27.3 Readings	26
_			^-
5		•	27
	5.1	Question: Recycling in a dataframe	
		v G	28
		Question: Vector addition and recycling	
	5.4	Exercise 2: vector addition	29
	5.5	Exercise 3: for loops	30
	5.6	Exercise 4: while loops	32
	5.7	Exercise 5: repeat loops	33
	5.8	Exercise 6: using the rep function	34
	5.9	Exercise 7	35
	5.10	Exercise 8	36

6	R C	haracter Exercise 39	9										
	6.1	Load Libraries	9										
	6.2	Useful RStudio cheatsheet	9										
	6.3	Scenario 1	9										
	6.4	Discussion Questions	0										
		6.4.1 Question 1	0										
		6.4.2 Answer 1	2										
		6.4.3 Question 2	2										
		6.4.4 Answer 2	3										
		6.4.5 Question 3	3										
		6.4.6 Answer 3	4										
	6.5	Scenario 2	5										
		6.5.1 Question 4	5										
		6.5.2 Answer 4	6										
7	R F	unctions Excercise 48	8										
	7.1	Load Libraries	8										
	7.2	Location	8										
	7.3	Data set creation code	8										
	7.4	Example	9										
		7.4.1 Question: How could we construct a list of file names? 50	0										
		7.4.2 Question: Outline a possible algorithm	0										
		7.4.3 Question: Construct a more detailed step-by-step algorithm 51	1										
		7.4.4 Task: Write a read_data_file function	1										
		7.4.5 Question: What does the above code assume?	2										
		7.4.6 Question: Extend your function to process all of the files	3										
		7.4.7 Bonus question	3										
8	R Tidyverse Exercise 55												
	8.1	Load Libraries	5										
	8.2	Untidy data	5										
	8.3	Tidy data	6										
	8.4	Gather	7										
	8.5	Pivot_longer	8										
	8.6	WHO TB data	8										
	8.7	Conclusion	0										
	8.8	Acknowledgment	1										
9	R R	ecoding Reshaping Exercise 62	2										
	9.1	Load Libraries	2										
	9.2	Project 1 Data	2										
	9.3	Exercise 1	3										
	9.4	Checking for duplicates	5										

	9.5	Counting the number of occurences of the ID	35
	9.6	Count sample_id duplicates	66
	9.7	Checking for duplicates	66
			67
			67
		9.7.3 Subject ID	38
	9.8	Exercise 2	38
		9.8.1 Comment	39
	9.9		39
		9.9.1 Comment:	70
		9.9.2 xtabs table with labels	70
	9.10	Exercise 4	72
			73
			74
		9.12.1 Comment	74
	9.13		75
			75
10	R M	erging Exercise 7	7
	10.1	Load Libraries	77
	10.2	Input data	77
	10.3	Select a subset of subject-level fields	77
	10.4	Unique records	78
		10.4.1 Comment	79
	10.5	Check that the subject_id's are now not duplicated	30
	10.6	Create random integer IDs	31
	10.7	Merge in new phenotype information	31
	10.8	Always be careful when merging	33
	10.9	Merge in new phenotype information	33
11		· F	37
			37
			37
			38
		v 1 v	39
	11.5	<u>o</u>	90
)2
	11.6	Always plot your data	93
	11.7	Identical box plots)4
		1)4
	11.9	Non-identical violin plots	95
	11.10	OSina plots	95
	11 11	Sina plots	96

	11.12S	Sina plots	96
	11.13S	Sina plots	97
	11.14I	Orawing multiple graphs	97
	11.15V	Vriting ggplot functions	98
	11.16E	Exercise 3	.01
	1	1.16.1 Exercise	.01
	11.17S	Source of data	.04
12		O Company of the comp	05
		Load Libraries	
		Create some example data	
		Task: Reorder rows in dd in the order of ds's columns	
		Assumption Check Question	
		Task: Reorder rows in \mathtt{dd} to match the order of the columns in \mathtt{ds}	
		Question: use arrange?	
		Question: use arrange?	
	12.8 (Question: use slice	.09
	12.9	Question: use select?	.10
	12.100	Question: use row names	.11
12	D F	Lauretoni Data Analysia Francisca	12
13		loratory Data Analysis Exercise Load Libraries	
		Explore Project 1 data	
		Dimensions	
		Dimensions	
		3.4.1 Data ds	
		3.4.2 Data dictionay dd	
		Arrangement	
		3.5.1 Samples or subjects	
		3.5.2 Unique values	
		3.5.3 Subject-level data set	
		Coding	
		3.6.1 Recode for understandability	
		Missing data	
	13.8 I	Distribution	
	13.9 V	Variation \ldots	.20
	1	3.9.1 Bar plots	21
		*	21
	1	3.9.3 QQ plots	23
		3.9.4 Correlation	
	1	3.9.5 ggpairs from the GGally R package	24
	13 100	1staFynlorer	26

14	Basi	c Shell Commands	127
	14.1	Acknowledgment and License	127
		Shell Basics:	
	14.3	Creating Things:	127
		14.3.1 How to create new files and directories	127
		14.3.2 How to delete files and directories	128
		14.3.3 How to copy and rename files and directories	128
	14.4	Pipes and Filters	128
		14.4.1 How to use wildcards to match filenames	128
		14.4.2 How to redirect to a file and get input from a file	129
	14.5	How to repeat operations using a loop	130
		14.5.1 For loop	
		14.5.2 While Loop	131
	14.6	Finding Things	132
		14.6.1 How to select lines matching patterns in text files	132
		14.6.2 How to find files with certain properties	132
15	Sum	mary	134
16	Web	R - R in the web browser	135
17	Tech	nical Details	136
	17.1	Quarto	136
		17.1.1 Callout blocks	
		17.1.2 Adding a chapter	137
	17.2	Previewing the book	137
		Deploying the book to GitHub Pages	
		Deploying the book to Netlify	
		WebR: R in the browser	
Po:	foron	cos	130

Preface

This is a Quarto book created from markdown and executable code using Quarto within RStudio.

Book web site: https://danieleweeks.github.io/HuGen2071/

Book source code: https://github.com/DanielEWeeks/HuGen2071

Created by Daniel E. Weeks

Website: https://www.sph.pitt.edu/directory/daniel-weeks

To learn more about Quarto books visit https://quarto.org/docs/books/.

1 Preparation

The first part of our HuGen 2071 course aims to teach you R in the context of applied data wrangling in a genetic context. In our experience, if you have never programmed much before, it moves kind of fast. As such, it would be useful to review these sources below.

1.1 Basic programming ideas

1.1.1 Introduction to Coding

This web page and two short videos discusses how computer programming is very similar to writing a recipe - you have to break a complex project down into precise smaller individual steps.

https://subjectguides.york.ac.uk/coding/introduction

1.2 R

1.2.1 PhD Training Workshop: Statistics in R

This online book has a nice introduction to the concepts of programming, RStudio, and R https://bookdown.org/animestina/R_Manchester/

See Chapters 1, 2, and 3

1.3 R and RStudio

1.3.1 R for the Rest of Us

Acquaint or refresh yourself with R and RStudio — including installing them on your computer with this "R for the Rest of Us course" (24 min of videos + exercises):

https://rfortherestofus.com/courses/getting-started/

Slides: https://rfortherestofus.github.io/getting-started/slides/slides.html

1.4 GitHub

To introduce yourself to GitHub:

https://docs.github.com/en/get-started/using-git/about-git

https://docs.github.com/en/get-started/quickstart/hello-world

1.5 R Markdown

To introduce yourself or refresh yourself on R Markdown:

https://rmarkdown.rstudio.com/

Scroll down and click on "Get Started", which will take you to Lesson 1:

https://rmarkdown.rstudio.com/lesson-1.html

1.6 Unix

And finally, to introduce yourself or refresh yourself with Unix (well, Linux in this case, but close enough), try Lessons 1–11 here:

https://www.webminal.org/

2 Introduction

This is a book created from markdown and executable code using Quarto within RStudio.

Book web site: https://danieleweeks.github.io/HuGen2071/

Book source code: https://github.com/DanielEWeeks/HuGen2071

Created by Daniel E. Weeks

Website: https://www.sph.pitt.edu/directory/daniel-weeks

3 Logistics

3.1 GitHub: Set up an account

Please go to https://github.com and set up a GitHub account.

Choose your GitHub user name carefully, as you may end up using it later in a professional context.

3.2 GitHub Classroom

As GitHub Classroom will be used to distribute course materials and to submit assignments, it would be best if you get git working on your own computer. The easiest way to do this is to install RStudio, R, and git on your computer.

Please follow the detailed instructions in https://github.com/jfiksel/github-classroom-for-students

In particular, see Step 5 re generating an ssh key so you don't need to login every time.

4 Active Learning and Readings

4.1 Introduction and Overview

4.1.1 Learning Objectives

- Review the syllabus
- Describe bioinformatics
- List various type of data used in genetics

4.1.2 Readings

Barnes (2007) Chapter 1 Carey MA, Papin JA. Ten simple rules for biologists learning to program. PLoS Comput Biol. 2018;14(1):e1005871. https://doi.org/10.1371/journal.pcbi.1005871

Dudley JT, Butte AJ. A quick guide for developing effective bioinformatics programming skills. PLoS Comput Biol. 2009;5(12):e1000589. https://doi.org/10.1371/journal.pcbi.1000589

4.2 GitHub

4.2.1 Learning Objectives

- To learn how to use GitHub
- To learn how to use GitHub Classroom
- To learn how to use GitHub within RStudio

4.2.2 Readings

Happy Git and GitHub for the useR. https://happygitwithr.com/

Perez-Riverol Y, Gatto L, Wang R, et al. Ten Simple Rules for Taking Advantage of Git and GitHub. PLoS Comput Biol. 2016;12(7):e1004947. https://doi.org/10.1371/journal.pcbi.1004947

4.3 R: Basics

4.3.1 Learning Objectives

- To become familiar with the R language and concepts
- To learn how to read and write data with R
- To learn control flow: choices and loops

4.3.2 Active Learning:

https://datacarpentry.org/R-genomics/01-intro-to-R.html

4.3.3 Readings

Buffalo (2015) Chapter 8 'R Language Basics' (Available online through PittCat+)

Read the first four sections, up to the end of 'Vectors, Vectorization, and Indexing'

 $https://pitt.primo.exlibrisgroup.com/permalink/01PITT_INST/i25aoe/cdi_askewsholts_v~lebooks~9781449367510$

https://datacarpentry.org/R-genomics/01-intro-to-R.html

Supplementary Reading: Spector (2008) Chapters 1 & 2 (Available online through PittCat+; link in syllabus)

4.4 R: Factors, Dates, Subscripting

4.4.1 Learning Objectives

- To learn how to handle factors and dates with R
- To learn how to subset data with R
- To learn how to manipulate characters with R

4.4.2 Active Learning:

https://datacarpentry.org/R-ecology-lesson/02-starting-with-data.html

Subsetting: https://swcarpentry.github.io/r-novice-gapminder/06-data-subsetting.html

4.4.3 Readings

Buffalo (2015) Chapter 8 'R Language Basics' (Available online through PittCat+)

Read the 'Factors and classes in R' subsection at the end of the 'Vectors, Vectorization, and Indexing' section.

Read the 'Exploring Data Through Slicing and Dicing: Subsetting Dataframes' section.

Read the 'Working with Strings' section.

 $https://pitt.primo.exlibrisgroup.com/permalink/01PITT_INST/i25aoe/cdi_askewsholts_v~lebooks_9781449367510$

https://datacarpentry.org/R-ecology-lesson/02-starting-with-data.html

Supplementary Readings: Spector (2008) Chapters 4, 5, 6

4.5 R: Character Manipulation

4.5.1 Learning Objectives

- To learn how to handle character data in R
- To learn how to use regular expressions in R

4.5.2 Readings

Buffalo (2015) Chapter 8 'R Language Basics' (Available online through PittCat+)

Read the 'Working with Strings' section (Oh, you already read this)

 $https://pitt.primo.exlibrisgroup.com/permalink/01PITT_INST/i25aoe/cdi_askewsholts_v~lebooks_9781449367510$

Read Chapter 14 "Strings" of "R for Data Science": https://r4ds.had.co.nz/strings.html

Supplementary Reading: Spector (2008) Chapter 7

4.6 R: Loops and Flow Control

4.6.1 Learning Objectives

- To learn how to implement loops in R
- To learn how to control flow in R
- To learn how to vectorize operations

4.6.2 Active Learning:

Flow control and loops: https://swcarpentry.github.io/r-novice-gapminder/07-control-flow.html

Vectorization: https://swcarpentry.github.io/r-novice-gapminder/09-vectorization.html

4.7 R: Functions and Packages, Debugging R

4.7.1 Learning Objectives

- To learn how to write R functions and packages
- To learn how to debug R code

4.7.2 Active Learning:

https://swcarpentry.github.io/r-novice-gapminder/10-functions.html

4.7.3 Readings

Functions Explained: https://swcarpentry.github.io/r-novice-gapminder/10-functions.html

Buffalo (2015) Chapter 8: Read the section 'Digression: Debugging R Code'

4.8 R: Tidyverse

4.8.1 Learning Objectives

- To learn how to use the pipe operator
- To learn how to use Tidyverse functions

4.8.2 Active Learning:

https://uomresearchit.github.io/r-day-workshop/04-dplyr/

4.8.3 Readings

Introduction to the Tidyverse: Manipulating tibbles with dplyr https://uomresearchit.github.io/r-day-workshop/04-dplyr/

Supplementary Reading: Buffalo (2015) Chapter 8: section 'Exploring Dataframes with dplyr'

4.9 R: Recoding and Reshaping Data

4.9.1 Learning Objectives

• To learn how to reformat and reshape data in R

4.9.2 Active Learning:

Recoding data: Pay particular attention to the Recoding values and Creating new variables sections

https://librarycarpentry.org/lc-r/03-data-cleaning-and-transformation.html

4.9.3 Readings

Reshaping data https://sscc.wisc.edu/sscc/pubs/dwr/reshape-tidy.html

Supplementary Reading: Spector (2008) Chapters 8 & 9

4.10 Merging Data

4.10.1 Learning Objectives

- To learn how to use the R 'merge' command
- To learn how to use the R Tidyverse join commands

4.10.2 Active Learning:

https://mikoontz.github.io/data-carpentry-week/lesson_joins.html

4.10.3 Readings

https://mikoontz.github.io/data-carpentry-week/lesson_joins.html#practice_with_joins_u sing_gapminder

Supplementary Reading: Buffalo (2015) Chapter 8 'Merging and Combining Data'. Spector (2008) Chapter 9.

4.11 R: Traditional Graphics & Advanced Graphics

4.11.1 Learning Objectives

- To learn the basic graphics commands of R
- To learn the R graphing package ggplot2

4.11.2 Active Learning:

https://datacarpentry.org/R-ecology-lesson/04-visualization-ggplot2.html

4.11.3 Readings

Plotting with ggplot2 https://datacarpentry.org/R-ecology-lesson/04-visualization-ggplot2.html

Supplementary Reading: Wickham (2009) Chapters 2 & 3

4.12 R: Exploratory Data Analysis

4.12.1 Learning Objectives

- To learn how to summarize data frames
- To learn how to visualize missing data patterns
- To learn how to visualize covariation

4.12.2 Readings

Missing value visualization with tidyverse in R https://towardsdatascience.com/missing-value-visualization-with-tidyverse-in-r-a9b0fefd2246

Suggested Reading: Buffalo (2015) Chapter 8 Sections: Exploring Data Visually with ggplot2 I: Scatterplots and Densities Exploring Data Visually with ggplot2 II: Smoothing Binning Data with cut() and Bar Plots with ggplot2 Using ggplot2 Facets.

4.13 R: Interactive and Dynamic Graphics

4.13.1 Learning Objectives

- To learn how to use interactive and dynamic graphics to explore your data more thoroughly
- To learn to use iPlots and Ggobi
- To learn to use plotly

4.13.2 Readings

Create interactive ggplot2 graphs with plotly https://www.littlemissdata.com/blog/interactiveplots

Suggested Reading: Wickham (2009) Chapters 2 & 3

4.14 Review and Help with Project

4.15 Data Quality Checking and Filters

4.15.1 Learning Objectives

• To learn how to check genotype data for quality

4.15.2 Readings

Anderson CA, Pettersson FH, Clarke GM, Cardon LR, Morris AP, Zondervan KT. Data quality control in genetic case-control association studies. Nat Protoc. 2010 Sep;5(9):1564–1573. DOI: https://doi.org/10.1038/nprot.2010.116

Suggested Reading: Laurie CC, Doheny KF, Mirel DB, Pugh EW, Bierut LJ, Bhangale T, Boehm F, Caporaso NE, Cornelis MC, Edenberg HJ, Gabriel SB, Harris EL, Hu FB, Jacobs KB, Kraft P, Landi MT, Lumley T, Manolio TA, McHugh C, Painter I, Paschall J, Rice JP, Rice KM, Zheng X, Weir BS, GENEVA Investigators. Quality control and quality assurance in genotypic data for genome-wide association studies. Genetic epidemiology. 2010 Sep;34(6):591–602. PMID: 20718045 DOI: https://doi.org/10.1002/gepi.20516

4.16 Unix: Basics, Streams, Redirection, & Pipe

4.16.1 Learning Objectives

- To learn basic Unix commands
- To learn how streams operate in Unix
- To learn out to pass streamed data from program to program in Unix

4.16.2 Readings

Buffalo (2015) Chapter 3

"Chapter 43: Redirecting Input and Output" in Unix Power Tools, 3rd Edition by Jerry Peek, Shelley Powers, Tim O'Reilly, Mike Loukides. Published by O'Reilly Media, Inc. https://pitt.primo.exlibrisgroup.com/permalink/01PITT_INST/e8h8hp/alma9998520758606236

Terminus, a web-based game for learning and practicing basic UNIX commands https://web.mit.edu/mprat/Public/web/Terminus/Web/main.html

4.17 PLINK & PLINK Format

4.17.1 Learning Objectives

- Describe PLINK formats
- Create PLINK datafiles
- Use PLINK to perform genetic association testing

4.17.2 Readings

Marees AT, de Kluiver H, Stringer S, Vorspan F, Curis E, Marie-Claire C, Derks EM. A tutorial on conducting genome-wide association studies: Quality control and statistical analysis. Int J Methods Psychiatr Res. 2018 Jun;27(2):e1608. PMID: 29484742 PMCID: PMC6001694 DOI: https://doi.org/10.1002/mpr.1608

https://github.com/MareesAT/GWA_tutorial/

4.18 PLINK Advanced

4.18.1 Learning Objectives

• To learn how to use PLINK to manipulate data files

4.19 PLINK Computer Lab

4.19.1 Learning Objectives

• To practice using PLINK to manipulate data files

4.20 Unix: Interacting with Processes, Cluster Jobs, Shell Scripting

4.20.1 Learning Objectives

- To learn how to interact with running processes
- To learn about the cluster and how to submit jobs there
- To learn how to write a script that can run in Unix

4.20.2 Active Learning:

Software Carpentry Unix Shell intro parts 1-3 https://swcarpentry.github.io/shell-novice/

4.20.3 Readings

Buffalo (2015) Chapter 7 up to the start of "Sorting Plain-Text Data with Sort" section.

Suggested Reading: Software Carpentry Unix Shell intro parts 1-3 (https://swcarpentry.github.io/shell-novice/)

4.21 Unix: Data Manipulation

4.21.1 Learning Objectives

• To learn Unix tools like sed and awk that can be used to manipulate data

4.21.2 Readings

Buffalo (2015) Chapter 7 from the "Sorting Plain-Text Data with Sort" section on.

4.22 Unix: Pipes & Parallelization

4.22.1 Learning Objectives

- To learn to string programs together to process data
- To learn how to parallelize functions in Unix

4.22.2 Active Learning:

Software Carpentry Unix Shell intro part 4 https://swcarpentry.github.io/shell-novice/04-pipefilter.html

4.22.3 Readings

Buffalo (2015) Chapter 12: "Bioinformatics Shell Scripting, Writing Pipelines, and Parallelizing Tasks"

4.23 Unix: Scripting, Control Structures and Variables

4.23.1 Learning Objectives

- To learn how to use control structures in Unix scripting
- To learning how to use variables in Unix

4.23.2 Active Learning:

Software Carpentry Unix Shell intro parts 5-7 https://swcarpentry.github.io/shell-novice/

4.23.3 Readings

Software Carpentry Unix Shell intro parts 5-7 (https://swcarpentry.github.io/shell-novice/)

4.24 Genetic Data Structures

4.24.1 Learning Objectives

• To learn about what genetic data is stored and principles for storing it

4.24.2 Readings

Introduction to PLINK (22n14-rlm-Introduction_to_PLINK.pdf, included in this lecture's folder)

Bennett RL, Steinhaus KA, Uhrich SB, O'Sullivan CK, Resta RG, Lochner-Doyle D, Markel DS, Vincent V, Hamanishi J. Recommendations for standardized human pedigree nomenclature. J Genet Couns. 1995 Dec;4(4):267-79. https://doi.org/10.1007/BF01408073. PMID: 24234481.

Bennett RL, French KS, Resta RG, Doyle DL. Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. J Genet Couns. 2008 Oct;17(5):424-33. https://doi.org/10.1007/s10897-008-9169-9. Epub 2008 Sep 16. PMID: 18792771.

Bennett RL, French KS, Resta RG, Austin J. Practice resource-focused revision: Standardized pedigree nomenclature update centered on sex and gender inclusivity: A practice resource of the National Society of Genetic Counselors. J Genet Couns. 2022 Sep 15. https://doi.org/10.1002/jgc4.1621. Epub ahead of print. PMID: 36106433.

4.25 VCF, bcftools, vcftools

4.25.1 Learning Objectives

- To learn about VCF data format
- To learn about beftools and veftools for manipulating VCF files

4.26 SAM & samtools

4.26.1 Learning Objectives

- To learn about SAM data format for sequence data
- To learn about samtools to manipulate SAM data files

4.26.2 Readings

Buffalo Chapter 11 "Working with Alignment Data"

Data Wrangling and Processing for Genomics https://data-lessons.github.io/wrangling-genomics/

Relevant links: The Sequence Alignment/Map Format Specification http://samtools.github.io/hts-specs/

4.27 Genetic Data in R, GDS

4.27.1 Learning Objectives

- To learn about data structures in R for storing genetic data
- To learn about the GDS format

4.27.2 Active Learning:

 $https://uw-gac.github.io/topmed_workshop_2017/gds-format.html~(Only~2.1-Exploring~a~GDS~file)$

4.27.3 Readings

Zheng X, Gogarten SM, Lawrence M, Stilp A, Conomos MP, Weir BS, Laurie C, Levine D. SeqArray-a storage-efficient high-performance data format for WGS variant calls. Bioinformatics. 2017 Aug 1;33(15):2251-2257. doi: 10.1093/bioinformatics/btx145. PMID: 28334390; PMCID: PMC5860110. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5860110/

5 R Basics Group Exercise

5.1 Question: Recycling in a dataframe

Suppose you have a dataframe df with three columns, A, B, and C, as follows:

```
df <- data.frame(
    A = c(1, 2, 3, 4),
    B = c(5, 6, 7, 8),
    C = c(9, 10, 11, 12)
    )
    df

A B C
1 1 5 9
2 2 6 10
3 3 7 11</pre>
```

Now, you want to insert a shorter vector D into the df dataframe:

```
df$D <- c(13,14)
```

4 4 8 12

What will be the D column of df after the operation?

- (A) c(13, 14, NA, NA)
- (B) c(13, 14, 11, 12)
- (C) c(13, 14, 13, 14)
- (D) c(13, 14)

Please select the correct option.

5.2 Exercise 1: recycling

This exercise should help answer this question: 'In what type of situations would "recycling" be useful?

First, let's set up the data frame a

```
a \leftarrow data.frame(n = 1:4)
  dim(a)
[1] 4 1
  а
  n
1 1
2 2
```

Use recycling to insert into the data frame a a column named rowNum1 that contains a 1 in even rows and a 2 in odd rows.



⚠ Warning

If the following WebR chunk is working properly, you should see an editor window below the Run code tab displaying this line of R code: (a <- data.frame(n = 1:4)).

```
(a \leftarrow data.frame(n = 1:4))
# Edit/add R code here
```



The R command

a\$rowNum1 <- NA

would insert a new row into the data frame a full of NA values.

```
Expand to see the answer

a$rowNum1 <- c(1,2)
a

n rowNum1
1 1 1
2 2 2 2
3 3 1
4 4 2</pre>
```

5.3 Question: Vector addition and recycling

Suppose you have two vectors in R:

Vector A: c(1, 2, 3)Vector B: c(4, 5)

If you perform the operation A + B, what will be the result of vector recycling?

- (A) c(5, 7, 3)
- (B) c(5, 7, 8)
- (C) c(5, 7, 7)
- (D) c(5, 5, 3)

Please select the correct option.

5.4 Exercise 2: vector addition

Use vector addition to construct a vector of length 4 that contains a 1 in even positions and a 2 in odd positions. Then insert this vector into the data frame a into a column named rowNum6.

Edit/add code here

```
Tip
What vector could you add to this vector so the sum is the vector (1, 2, 1, 2)?
rep(1, 4)
[1] 1 1 1 1
```

```
? Expand to see the answer
  r1 < -rep(1, times = 4)
  r2 \leftarrow rep(c(0,1), times = 2)
  r1
[1] 1 1 1 1
  r2
[1] 0 1 0 1
  r1 + r2
[1] 1 2 1 2
  a$rowNum6 <- r1 + r2
  n rowNum1 rowNum6
1 1
           1
                   1
2 2
           2
                   2
3 3
           1
                   1
4 4
           2
                   2
```

5.5 Exercise 3: for loops

Loops allow you to repeat actions on each item from a vector of items.

Here is an example for loop, iterating through the values of i from 1 to 3:

```
for (i in 1:3) {
   print(paste("i =",i))
}

[1] "i = 1"
[1] "i = 2"
[1] "i = 3"
```

This does the same thing as this repetitive code:

```
i.vector <- c(1,2,3)
i <- i.vector[1]
print(paste("i =",i))

[1] "i = 1"

   i <- i.vector[2]
   print(paste("i =",i))

[1] "i = 2"

   i <- i.vector[3]
   print(paste("i =",i))</pre>
```

Use a for loop to insert into the data frame a a column named rowNum2 that contains a 1 in even rows and a 2 in odd rows.

Edit/add code here



Think about how as i increments from 1 to nrow(a), how could we map that sequence (e.g. 1, 2, 3, 4) to the desired sequence of 1, 2, 1, 2.

```
? Expand to see the answer
  # Set value that we want to iterate 1, 2, 1, 2, ...
  j <- 1
  # Initialize rowNum2 to all missing values
  a$rowNum2 <- NA
  # Start the for loop, looping over the number of rows in a
  for (i in c(1:nrow(a))) {
     # Assign value j to row i
     a$rowNum2[i] <- j
     # Increment j
     j <- j + 1
     # If j is greater than 2, set it back to 1
     if (j > 2) {
       j <- 1
     }
  }
  a
 n rowNum1 rowNum6 rowNum2
1 1
          1
                  1
                           1
2 2
          2
                  2
                          2
3 3
          1
                  1
                           1
          2
                           2
4 4
                  2
```

5.6 Exercise 4: while loops

Here's an example while loop:

```
i <- 1
while (i < 4) {
  print(paste("i =",i))
  i <- i + 1
}

[1] "i = 1"
[1] "i = 2"
[1] "i = 3"</pre>
```

Use a while loop to insert into the data frame a a column named rowNum3 that contains a 1 in even rows and a 2 in odd rows.

Edit/add code here

```
Expand to see the answer
  a$rowNum3 = NA
  i <- 1 #set index
  while(i <= nrow(a)){ #set conditions for while loop</pre>
    if ((i \% 2)) { #if statement for when "i" is odd
      a$rowNum3[i] <- 1
    }
    else #else statement for when "i" is even
      a$rowNum3[i] <- 2
    i <- i + 1 #counter for "i", increments by 1 with each loop iteration
  }
  a
  n rowNum1 rowNum6 rowNum2 rowNum3
                  1
                                   2
          2
                  2
                          2
3 3
          1
                  1
                          1
                                   1
4 4
          2
                  2
                           2
                                   2
```

5.7 Exercise 5: repeat loops

Here's an example repeat loop:

```
i <- 1
repeat {
   print(paste("i =",i))
   i <- i + 1
   if (i > 3) break
}
```

```
[1] "i = 1"
```

```
[1] "i = 2"
[1] "i = 3"
```

Use a repeat loop to insert into the data frame a a column named rowNum4 that contains a 1 in even rows and a 2 in odd rows.

Edit/add code here

```
Expand to see the answer
  a$rowNum4 <- NA
  i <- 1 #set index
  repeat {
    if ((i \% 2)) { #if statement for when "i" is odd
      a$rowNum4[i] <- 1
    else #else statement for when "i" is even
      a$rowNum4[i] <- 2
    i <- i + 1 #counter for "i", increments by 1 with each loop iteration
    if (i > nrow(a)) {
      break
    }
  }
 n rowNum1 rowNum6 rowNum2 rowNum3 rowNum4
1 1
                 1
                         1
                                  1
          2
                  2
                          2
                                  2
                                          2
2 2
3 3
                                          1
          1
                  1
                          1
                                  1
```

5.8 Exercise 6: using the rep function

Use the rep command to insert into the data frame a a column named rowNum5 that contains a 1 in even rows and a 2 in odd rows.

```
# Edit/add code here
```

```
? Expand to see the answer
  # This will only work correctly if nrow(a) is even
  a$rowNum5 <- rep(c(1,2), nrow(a)/2)
  n rowNum1 rowNum6 rowNum2 rowNum3 rowNum4 rowNum5
1 1
                  1
                           1
                                   1
                                                    2
2 2
          2
                  2
                           2
                                            2
3 3
          1
                  1
                           1
                                   1
                                            1
                                                    1
4 4
          2
                   2
                           2
                                            2
                                                    2
```

5.9 Exercise 7

List all even rows of the data frame a.

List rows 3 and 4 of the data frame a.

Edit/add code here

```
Expand to see the answer
  # All even rows
  a[a$rowNum1==2,]
  n rowNum1 rowNum6 rowNum2 rowNum3 rowNum4 rowNum5
          2
                  2
                           2
                                   2
          2
                  2
                           2
                                   2
                                            2
                                                    2
  # All odd rows
  a[a$rowNum1==1,]
  n rowNum1 rowNum6 rowNum2 rowNum3 rowNum4 rowNum5
1 1
          1
                  1
                           1
                                   1
                                            1
                                                    1
3 3
          1
                  1
                           1
                                   1
                                            1
                                                    1
```

5.10 Exercise 8

Note

Learning objective: Learn how to alter the options of an R command to achieve your goals.

This exercise should help answer this question: "When reading a file, will missing data be automatically represented as NA values, or does that need to be coded/manually curated?"

The tab-delimited file in testdata.txt contains the following data:

```
1 1 1
2 2 2
3 NA 99
4 4 4
```

Your collaborator who gave you these data informed you that in this file 99 stands for a missing value, as does NA.

However if we use the read.table command with its default options to read this in, we fail to accomplish the desired task, as 99 is not reading as a missing value:

```
infile <- "data/testdata.txt"</pre>
  # Adjust the read.table options to read the file correctly as desired.
  b <- read.table(infile)</pre>
  V1 V2 V3
1
  1
      1
         1
2
      2
  3 NA 99
      4
  str(b)
                 4 obs. of 3 variables:
'data.frame':
            1 2 3 4
$ V1: int
$ V2: int
            1 2 NA 4
$ V3: int 1 2 99 4
```

Use the read.table command to read this file in while automatically setting both the 'NA" and the 99 to NA. This can be done by adjusting the various options of the read.table command.

```
dir.create("data")
infile <- "data/testdata.txt"
srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/data/testdata.tx
download.file(srcfile, infile)
# Adjust the read.table options to read the file correctly as desired.
b <- read.table(infile)
b</pre>
```

Tip

Read the help page for the read.table command

? Expand to see the answer

To read this in properly, we have to let 'read.table' know that there is no header and that which values should be mapped to the missing NA value:

```
b <- read.table(infile, header = FALSE, na.strings = c("NA","99"))
  b
 V1 V2 V3
    2 2
  3 NA NA
     4 4
  str(b)
'data.frame':
                4 obs. of 3 variables:
$ V1: int 1 2 3 4
$ V2: int 1 2 NA 4
$ V3: int 1 2 NA 4
  summary(b)
       V1
                                       VЗ
        :1.00
                       :1.000
                                 {\tt Min.}
                                        :1.000
Min.
                Min.
```

```
1st Qu.:1.75
              1st Qu.:1.500
                             1st Qu.:1.500
Median :2.50
              Median :2.000
                             Median :2.000
Mean :2.50
              Mean :2.333
                              Mean
                                   :2.333
3rd Qu.:3.25
                              3rd Qu.:3.000
              3rd Qu.:3.000
Max. :4.00
              Max.
                     :4.000
                              Max.
                                    :4.000
              NA's
                              NA's
                     :1
                                    :1
```

6 R Character Exercise

6.1 Load Libraries

```
library(tidyverse)
# library(tidylog)
library(knitr)
```

6.2 Useful RStudio cheatsheet

See the "String manipulation with stringr cheatsheet" at https://www.rstudio.com/resources/cheatsheets/

6.3 Scenario 1

You are working with three different sets of collaborators: 1) the clinical group that did the field work and generated the anthropometric measurements; 2) the medical laboratory that measured blood pressure in a controlled environment; and 3) the molecular laboratory that generated the genotypes.

```
clin <- read.table(file = "data/clinical_data.txt", header=TRUE)
kable(clin)</pre>
```

ID	height
1	152
104	172
2112	180
2543	163

```
lab <- read.table(file = "data/lab_data.txt", header = TRUE)
kable(lab)</pre>
```

ID	SBP
SG0001	120
SG0104	111
SG2112	125
SG2543	119

```
geno <- read.table(file = "data/genotype_data.txt", header = TRUE)
kable(geno)</pre>
```

rs1212
G/C
G/G
C/C
C/G

6.4 Discussion Questions

6.4.1 Question 1

The clinical group, which measured height, used integer IDs, but the medical group, which measured the blood pressure, decided to prefix the integer IDs with the string 'SG' (so as to distinguish them from other studies that were also using integer IDs). So ID '1' was mapped to ID 'SG0001'.

Table 6.4: The clin data frame

ID	height
1	152
104	172
2112	180
2543	163

Discuss how, using R commands, you would reformat the integer IDs to be in the format

"SGXXXX". Write down your ideas in the next section, and, if you have time, try them out within an R chunk.

Hint: Use the formatC function.

6.4.1.1 Interactive WebR chunk

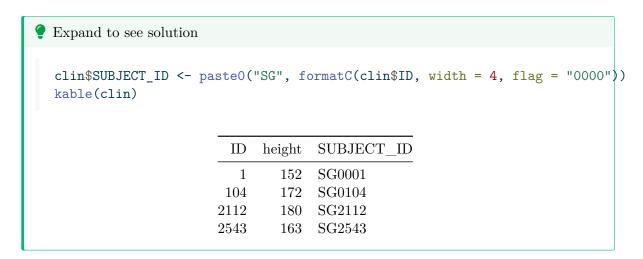
You can interactively run R within this WebR chunk by clicking the Run code tab. Note that this is a limited version of R which runs within your web browser.

Note

This Run code WebR chunk needs to be run first, before the later ones, as it downloads and reads in the required data files. The WebR chunks should be run in order, as you encounter them, from beginning to end.

```
# Download files within the WebR environment
dir.create("data")
infiles <- c("data/clinical_data.txt","data/lab_data.txt","data/genotype_data.txt")</pre>
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"</pre>
for (i in 1:length(infiles)) {
   download.file(paste0(root_srcfile,infiles[i]), infiles[i])
}
# kable is not available in WebR
kable <- head
# Read the three files in:
clin <- read.table(file = "data/clinical data.txt", header=TRUE)</pre>
kable(clin)
lab <- read.table(file = "data/lab_data.txt", header = TRUE)</pre>
kable(lab)
geno <- read.table(file = "data/genotype_data.txt", header = TRUE)</pre>
kable(geno)
# Edit/add R code here
```

6.4.2 Answer 1



6.4.3 Question 2

Discuss how, using R commands, you would reformat the "SGXXXX" IDs to be integer IDs. Write down your ideas in the next section, and, if you have time, try them out within an R chunk.

Table 6.6: The lab data frame

ID	SBP
SG0001	120
SG0104	111
SG2112	125
SG2543	119

Hint: Use either the gsub command or the str_replace_all command from the stringr package.

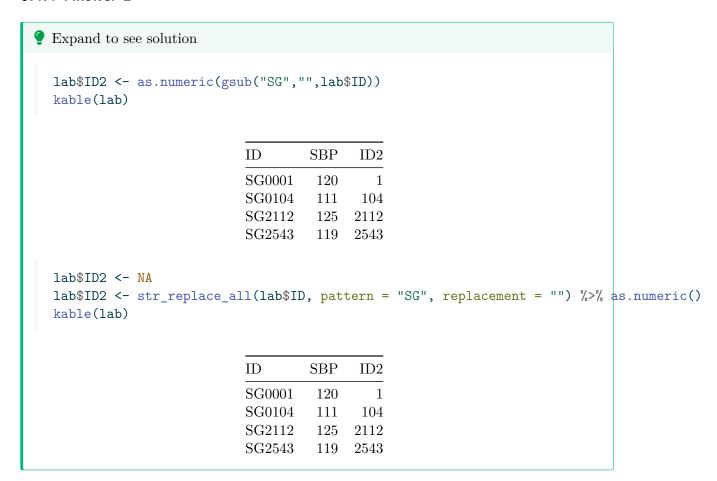


Warning

To read in and load the data within the WebR environment, be sure to run all of the WebR chunks in order. For example, to usefully run R code in this WebR chunk here, you first need to run the WebR chunk above in Question 1.

```
# str_replace_all is in the stringr R package
library(stringr)
# Edit/add code here
```

6.4.4 Answer 2



6.4.5 Question 3

The genotype group used IDs in the style "TaqMan-SG0001-190601", where the first string is "TaqMan" and the ending string is the date of the genotyping experiment.

Discuss how, using R commands, you would extract an "SGXXXX" style ID from the "TaqMan-SG0001-190601" style IDs. Write down your ideas in the next section, and, if you have time, try them out within an R chunk.

Note that one of the IDs has a lower case 'g' in it - how would you correct this, using R commands?

Table 6.9: The geno data frame

rs1212
G/C
G/G
C/C
C/G

Hint: Use either the str_split_fixed function from the stringr package or the separate function from the tidyr package.

```
# separate is in the tidyr R package
library(tidyr)
# Edit/add code here
```

6.4.6 Answer 3

```
Expand to see solution
  a <- str_split_fixed(geno$Sample, pattern = "-",n=3)
     [,1]
              [,2]
                        [,3]
[1,] "TaqMan" "SG0001" "190601"
[2,] "TaqMan" "SG0104" "190602"
[3,] "TaqMan" "SG2112" "190603"
[4,] "TaqMan" "Sg2543" "190603"
  geno$ID <- toupper(a[,2])</pre>
  kable(geno)
                   Sample
                                          rs1212
                                                 ID
                   TaqMan-SG0001-190601
                                          G/C
                                                  SG0001
                   TaqMan-SG0104-190602
                                          G/G
                                                  SG0104
                   TaqMan-SG2112-190603
                                          C/C
                                                  SG2112
```

```
TaqMan-Sg2543-190603 C/G SG2543

The separate function from the tidyr package is also useful:

geno %>% separate(Sample, into=c("Tech","ID","Suffix"), sep="-")

Tech ID Suffix rs1212
1 TaqMan SG0001 190601 G/C
2 TaqMan SG0104 190602 G/G
3 TaqMan SG2112 190603 C/C
4 TaqMan Sg2543 190603 C/G
```

6.5 Scenario 2

A replication sample has been measured, and that is using IDs in the style "RP5XXX".

```
joint <- read.table(file = "data/joint_data.txt", header = TRUE)
kable(joint)</pre>
```

ID	SBP
SG0001	120
SG0104	111
SG2112	125
SG2543	119
RP5002	121
RP5012	118
RP5113	112
RP5213	142

6.5.1 Question 4

Discuss how you would use R commands to split the 'joint' data frame into an 'SG' and 'RP' specific piece? Write down your ideas in the next section, and, if you have time, try them out within an R chunk.

Table 6.12: The joint data frame

ID	SBP
SG0001	120
SG0104	111
SG2112	125
SG2543	119
RP5002	121
RP5012	118
RP5113	112
RP5213	142

```
# Download files within the WebR environment
dir.create("data")
infiles <- c("data/joint_data.txt")
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
for (i in 1:length(infiles)) {
    download.file(paste0(root_srcfile,infiles[i]), infiles[i])
}
joint <- read.table(file = "data/joint_data.txt", header = TRUE)
kable(joint)
# Edit/add code here</pre>
```

6.5.2 Answer 4

```
Expand to see solution

grep(pattern = "SG",joint$ID)

[1] 1 2 3 4

grep(pattern = "RP", joint$ID)

[1] 5 6 7 8

joint.SG <- joint[grep(pattern = "SG",joint$ID), ]
 joint.RP <- joint[grep(pattern = "RP", joint$ID), ]
 kable(joint.SG)</pre>
```

ID	SBP
SG0001	120
SG0104	111
SG2112	125
SG2543	119

kable(joint.RP)

	ID	SBP
5	RP5002	121
6	RP5012	118
7	RP5113	112
8	RP5213	142

Reset row names
rownames(joint.RP) <- NULL
kable(joint.RP)</pre>

ID	SBP
RP5002	121
RP5012	118
RP5113	112
RP5213	142

7 R Functions Excercise

7.1 Load Libraries

```
library(tidyverse)
# library(tidylog)
```

7.2 Location

This file exercise1_solution.Qmd is in the "HuGen2071_book" sub-folder of the "hugen2071" folder of our Lectures repository.

```
paste0(basename(dirname(getwd())),"/",basename(getwd()))
[1] "hugen2071/HuGen2071_book"
```

7.3 Data set creation code

```
i <- 6
for (i in 1:10) {
fl <- data.frame(name=rep(paste0("name",i),26))
b <- data.frame(name = rep(NA, 26))
b$name <- paste0(fl$name,"_",letters)
b$trait <- rnorm(26)
write_tsv(b,paste0("data/dataset",i,".txt"))
}</pre>
```

7.4 Example

Here we have been sent three data sets in the files that contain the trait quantitative values for each person in the data set:

```
"dataset1.txt" "dataset2.txt" "dataset3.txt"
```

And we've been asked to make a table that gives, for each dataset, the sample size (N), the mean of the trait, the median, and the variance.

We could do this by reading in each data set, one by one, as follows:

```
results <- data.frame(dataset=rep(NA,3),N=NA, mean=NA, median=NA, var=NA)
  fl1 <- read.table("data/dataset1.txt",sep="\t",header=TRUE)</pre>
  results$dataset[1] <- "dataset1"</pre>
  results$N <- nrow(fl1)
  results$mean[1] <- mean(fl1$trait)</pre>
  results$median[1] <- median(fl1$trait)</pre>
  results$var[1] <- var(fl1$trait)</pre>
  results
   dataset N
                              median
                     mean
1 dataset1 26 0.09762111 0.2198957 0.5974116
2
      <NA> 26
                       NA
                                   NA
                                              NA
3
      <NA> 26
                       NA
                                   NA
                                              NA
  f12 <- read.table("data/dataset2.txt",sep="\t",header=TRUE)</pre>
  results$dataset[2] <- "dataset2"</pre>
  results$N <- nrow(fl2)
  results$mean[2] <- mean(fl2$trait)</pre>
  results$median[2] <- median(fl2$trait)</pre>
  results$var[2] <- var(fl2$trait)</pre>
  results
   dataset
                              median
           N
                     mean
                                             var
1 dataset1 26 0.09762111 0.2198957 0.5974116
2 dataset2 26 0.43486401 0.3558736 1.0936651
3
      <NA> 26
                       NΑ
                                  NA
                                              NA
  f13 <- read.table("data/dataset3.txt", sep="\t", header=TRUE)
  results$dataset[3] <- "dataset3"
```

```
results$N <- nrow(fl3)
results$mean[3] <- mean(fl3$trait)
results$median[3] <- median(fl3$trait)
results$var[3] <- var(fl3$trait)
results

dataset N mean median var
1 dataset1 26 0.09762111 0.2198957 0.5974116
2 dataset2 26 0.43486401 0.3558736 1.0936651
3 dataset3 26 0.07508335 0.0445614 0.7950574</pre>
```

Your colleague initially sent you the three data sets above, but now your colleague has sent you three more data sets and asked you to update the 'results' table.

As you can see, the code above is very repetitive. So let's automate this by writing a function that loops through a list of data set files named "dataset1.txt", "dataset2.txt", "dataset3.txt", etc., building up the results table as above.

7.4.1 Question: How could we construct a list of file names?

How could we construct a list of file names?

```
Expand to see solution

Hint: the list.files command provides a handy way to get a list of the input files:

fls <- list.files(path="data",pattern="dataset*")
fls

[1] "dataset1.txt" "dataset2.txt" "dataset3.txt" "dataset4.txt" "dataset5.txt"
[6] "dataset6.txt"</pre>
```

7.4.2 Question: Outline a possible algorithm

Outline a possible algorithm that loops through a list of input data set files named "dataset1.txt", "dataset2.txt", "dataset3.txt", etc., building up the results table as above.

? Expand to see solution

- Read in the input file names into a list
- Set up an empty results table
- For each file in our file name list
 - Read the file
 - Compute the statistics
 - Insert the information into the results table
 - Return the filled-in results table

7.4.3 Question: Construct a more detailed step-by-step algorithm.

Construct a more detailed step-by-step algorithm.

? Expand to see solution

- Input the path to the folder containing the data files
- Read in the input file names into a list fls
- Count the number of input files N
- Set up an empty results table with N rows
- For each file in our file name list fls
 - Read the file
 - Compute the statistics
 - Insert the information into the correct row of the results table
- Return the filled-in results table

7.4.4 Task: Write a read data file function.

Write a read_data_file function to accomplish the required steps for a single input data file.

1. Make the number in the data file name an argument.



Here we make the number in the data file name an argument

```
results <- data.frame(dataset=rep(NA,6),N=NA, mean=NA, median=NA, var=NA)
read_data_file <- function(n=1, results) {
  fl1 <- read.table(paste0("data/dataset",n,".txt"),sep="\t",header=TRUE)
  results$dataset[n] <- paste0("dataset",n,".txt")
  results$N <- nrow(fl1)
  results$mean[n] <- mean(fl1$trait)
  results$median[n] <- median(fl1$trait)
  results$var[n] <- var(fl1$trait)
  invisible(results)
}</pre>
```

2. Make the path to the input file an argument to your read_data_file function.

```
Expand to see solution

Here we make the path to the input file an argument.

read_data_file_v2 <- function(flnm, results) {
   fl1 <- read.table(paste0("data/",flnm),sep="\t",header=TRUE)
   results$dataset[n] <- flnm
   results$N <- nrow(fl1)
   results$mean[n] <- mean(fl1$trait)
   results$median[n] <- median(fl1$trait)
   results$var[n] <- var(fl1$trait)
   invisible(results)
}</pre>
```

7.4.5 Question: What does the above code assume?

What does the above code assume?

? Expand to see solution

Assumes a file naming style of 'dataset*.txt' where the asterisk represents 1, 2, 3, ... Assumes the files are in the "data" folder.

7.4.6 Question: Extend your function to process all of the files

The above function read_data_file processes one file at a time. How would you write a function to loop this over to process all of our files?

```
Expand to see solution
  fls <- list.files(path="data",pattern="dataset*")</pre>
  loop_over_dataset <- function(fls) {</pre>
    # Input: the list of file names
    # Output: the 'results table
    # Count the number of data set file names in fls
    n datasets <- length(fls)</pre>
    # Set up a results dataframe with n_datasets rows
    results <- data.frame(dataset=rep(NA, n datasets), N=NA, mean=NA, median=NA, var=NA)
    for (n in 1:n_datasets) {
      results <- read_data_file(n=n, results=results)</pre>
    return(results)
  }
  loop_over_dataset(fls = fls)
       dataset N
                                    median
                          mean
1 dataset1.txt 26  0.09762111  0.21989574  0.5974116
2 dataset2.txt 26  0.43486401  0.35587359  1.0936651
3 dataset3.txt 26  0.07508335  0.04456140  0.7950574
4 dataset4.txt 26 0.06259720 0.04813915 0.9186042
5 dataset5.txt 26 -0.09288522 -0.19155759 0.9978161
6 dataset6.txt 26 -0.20266667 -0.23845426 1.5605823
```

7.4.7 Bonus question

Can you find a subtle mistake in the read_data_file function?

```
results <- data.frame(dataset=rep(NA,6),N=NA, mean=NA, median=NA, var=NA)
read_data_file <- function(n=1, results) {
  fl1 <- read.table(paste0("data/dataset",n,".txt"),sep="\t",header=TRUE)
  results$dataset[n] <- paste0("dataset",n,".txt")</pre>
```

```
results$N <- nrow(fl1)
results$mean[n] <- mean(fl1$trait)
results$median[n] <- median(fl1$trait)
results$var[n] <- var(fl1$trait)
invisible(results)
}</pre>
```

? Expand to see solution

If ${\tt N}$ varies across the data sets, then this line will not do the right thing:

```
results$N <- nrow(fl1)

results <- data.frame(dataset=rep(NA,6),N=NA, mean=NA, median=NA, var=NA)
read_data_file <- function(n=1, results) {
   fl1 <- read.table(paste0("data/dataset",n,".txt"),sep="\t",header=TRUE)
   results$dataset[n] <- paste0("dataset",n,".txt")
   results$N[n] <- nrow(fl1)
   results$mean[n] <- mean(fl1$trait)
   results$var[n] <- var(fl1$trait)
   invisible(results)
}</pre>
```

8 R Tidyverse Exercise

8.1 Load Libraries

Load the tidyverse packages

```
library(tidyverse)
# library(tidylog)
```

8.2 Untidy data

Let's use the World Health Organization TB data set from the tidyr package

```
who <- tidyr::who
dim(who)

[1] 7240 60

head(who[,1:6] %>% filter(!is.na(new_sp_m014)))
```

```
# A tibble: 6 x 6
  country
             iso2 iso3
                           year new_sp_m014 new_sp_m1524
  <chr>>
              <chr> <chr> <dbl>
                                       <dbl>
                                                     <dbl>
1 Afghanistan AF
                    AFG
                           1997
                                          0
                                                        10
2 Afghanistan AF
                    AFG
                           1998
                                          30
                                                       129
3 Afghanistan AF
                    AFG
                           1999
                                           8
                                                        55
                    AFG
                                          52
                                                       228
4 Afghanistan AF
                           2000
5 Afghanistan AF
                    AFG
                           2001
                                         129
                                                       379
                    AFG
                                          90
                                                       476
6 Afghanistan AF
                           2002
```

See the help page for who for more information about this data set.

In particular, note this description:

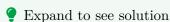
"The data uses the original codes given by the World Health Organization. The column names for columns five through 60 are made by combining new_ to a code for method of diagnosis (rel = relapse, sn = negative pulmonary smear, sp = positive pulmonary smear, ep = extrapulmonary) to a code for gender (f = female, m = male) to a code for age group (014 = 0-14 yrs of age, 1524 = 15-24 years of age, 2534 = 25 to 34 years of age, 3544 = 35 to 44 years of age, 4554 = 45 to 54 years of age, 5564 = 55 to 64 years of age, 65 = 65 years of age or older)."

So new_sp_m014 represents the counts of new TB cases detected by a positive pulmonary smear in males in the 0-14 age group.

8.3 Tidy data

Tidy data: Have each variable in a column.

Question: Are these data tidy?



No these data are not tidy because aspects of the data that should be variables are encoded in the name of the variables.

These aspects are

- 1. test type.
- 2. sex of the subjects.
- 3. age range of the subjects.

Question: How would we make these data tidy?

Consider this portion of the data:

```
head(who[,1:5] %>% filter(!is.na(new_sp_m014) & new_sp_m014>0), 1)
# A tibble: 1 x 5
country iso2 iso3 year new sp m014
```

? Expand to see solution

We would replace the new_sp_m014 with the following four columns:

```
\begin{array}{ccccc} \text{type} & \text{sex} & \text{age} & \text{n} \\ \text{sp} & \text{m} & \text{014} & \text{30} \end{array}
```

This would place each variable in its own column.

8.4 Gather

```
stocks <- tibble(</pre>
    time = as.Date('2009-01-01') + 0:9,
    X = rnorm(10, 0, 1),
    Y = rnorm(10, 0, 2),
    Z = rnorm(10, 0, 4)
  )
  head(stocks)
# A tibble: 6 x 4
 time
                  Х
                         Y
  <date>
              <dbl> <dbl>
                            <dbl>
1 2009-01-01 0.760 -4.29
                            5.56
2 2009-01-02 -1.94 -1.41
                            2.35
3 2009-01-03 -0.381 -1.28 -0.542
4 2009-01-04 -0.751 -0.326 2.07
5 2009-01-05 -0.270 0.716 -1.72
6 2009-01-06 -0.218 -2.06 -2.34
  stocks %>% gather("stock", "price", -time) %>% head()
# A tibble: 6 x 3
             stock price
 time
             <chr> <dbl>
  <date>
1 2009-01-01 X
                    0.760
2 2009-01-02 X
                   -1.94
3 2009-01-03 X
                   -0.381
4 2009-01-04 X
                   -0.751
```

```
5 2009-01-05 X -0.270
6 2009-01-06 X -0.218
```

8.5 Pivot_longer

```
stocks %>% pivot_longer(c(X,Y,Z), names_to= "stock", values_to = "price") %>%
    head()
# A tibble: 6 x 3
             stock price
  time
  <date>
             <chr> <dbl>
1 2009-01-01 X
                    0.760
                   -4.29
2 2009-01-01 Y
3 2009-01-01 Z
                    5.56
4 2009-01-02 X
                   -1.94
5 2009-01-02 Y
                   -1.41
6 2009-01-02 Z
                    2.35
```

8.6 WHO TB data

Question: How would we convert this to tidy form?

```
head(who[,1:6] %>% filter(!is.na(new_sp_m014)))
```

```
# A tibble: 6 x 6
                            year new_sp_m014 new_sp_m1524
  country
              iso2 iso3
  <chr>
              <chr> <chr> <dbl>
                                       <dbl>
                                                     <dbl>
                    AFG
1 Afghanistan AF
                            1997
                                           0
                                                        10
2 Afghanistan AF
                    AFG
                                          30
                                                       129
                            1998
                    AFG
3 Afghanistan AF
                            1999
                                           8
                                                        55
4 Afghanistan AF
                    AFG
                            2000
                                          52
                                                       228
5 Afghanistan AF
                    AFG
                            2001
                                         129
                                                       379
6 Afghanistan AF
                    AFG
                            2002
                                          90
                                                       476
```

```
Expand to see solution
  who.long <- who %>% pivot_longer(starts_with("new"), names_to = "demo", values_to = "n")
  head(who.long)
# A tibble: 6 x 6
  country
              iso2
                           year demo
                    iso3
  <chr>
              <chr> <chr> <dbl> <chr>
                                              <dbl>
1 Afghanistan AF
                    AFG
                           1997 new_sp_m014
                                                  0
2 Afghanistan AF
                    AFG
                           1997 new_sp_m1524
                                                 10
3 Afghanistan AF
                           1997 new_sp_m2534
                    AFG
                                                  6
4 Afghanistan AF
                    AFG
                           1997 new_sp_m3544
                                                  3
5 Afghanistan AF
                    AFG
                            1997 new_sp_m4554
                                                  5
6 Afghanistan AF
                    AFG
                            1997 new_sp_m5564
                                                  2
```

Question: How would we split demo into variables?

```
head(who.long)
```

```
# A tibble: 6 x 6
  country
              iso2 iso3
                           year demo
  <chr>
              <chr> <chr> <dbl> <chr>
                                              <dbl>
                           1997 new_sp_m014
1 Afghanistan AF
                    AFG
                                                  0
2 Afghanistan AF
                           1997 new_sp_m1524
                    AFG
                                                 10
3 Afghanistan AF
                    AFG
                           1997 new_sp_m2534
                                                  6
4 Afghanistan AF
                    AFG
                           1997 new_sp_m3544
                                                  3
5 Afghanistan AF
                    AFG
                           1997 new_sp_m4554
                                                  5
6 Afghanistan AF
                    AFG
                                                  2
                           1997 new_sp_m5564
```

Look at the variable naming scheme:

```
names(who) %>% grep("m014",., value=TRUE)
```

```
[1] "new_sp_m014" "new_sn_m014" "new_ep_m014" "newrel_m014"
```

Question: How should we adjust the demo strings so as to be able to easily split all of them into the desired variables?

```
who.long <- who.long %>%
    mutate(demo = str_replace(demo, "newrel", "new_rel"))
    grep("m014",who.long$demo, value=TRUE) %>% unique()

[1] "new_sp_m014" "new_sn_m014" "new_ep_m014" "new_rel_m014"
```

Question: After adjusting the demo strings, how would we then separate them into the desired variables?

```
Expand to see solution
  who.long <- who.long %>%
    separate(demo, into = c("new", "type", "sexagerange"), sep="_") %>%
    separate(sexagerange, into=c("sex", "age_range"), sep=1) %>%
    select(-new)
  head(who.long)
# A tibble: 6 x 8
              iso2 iso3
  country
                           year type sex
                                             age_range
  <chr>
              <chr> <chr> <chr> <chr> <chr> <chr> <chr>
                                                        <dbl>
1 Afghanistan AF
                    AFG
                                             014
                            1997 sp
                                                            0
2 Afghanistan AF
                    AFG
                            1997 sp
                                             1524
                                                           10
                                       m
                            1997 sp
3 Afghanistan AF
                    AFG
                                             2534
                                                            6
                                       m
4 Afghanistan AF
                    AFG
                            1997 sp
                                             3544
                                                            3
                                       m
5 Afghanistan AF
                    AFG
                            1997 sp
                                             4554
                                                            5
                                       m
6 Afghanistan AF
                    AFG
                                                            2
                            1997 sp
                                             5564
```

8.7 Conclusion

Now our untidy data are tidy.

1	Afghanistan	AF	AFG	1997	sp	m	014	0
2	Afghanistan	AF	AFG	1997	sp	m	1524	10
3	Afghanistan	AF	AFG	1997	sp	m	2534	6
4	Afghanistan	AF	AFG	1997	sp	m	3544	3
5	Afghanistan	AF	AFG	1997	sp	m	4554	5
6	Afghanistan	AF	AFG	1997	sp	m	5564	2

8.8 Acknowledgment

This exercise was modeled, in part, on this exercise:

 $https://people.duke.edu/\sim ccc14/cfar-data-workshop-2018/CFAR_R_Workshop_2018_Exercisees.html$

9 R Recoding Reshaping Exercise

9.1 Load Libraries

```
library(tidyverse)
# library(tidylog)
```

9.2 Project 1 Data

In the ds data frame we have the synthetic yet realistic data we will be using in Project 1. In the dd data frame we have the corresponding data dictionary.

```
load("data/exercise.RData", verbose = TRUE)
Loading objects:
  ds
  dd
  DictPer
  dim(ds)
[1] 191 24
  names(ds)
 [1] "sample_id"
                                 "Sample_trimester"
 [3] "Gestationalage_sample"
                                 "subject_id"
 [5] "strata"
                                 "race"
 [7] "maternal_age_delivery"
                                 "case_control_status"
 [9] "prepregnancy_weight"
                                 "height"
```

```
[11] "prepregnancy_BMI"
                                 "gravidity"
[13] "parity"
                                 "gestationalage_delivery"
[15] "average_SBP_lt20weeks"
                                 "average_DBP_lt20weeks"
[17] "average_SBP_labor"
                                 "average_DBP_labor"
[19] "smoke_lifetime"
                                 "baby_birthweight"
                                 "baby_birthweight_centile"
[21] "baby_sex"
[23] "baby_SGA"
                                 "placental_pathology"
  dim(dd)
[1] 27 5
  names (dd)
[1] "Original.Variable.Name" "R21.Variable.Name"
                                                        "Description"
[4] "Variable.Units"
                              "Variable.Coding"
```

9.3 Exercise 1

Skill: Checking for duplicated IDs

```
ds %>%
      select(subject_id, sample_id, height) %>%
      head(n = 10)
  subject_id sample_id height
      SUBJ48
               SAMP149
                          64.6
1
2
      SUBJ46
               SAMP037
                         65.7
3
                         63.3
      SUBJ28
               SAMP120
4
      SUBJ26
               SAMP187
                         61.1
5
      SUBJ49
               SAMP082
                         67.6
6
      SUBJ48
               SAMP149
                         64.6
7
      SUBJ19
               SAMPO74
                         66.1
8
      SUBJ07
               SAMP063
                         64.4
9
      SUBJ28
               SAMP053
                         63.3
                          65.7
10
      SUBJ43
               SAMP085
```

Check if there are any duplicated sample_id's using the duplicated command.

```
Expand to see solution

sum(duplicated(ds$sample_id))

[1] 72
```

Construct a table of the number of times each sample_id is duplicated:

```
? Expand to see solution
  table(table(ds$sample_id))
67 35 13 2 1
  # But?
  sum(duplicated(ds$sample_id))
[1] 72
  35 + 13 * 2 + 2 * 3 + 1 * 4
[1] 71
  sum(duplicated(ds$sample_id, incomparables = NA))
[1] 71
  table(table(ds$sample_id, useNA = "always"))
 1 2 3
67 36 13 2 1
  36 + 13 * 2 + 2 * 3 + 1 * 4
[1] 72
```

Check if there are any duplicated subject_ids

```
Expand to see solution

sum(duplicated(ds$subject_id))

[1] 137
```

9.4 Checking for duplicates

How do we return every row that contains a duplicate?

9.5 Counting the number of occurences of the ID

9.6 Count sample_id duplicates

Using Tidyverse commands, count how many times each sample_id occcurs in the ds data frame, reporting the counts in descending order, from highest to lowest.

```
? Expand to see solution
  ds %>%
      group_by(sample_id) %>%
      summarise(n = n()) \%>\%
      filter(n > 1) %>%
      arrange(desc(n)) %>%
      head()
# A tibble: 6 x 2
  sample_id
  <chr>
            <int>
1 SAMP100
2 SAMP125
                 4
3 SAMP155
                 4
4 SAMP017
                 3
                 3
5 SAMP048
6 SAMP058
                3
  ds %>%
      group_by(sample_id) %>%
      summarise(n = n()) \%>\%
      filter(n > 1) \%>\%
      arrange(desc(n)) %>%
      pull(n) %>%
      table()
    3
       4 5
36 13
       2 1
```

9.7 Checking for duplicates

Here we list all of the rows containing a duplicated 'ID' value using functions from the 'tidy-verse' package:

```
f %>%
    group_by(ID) %>%
    filter(n() > 1)

# A tibble: 2 x 2
# Groups: ID [1]
    ID    c2
    <dbl> <dbl>
1     1    1
2     1    2
```

9.7.1 How to list all duplicates

Use Tidyverse commands to list all duplicates for sample_id and for subject_id. Sort the results by the ID.

```
? Expand to see solution
9.7.2 Sample ID
  ds %>%
      group_by(sample_id) %>%
      filter(n() > 1) \%>\%
      select(sample_id, subject_id, Sample_trimester, Gestationalage_sample) %>%
      arrange(sample_id, Sample_trimester, Gestationalage_sample) %>%
      head()
# A tibble: 6 x 4
# Groups:
            sample_id [3]
  sample_id subject_id Sample_trimester Gestationalage_sample
  <chr>
            <chr>
                                   <dbl>
                                                          <dbl>
1 SAMP002
            SUBJ20
                                       2
                                                          19.3
2 SAMP002
            SUBJ20
                                       2
                                                          19.7
3 SAMP003
                                                          8.25
            SUBJ12
                                       1
4 SAMP003
            SUBJ12
                                       1
                                                          8.35
5 SAMP004
                                       2
                                                          20.4
            SUBJ35
6 SAMP004
            SUBJ35
                                       2
                                                          20.9
```

9.7.3 Subject ID ds %>% group_by(subject_id) %>% filter(n() > 1) %>% select(subject_id, sample_id, Sample_trimester, Gestationalage_sample) %>% arrange(subject_id, sample_id, Sample_trimester, Gestationalage_sample) %>% head(10) # A tibble: 10 x 4 # Groups: subject_id [2] subject_id sample_id Sample_trimester Gestationalage_sample <chr>> <chr> <dbl> <dbl> 1 SUBJ01 SAMP011 9.00 1 2 SUBJ01 SAMP034 3 39.8 3 SUBJ01 SAMP034 3 42.1 2 4 SUBJ01 SAMP103 19.9 2 5 SUBJ01 SAMP103 20.0 SAMP155 6 SUBJ01 3 40.0 7 SUBJ01 SAMP155 3 40.5 8 SUBJ01 3 40.7 SAMP155 9 SUBJ01 3 41.6 SAMP155 3 10 SUBJ02 38.6 SAMP113

9.8 Exercise 2

Skill: Reshaping data

Select only three columns "sample_id", "Sample_trimester", "Gestationalage_sample", and then reshape from 'long' format to 'wide' format using pivot_wider, taking time as the "Sample_trimester".

```
Expand to see solution

b <- ds %>%
    select(sample_id, Sample_trimester, Gestationalage_sample)

b2 <- b %>%
    pivot_wider(id_cols = sample_id, names_from = Sample_trimester, values_from = Gestatent
```

```
Warning: Values from `Gestationalage_sample` are not uniquely identified; output will
contain list-cols.
* Use `values_fn = list` to suppress this warning.
* Use `values_fn = {summary_fun}` to summarise duplicates.
* Use the following dplyr code to identify duplicates.
  {data} %>%
 dplyr::group_by(sample_id, Sample_trimester) %>%
 dplyr::summarise(n = dplyr::n(), .groups = "drop") %>%
 dplyr::filter(n > 1L)
  head(b2)
# A tibble: 6 x 5
                                `2`
  sample_id `1`
                      `3`
                                          `NA`
  <chr>
            t>
                      t>
                                t>
                                          st>
1 SAMP149
            <dbl [3]> <NULL>
                                <NULL>
                                          <NULL>
2 SAMP037
            <dbl [2]> <NULL>
                                <NULL>
                                          <NULL>
            <dbl [3]> <NULL>
3 SAMP120
                                <NULL>
                                          <NULL>
4 SAMP187
            <NULL>
                      <dbl [1]> <NULL>
                                          <NULL>
5 SAMP082
            <NULL>
                      <NULL>
                                <dbl [1]> <NULL>
6 SAMP074
            <NULL>
                      <NULL>
                                <dbl [3]> <NULL>
9.8.1 Comment
```

9.9 Exercise 3

Skill: Aggregating data

Make a table showing the proportion of blacks and whites that are controls and cases.

View b2 via the View(b2) command in RStudio - it nicely put all the different gestational age observations into one list for each sample_id x Sample_trimester combination.

```
Expand to see solution

prop.table(table(ds$case_control_status, ds$race), margin = 2)

B     W     White
0 0.5396825 0.5156250 0.0000000
```

```
1 0.4603175 0.4843750 1.0000000
```

9.9.1 Comment:

The margin parameter of the prop.table command has to be specified in order to get the desired answer: "1 indicates rows, 2 indicates columns.

Construct more readable tables with labels using xtabs

Create a count cross table using Tidyverse commands

Expand to see solution ds %>% group_by(case_control_status, race) %>% summarize(n = n()) %>%spread(race, n) `summarise()` has grouped output by 'case_control_status'. You can override using the `.groups` argument. # A tibble: 2 x 4 case_control_status [2] # Groups: case_control_status В W White <dbl> <int> <int> <int> 1 68 33 NΑ 2 1 58 31 1 addmargins(xtabs(~case_control_status + race, data = ds)) race case_control_status В W White Sum 68 33 0 101 1 90 1 58 31 Sum 126 64 1 191

Create a proportion cross table using Tidyverse commands

```
Expand to see solution

ds %>%
    group_by(case_control_status, race) %>%
    summarize(n = n()) %>%
    mutate(prop = n/sum(n)) %>%
    select(-n) %>%
    spread(race, prop)

`summarise()` has grouped output by 'case_control_status'. You can override using the `.groups` argument.

# A tibble: 2 x 4
```

9.10 Exercise 4

Skill: Summarizing within groups

Apply the **summary** command to the "Gestationalage_sample" within each "Sample_trimester" group.

```
? Expand to see solution
  f <- split(ds[, "Gestationalage_sample"], ds$Sample_trimester)</pre>
  sapply(f, summary)
                1
Min.
         4.934325 16.53800 31.44880
1st Qu. 7.838825 18.45761 35.16305
         8.565282 19.72388 37.71093
Mean
         8.616799 19.83310 37.37827
3rd Qu.
         9.193104 20.69576 39.11360
        13.026958 24.60659 42.09340
Max.
  # Or 'tapply' can be used:
  tapply(ds$Gestationalage_sample, ds$Sample_trimester, summary)
$`1`
  Min. 1st Qu. Median
                            Mean 3rd Qu.
                                            Max.
  4.934
          7.839
                  8.565
                          8.617
                                   9.193 13.027
$`2`
                           Mean 3rd Qu.
  Min. 1st Qu.
                 Median
                                            Max.
                  19.72
                                   20.70
                                           24.61
  16.54
          18.46
                           19.83
$`3`
  Min. 1st Qu.
                 Median
                          Mean 3rd Qu.
                                            Max.
  31.45
                  37.71
                          37.38
                                   39.11
                                           42.09
          35.16
```

Note: With split(x, f), any missing values in f are dropped together with the corresponding values of x.

9.11 Exercise 5: Recoding data

Approach 1

- Implement our dictionaries using look-up tables
 - Use a named vector.

Skill:: Recoding IDs using a dictionary

Create a new subject ID column named "subjectID" where you have used the DictPer named vector to recode the original "subject_id" IDs into integer IDs.

```
head(DictPer)
```

```
SUBJ48 SUBJ46 SUBJ28 SUBJ26 SUBJ49 SUBJ19
40 2 23 38 10 27
```

```
Expand to see solution
  a5 <- ds
  a5$ID <- DictPer[a5$subject_id]
  a5 %>%
      select(subject_id, ID) %>%
      head
  subject_id ID
1
      SUBJ48 40
2
      SUBJ46 2
3
      SUBJ28 23
      SUBJ26 38
5
      SUBJ49 10
      SUBJ48 40
  head(DictPer)
SUBJ48 SUBJ46 SUBJ28 SUBJ26 SUBJ49 SUBJ19
    40
            2
                   23
                          38
                                  10
                                         27
```

9.12 Recoding data

Approach 2

• Implement our dictionaries using left joins

9.12.1 Comment

I usually prefer to use a merge command like left_join to merge in the new IDs into my data frame.

```
? Expand to see solution
  key <- data.frame(SubjID = names(DictPer), ID = DictPer)</pre>
  head(key)
       SubjID ID
SUBJ48 SUBJ48 40
SUBJ46 SUBJ46 2
SUBJ28 SUBJ28 23
SUBJ26 SUBJ26 38
SUBJ49 SUBJ49 10
SUBJ19 SUBJ19 27
  b5 <- left_join(ds, key, by = c(subject_id = "SubjID"))
      select(subject_id, ID) %>%
      head()
  subject_id ID
1
      SUBJ48 40
2
      SUBJ46 2
3
      SUBJ28 23
4
      SUBJ26 38
5
      SUBJ49 10
      SUBJ48 40
```

9.13 Exercise 6

Skill: Filtering rows.

Create a data frame tri1 containing the records for Trimester 1, and a second data frame tri2 containing the records for Trimester 2.

```
? Expand to see solution
  tri1 <- ds %>%
      filter(Sample_trimester == 1)
      select(subject_id, sample_id, Sample_trimester) %>%
      head()
  subject_id sample_id Sample_trimester
1
      SUBJ48
               SAMP149
2
      SUBJ46
               SAMP037
                                        1
3
      SUBJ28
               SAMP120
                                        1
4
      SUBJ48
               SAMP149
5
      SUBJ07
               SAMP063
                                        1
      SUBJ28
               SAMP053
  tri2 <- ds %>%
      filter(Sample_trimester == 2)
      select(subject_id, sample_id, Sample_trimester) %>%
      head()
  subject_id sample_id Sample_trimester
1
      SUBJ49
               SAMP082
                                        2
2
      SUBJ19
               SAMPO74
                                        2
3
      SUBJ10
                                        2
               SAMP121
4
      SUBJ22
               SAMP184
                                        2
5
                                        2
      SUBJ29
               SAMP100
                                        2
      SUBJ19
               SAMP074
```

9.14 Exercise 7

Skill: Selecting columns

Update tri1 and tri2 to only contain the three columns "sample_id", "Sample_trimester", "Gestationalage_sample"

```
Expand to see solution
  tri1 <- tri1 %>%
      select(sample_id, Sample_trimester, Gestationalage_sample)
  head(tri1)
 sample_id Sample_trimester Gestationalage_sample
   SAMP149
                                          8.094299
                           1
   SAMP037
                           1
                                          7.146034
3
   SAMP120
                           1
                                          7.122495
   SAMP149
                                          8.473876
                           1
   SAMP063
                           1
                                          7.510132
   SAMP053
                           1
                                          7.446434
  tri2 <- tri2 %>%
      select(sample_id, Sample_trimester, Gestationalage_sample)
  head(tri2)
 sample_id Sample_trimester Gestationalage_sample
   SAMP082
                                          21.89337
1
                           2
2 SAMP074
                           2
                                          21.26259
   SAMP121
                           2
3
                                          18.29106
   SAMP184
                           2
                                          18.76825
5
                           2
                                          24.48074
   SAMP100
   SAMP074
                           2
                                          21.24652
```

10 R Merging Exercise

10.1 Load Libraries

```
library(tidyverse)
library(tidylog)
```

10.2 Input data

Let's load the synthetic simulated Project 1 data and associated data dictionary:

```
load("data/project1.RData", verbose = TRUE)
Loading objects:
   ds
   dd
```

10.3 Select a subset of subject-level fields

```
Set up a data frame 'a' that has these subject-level fields: "subject_id" "mater-
nal_age_delivery" "case_control_status"
"prepregnancy_BMI"

a <- ds %>%
    select("subject_id", "maternal_age_delivery", "case_control_status", "prepregnancy_BMI
    arrange(subject_id)
```

select: dropped 20 variables (sample_id, Sample_trimester, Gestationalage_sample, strata, ra-

```
head(a, 10)
```

```
subject_id maternal_age_delivery case_control_status prepregnancy_BMI
       SUBJ01
                             20.01345
                                                                    45.29379
1
2
                                                         0
       SUBJ01
                             20.01345
                                                                    45.29379
3
       SUBJ01
                                                         0
                                                                    45.29379
                            20.01345
4
                                                         0
       SUBJ01
                            20.01345
                                                                    45.29379
5
       SUBJ01
                                                         0
                                                                    45.29379
                            20.01345
6
       SUBJ01
                            20.01345
                                                         0
                                                                    45.29379
7
       SUBJ01
                            20.01345
                                                         0
                                                                    45.29379
8
       SUBJ01
                            20.01345
                                                         0
                                                                    45.29379
9
       SUBJ01
                            20.01345
                                                         0
                                                                    45.29379
10
       SUBJ02
                            22.22541
                                                         1
                                                                    41.63679
  tail(a)
    subject_id maternal_age_delivery case_control_status prepregnancy_BMI
186
        SUBJ55
                             23.79660
                                                                     30.73757
                                                          1
187
        SUBJ56
                             20.77767
                                                                     32.30103
188
        SUBJ56
                             20.77767
                                                          1
                                                                     32.30103
189
                             20.77767
                                                          1
                                                                     32.30103
        SUBJ56
190
                             20.77767
                                                          1
                                                                     32.30103
        SUBJ56
```

10.4 Unique records

SUBJ56

191

The data were given to us in a way that repeated subject-level information, once for each sample from each individual subject.

1

32.30103

From your data frame 'a' select only the unique records, creating data frame b.

20.77767

```
Expand to see solution
  b <- unique(a)
  head(b)
   subject_id maternal_age_delivery case_control_status prepregnancy_BMI
                                                         0
1
       SUBJ01
                            20.01345
                                                                    45.29379
                                                         1
10
       SUBJ02
                            22.22541
                                                                    41.63679
13
       SUBJ03
                            20.80036
                                                         1
                                                                    32.55473
15
       SUBJ04
                            21.94422
                                                         0
                                                                    35.09978
19
       SUBJ05
                            20.18760
                                                                    41.85877
```

```
22
       SUBJ06
                            25.70581
                                                                    38.02936
  b1 <- a %>%
      distinct()
distinct: removed 137 rows (72%), 54 rows remaining
  all.equal(b, b1)
[1] "Attributes: < Component \"row.names\": Mean relative difference: 0.7157633 >"
  all.equal(b, b1, check.attributes = FALSE)
[1] TRUE
  head(rownames(b))
[1] "1" "10" "13" "15" "19" "22"
  head(rownames(b1))
[1] "1" "2" "3" "4" "5" "6"
  # Reset row names
  rownames(b) <- NULL
  rownames(b1) <- NULL
  all.equal(b, b1)
[1] TRUE
10.4.1 Comment
It is better to apply unique to the whole data frame, not just to the subject_id column,
as that ensures that you are selecting whole records that are unique across all of their
columns.
```

 $(ex1 \leftarrow data.frame(ID = c(1, 1, 1, 2), trait = c(10, 9, 9, 11)))$

```
ID trait
3 1
         9
        11
  unique(ex1)
  ID trait
        10
  1
         9
  2
        11
  ex1 %>%
      distinct()
distinct: removed one row (25%), 3 rows remaining
  ID trait
        10
2 1
        9
3
  2
        11
```

10.5 Check that the subject_id's are now not duplicated

Are the subject_id's unique?

```
Expand to see solution

sum(duplicated(b$subject_id))

[1] 0

b %>%
    group_by(subject_id) %>%
    filter(n() > 1)

group_by: one grouping variable (subject_id)
```

```
filter (grouped): removed all rows (100%)

# A tibble: 0 x 4

# Groups: subject_id [0]

# ... with 4 variables: subject_id <chr>, maternal_age_delivery <dbl>,

# case_control_status <dbl>, prepregnancy_BMI <dbl>
```

10.6 Create random integer IDs

Create a new column ID containing randomly chosen integer IDs; this is necessary to de-identify the data. To do this, use the sample command, sampling integers from 1 to the number of rows in data frame b.

```
Expand to see solution
  set.seed(10234)
  b$ID <- sample(c(1:nrow(b)), replace = FALSE)</pre>
  head(b %>%
      select(subject_id, ID))
select: dropped 3 variables (maternal_age_delivery, case_control_status, prepregnancy_BMI)
  subject_id ID
      SUBJ01 6
1
2
      SUBJ02 4
3
      SUBJ03 41
      SUBJ04 14
      SUBJ05 51
      SUBJ06 37
  sum(duplicated(b$ID))
[1] 0
```

10.7 Merge in new phenotype information

The PI has sent you new trait data for your subjects.

```
new <- read_tsv("data/newtrait.tsv")</pre>
Rows: 54 Columns: 2
-- Column specification -----
Delimiter: "\t"
chr (1): subject_id
dbl (1): trait
i Use `spec()` to retrieve the full column specification for this data.
i Specify the column types or set `show_col_types = FALSE` to quiet this message.
  head(new)
# A tibble: 6 x 2
 subject_id trait
 <chr>
            <dbl>
1 SUBJ48
             130.
2 SUBJ46
             104.
3 SUBJ28
             125.
4 SUBJ26
             108.
5 SUBJ49
             129.
6 SUBJ19
             117.
  dim(new)
[1] 54 2
  dim(b)
[1] 54 5
```

Carefully merge this in using tidyverse commands.

If you notice any problems with this merge, prepare a report for the PI detailing what you noticed and what you'd like to ask the PI about.

10.8 Always be careful when merging.

- Always check for duplicated IDs before doing the merge.
- Always check that your ID columns do not contain any missing values.
- Check that the values in the ID columns (e.g., the keys) match.
 - Can use an 'anti_join' to check this.
 - Inconsistencies in the values of the keys can be hard to fix.
- Always check the dimensions to make sure the merged object has the expected number of rows and columns.
- Always explicitly name the keys you are merging on.
 - If you don't name them, then the join command will use all variables in common across x and y.

10.9 Merge in new phenotype information

Carefully merge in the new data in using tidyverse commands.

If you notice any problems with this merge, prepare a report for the PI detailing what you noticed and what you'd like to ask the PI about.

```
# Expand to see solution

# Check for duplicated IDs
sum(duplicated(b$subject_id))

[1] 0

sum(duplicated(new$subject_id))

[1] 1

# Which one is duplicated
new %>%
group_by(subject_id) %>%
mutate(n = n()) %>%
filter(n > 1)

group_by: one grouping variable (subject_id)
```

```
mutate (grouped): new variable 'n' (integer) with 2 unique values and 0% NA
filter (grouped): removed 52 rows (96%), 2 rows remaining
# A tibble: 2 x 3
# Groups:
           subject_id [1]
  subject_id trait
  <chr>
           <dbl> <int>
1 SUBJ09
              115.
2 SUBJ09
              115.
                      2
  # Check for missing IDs
  sum(is.na(b$subject_id))
Γ1 0
  sum(is.na(new$subject_id))
[1] 0
  # Check the dimensions
  dim(b)
[1] 54 5
  dim(new)
[1] 54 2
  b2 <- left_join(b, new, by = "subject_id")
left_join: added one column (trait)
           > rows only in x
                              2
           > rows only in y (1)
           > matched rows
                                   (includes duplicates)
                            53
```

```
> rows total
                              55
  head(b2)
  subject_id maternal_age_delivery case_control_status prepregnancy_BMI ID
     SUBJ01
                          20.01345
                                                                 45.29379 6
1
2
      SUBJ02
                          22.22541
                                                      1
                                                                 41.63679 4
3
     SUBJ03
                          20.80036
                                                       1
                                                                 32.55473 41
                          21.94422
                                                      0
                                                                 35.09978 14
     SUBJ04
     SUBJ05
                          20.18760
                                                                 41.85877 51
                                                                 38.02936 37
     SUBJ06
                          25.70581
     trait
1 138.1346
2 113.3822
3 116.0071
4 127.5887
5 113.8754
6 110.7376
  dim(b2)
[1] 55 6
  b3 <- full_join(b, new, by = "subject_id")
full_join: added one column (trait)
           > rows only in x
                               2
           > rows only in y
           > matched rows
                              53
                                     (includes duplicates)
           > rows total
                              56
  dim(b3)
[1] 56 6
```

```
anti_join() return all rows from x without a match in y.
  anti_join(b, new, by = "subject_id")
anti_join: added no columns
           > rows only in x
                             2
           > rows only in y (1)
           > matched rows
                            (52)
                             ====
           > rows total
                               2
  subject_id maternal_age_delivery case_control_status prepregnancy_BMI ID
      SUBJ18
                          27.54075
                                                                44.14983 18
                                                      0
      SUBJ24
                          21.93645
                                                                31.73762 9
  anti_join(new, b, by = "subject_id")
anti_join: added no columns
           > rows only in x
           > rows only in y (2)
           > matched rows
                             (53)
                             ====
           > rows total
                             1
# A tibble: 1 x 2
  subject_id trait
  <chr>
             <dbl>
1 SUBJOO
              127.
```

11 R Graphics Exercise

11.1 Load Libraries

```
library(tidyverse)
library(ggforce)
# library(tidylog)
# Set the default font to be a bit larger:
theme_set(theme_gray(base_size = 18))
```

11.2 Exercise 1

Read in and set up the data set b, a cleaned version of our simulated data set:

```
a <- read.csv("data/study1.csv")
a$ind <- seq_along(a$t)
b <- a[-c(1001:1004),]
b$g.f <- factor(b$g)
b$geno <- paste(b$all1,b$all2,sep="/")</pre>
```

Using ggplot and data set b, plot ind vs. t, coloring by case-control status (trait). What do you observe about the data?

```
Expand to see solution

ggplot(data=b, aes(x=ind, y=t, color=trait)) +
    geom_point()
```



11.3 Exercise 2

Using ggplot, plot ind vs. t, coloring by case-control status (trait) and faceting by geno. What do you observe about the data?

```
Expand to see solution

ggplot(data=b, aes(x=ind, y=t, color=trait)) +
  geom_point() +
  facet_grid(~ geno)
```



11.4 Always plot your data

```
library(tidyverse)
  d <- read_tsv("data/example.tsv")</pre>
New names:
Rows: 142 Columns: 26
-- Column specification
----- Delimiter: "\t" dbl
(26): x...1, y...2, x...3, y...4, x...5, y...6, x...7, y...8, x...9, y....
i Use `spec()` to retrieve the full column specification for this data. i
Specify the column types or set `show_col_types = FALSE` to quiet this message.
* `x` -> `x...1`
* `y` -> `y...2`
* `x` -> `x...3`
 `y` -> `y...4`
 `x` -> `x...5`
 `y` -> `y...6`
  `x` -> `x...7`
 `y` -> `y...8`
* `x` -> `x...9`
```

```
* `y` -> `y...10`
* `x` -> `x...11`
* `y` -> `y...12`
* `x` -> `x...13`
* `y` -> `y...14`
* `x` -> `x...15`
* `y` -> `y...16`
* `x` -> `x...17`
* `y` -> `y...18`
* `x` -> `x...19`
* `y` -> `y...20`
* `x` -> `x...21`
* `y` -> `y...22`
* `x` -> `x...23`
* `y` -> `y...24`
* `x` -> `x...25`
* `y` -> `y...26`
  n1 \leftarrow rep(c("x","y"), 13)
  n2 <- c("","",rep("_",24))
  n3 \leftarrow c("", "", c(sort(rep(c(1:12), 2))))
  names(d) \leftarrow paste0(n1,n2,n3)
  names(d)
 [1] "x" "y" "x_1" "y_1" "x_2" "y_2" "x_3" "y_3" "x_4" "y_4"
[11] "x_5" "y_5" "x_6" "y_6" "x_7" "y_7" "x_8" "y_8" "x_9" "y_9"
[21] "x_10" "y_10" "x_11" "y_11" "x_12" "y_12"
```

11.5 Similar regression lines

These three data sets have very similar regression lines:

```
Estimate Std. Error t value Pr(>|t|)

(Intercept) 56.31108156 2.87906158 19.5588319 7.158847e-42
y_1 -0.04269949 0.05249244 -0.8134407 4.173467e-01

summary(lm(x_3 ~ y_3, data=d)) %>% coef()

Estimate Std. Error t value Pr(>|t|)

(Intercept) 56.18271411 2.87924135 19.5130270 9.107718e-42
y_3 -0.04012859 0.05249468 -0.7644316 4.458966e-01

ggplot(d,aes(x=x,y=y)) + geom_point() +
geom_smooth(method="lm") + ggtitle("Linear regression of y ~ x")
```

Linear regression of y ~ x



Now try this:

```
ggplot(d,aes(x=x_1,y=y_1)) + geom_point() +
  geom_smooth(method="lm")
```

Expand to see solution ggplot(d,aes(x=x_1,y=y_1)) + geom_point() + geom_smooth(method="lm") + ggtitle("Linear regression of y_1 ~ x_1") `geom_smooth()` using formula = 'y ~ x' Linear regression of y_1 ~ x_1 80 - 60 - 60 - 60 - 60 - 80 x_1

And now try this:

```
ggplot(d,aes(x=x_3,y=y_3)) + geom_point() +
geom_smooth(method="lm")
```

Expand to see solution 11.5.1 Always plot your data! ggplot(d,aes(x=x_3,y=y_3)) + geom_point() + geom_smooth(method="lm") + ggtitle("Linear regression of y_3 ~ x_3")

`geom_smooth()` using formula = 'y ~ x'



11.6 Always plot your data

Delete the first column

f <- read_tsv("data/BoxPlots.tsv")</pre>

5 -9.76 -9.77 -9.6 -8.82 -9.77 6 -9.77 -9.76 -9.56 -8.07 -9.76

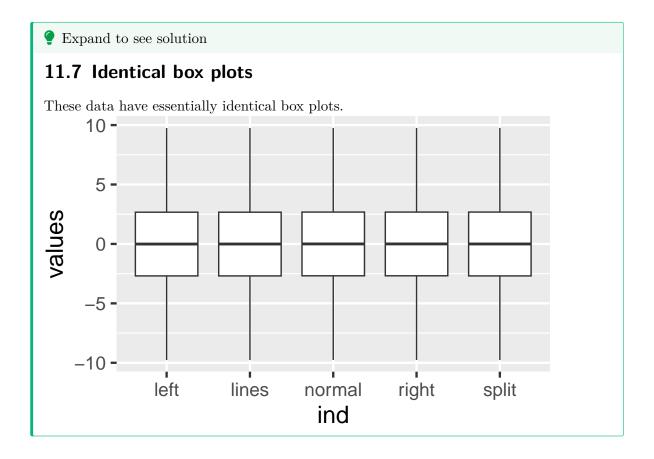
Stacking vectors concatenates multiple vectors into a single vector along with a factor indicating where each observation originated.

```
head(stack(f),2)

values ind
1 -9.769107 left
2 -9.763145 left

Now try this:

ggplot(stack(f), aes(x = ind, y = values)) +
   geom_boxplot()
```



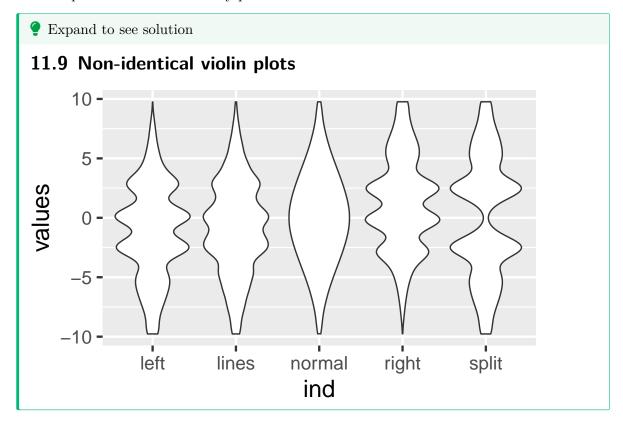
11.8 Boxplots

While the box plots are identical, box plots may not tell the whole story.

Let's try violin plots instead:

```
ggplot(stack(f), aes(x = ind, y = values)) +
  geom_violin()
```

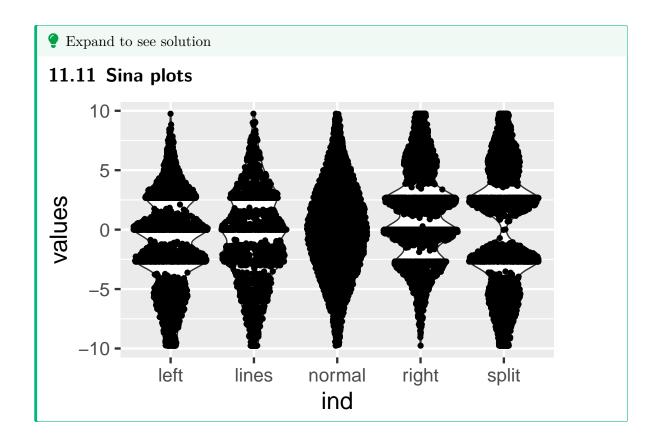
A violin plot is a mirrored density plot.



11.10 Sina plots

Sidiropoulos N, Sohi SH, Pedersen TL, Porse BT, Winther O, Rapin N, Bagger FO. SinaPlot: An Enhanced Chart for Simple and Truthful Representation of Single Observations Over Multiple Classes. Journal of Computational and Graphical Statistics. Taylor & Francis; 2018 Jul 3;27(3):673–676. DOI: https://doi.org/10.1080/10618600.2017.1366914

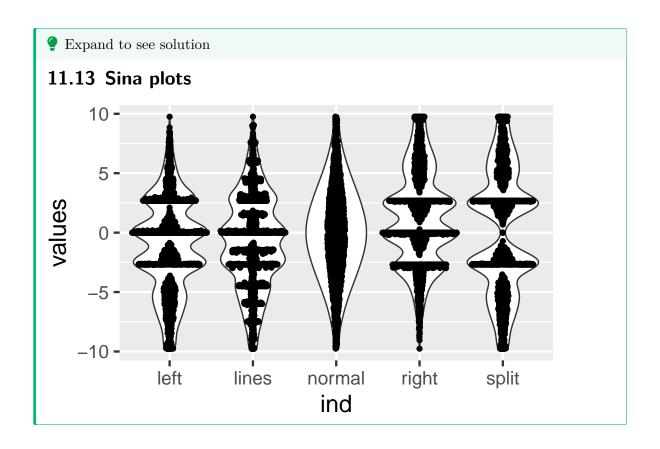
```
library(ggforce)
ggplot(stack(f), aes(x = ind, y = values)) +
    geom_violin() + geom_sina()
```



11.12 Sina plots

 ${\tt method} == {\tt "counts"}$: The borders are defined by the number of samples that occupy the same bin.

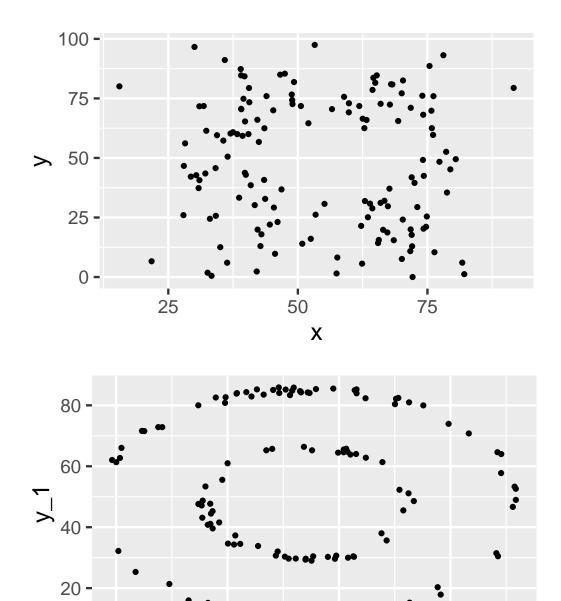
```
ggplot(stack(f), aes(x = ind, y = values)) +
  geom_violin() + geom_sina(method="count")
```



11.14 Drawing multiple graphs

Sometimes we'd like to draw multiple plots, looping across variables. Doing this within an R Markdown or Quarto Markdown document using ggplot2 is tricky. See https://dplyr.tidyverse.org/articles/programming.html and https://r4ds.hadley.nz/functions.html#plot-functions for details.

Here's one way to do this - this example code will generate two scatter plots:



11.15 Writing ggplot functions

See https://r4ds.hadley.nz/functions.html#plot-functions

x_1

```
library(palmerpenguins)

PlDensity <- function(fill, ...) {
    ggplot(penguins %>% filter(!is.na(bill_length_mm)),
        aes(bill_length_mm, fill = {{ fill }})) +
    geom_density(alpha = 0.5) +
    facet_wrap(vars(...))
}
```

 $\label{lem:example from: https://twitter.com/yutannihilat_en/status/1574387230025875457?s=20\&t=FLbwErwEKQKWtKIGufDLIQ$

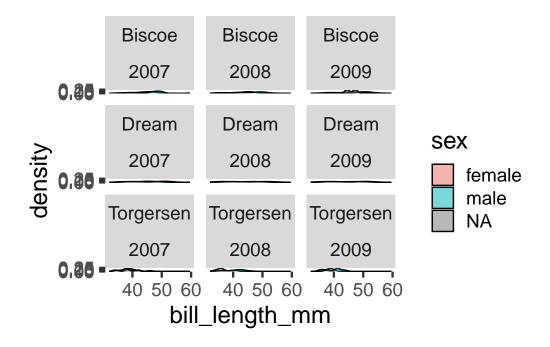
PlDensity(species)



PlDensity(island, sex) %>% print() %>% suppressWarnings()



PlDensity(sex, island, year) %>% print() %>% suppressWarnings()



11.16 Exercise 3

Consider this example code:

```
histogram <- function(df, var, binwidth) {
   df |>
      ggplot(aes({{ var }})) +
      geom_histogram(binwidth = binwidth)
}
```

 $From: \ https://twitter.com/hadleywickham/status/1574373127349575680?s = 20\&t = FLbwErwEKQKWtKIGufDLIQ$

When applied to the quantitative trait t from the data frame b, this generates this histogram:

```
histogram(b, t, 0.1)
```



11.16.1 Exercise

After reading the example above, extend the histogram function to allow facetting and use it to draw a histogram of the quantitative trait t facetted by geno using the data set b that we set up above.

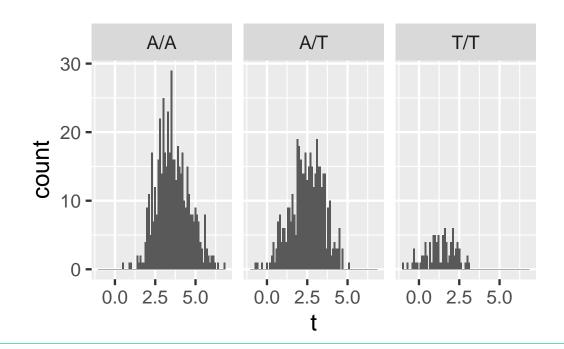
Hints

- $\bullet \ \ See \ https://r4ds.hadley.nz/functions.html\#plot-functions$
- Use the {{ var }} approach

```
? Expand to see solution
Hadley Wickham states:
You have to use the vars() syntax
foo <- function(x) {</pre>
 ggplot(mtcars) +
   aes(x = mpg, y = disp) +
   geom_point() +
   facet_wrap(vars({{ x }}))
}
LbwErwEKQKWtKIGufDLIQ\\
  histogram <- function(df, var, binwidth, grp) {</pre>
    df |>
      ggplot(aes({{ var }})) +
     geom_histogram(binwidth = binwidth) +
     facet_wrap(vars({{ grp }}))
  }
  histogram(b, t, 0.1)
```



histogram(b, t, 0.1, geno)



11.17 Source of data

 $Illustrative\ data\ sets\ from\ https://www.research.autodesk.com/publications/same-stats-different-graphs/$

12 R Reordering Exercise

12.1 Load Libraries

```
library(tidyverse)
library(tidylog)
```

12.2 Create some example data

Here we set up a data dictionary dd and some corresponding data ds. However, it is better if the order of the rows in the data dictionary dd match the order of the columns in the data ds.

```
set.seed(1562345)
  # Set up a data dictionary
  dd <- data.frame(VARNAME = sample(letters, 26), TYPE = "numeric")</pre>
  # Set up data
  ds <- as.data.frame(t(dd %>%
       arrange(VARNAME)))
  names(ds) <- letters</pre>
  rownames(ds) <- NULL
  ds[1, ] <- rnorm(26)
  ds[2, ] <- runif(26)
  ds$ID <- c(1, 2)
  ds <- ds %>%
       select(ID, everything())
select: columns reordered (ID, a, b, c, d, ...)
  # Randomly rearrange the columns
  idx <- sample(letters, 26)</pre>
  idx \leftarrow c("ID", idx)
  ds <- ds %>%
```

```
select(all_of(idx))
select: columns reordered (ID, b, z, a, p, ...)
  dd <- bind_rows(dd, data.frame(VARNAME = "ID", TYPE = "string"))</pre>
  dim(dd)
[1] 27 2
  head(dd)
 VARNAME
            TYPE
       c numeric
       m numeric
3
       f numeric
4
       e numeric
       a numeric
       d numeric
  dim(ds)
[1] 2 27
  head(ds[1:3])
 1 1.02333343074042 0.47956883003516
2 2 0.858655267162248 0.136965574463829
  names(ds)
 [1] "ID" "b" "z" "a" "p" "f" "u" "m" "q" "n" "d" "o" "s" "k" "e"
[16] "x" "c" "h" "i" "g" "j" "r" "t" "y" "l" "w" "v"
```

12.3 Task: Reorder rows in dd in the order of ds's columns

```
colnames(ds)

[1] "ID" "b" "z" "a" "p" "f" "u" "m" "q" "n" "d" "o" "s" "k" "e" [16] "x" "c" "h" "i" "g" "j" "r" "t" "y" "l" "w" "v" 

dd$VARNAME

[1] "c" "m" "f" "e" "a" "d" "v" "h" "k" "t" "p" "j" "l" "x" "w" [16] "y" "b" "o" "s" "r" "i" "z" "u" "n" "g" "q" "ID"
```

This assumes that every row of dd is in colnames(ds) and every colnames(ds) value is represented in dd. Perhaps that should be checked first.

12.4 Assumption Check Question

How would you check that every variable listed in the data dictionary dd is named in colnames(ds) and every colnames(ds) value is represented in the data dictionary dd?

```
Expand to see solution

table(dd$VARNAME %in% colnames(ds))

TRUE
27

table(colnames(ds) %in% dd$VARNAME)

TRUE
27

Note that we should also check to see if the VARNAME's are unique and the colnames of ds are unique.

sum(duplicated(dd$VARNAME))

[1] 0
```

```
sum(duplicated(colnames(ds)))
[1] 0
```

12.5 Task: Reorder rows in dd to match the order of the columns in ds

Task: Reorder rows in the data dictionary dd to match the order of the columns in the data ds

• What are various ways you could rearrange the rows of a data frame?

```
# Expand to see solution

# Assign VARNAME to be the rownames of dd
rownames(dd) <- dd$VARNAME
# Rearrange by row names:
dd2 <- dd[colnames(ds), ]
# Check if this worked:
all.equal(dd2$VARNAME, colnames(ds))

[1] TRUE

We can use match also:

# match returns a vector of the positions of (first) matches of its first
# argument in its second.
dd3 <- dd[match(colnames(ds), dd$VARNAME), ]
# Check if this worked:
all.equal(dd3$VARNAME, colnames(ds))</pre>
[1] TRUE
```

12.6 Question: use arrange?

Question: Is there a way to do this using arrange?

Expand to see the first attempt This does not work, because tidyverse wants to work on columns of data within dd: dd4 <- dd %>% arrange(colnames(ds)) # Check if this worked: all.equal(dd4\$VARNAME, colnames(ds)) [1] "26 string mismatches"

12.7 Question: use arrange?

Question: Is there a way to do this using arrange?

arrange() orders the rows of a data frame by the values of selected columns.

```
Expand to see solution

dd4 <- dd %>%
    mutate(neworder = match(.$VARNAME, colnames(ds))) %>%
    arrange(neworder) %>%
    select(-neworder)

mutate: new variable 'neworder' (integer) with 27 unique values and 0% NA
select: dropped one variable (neworder)

all.equal(dd4$VARNAME, colnames(ds))

[1] TRUE
```

12.8 Question: use slice

Question: Is there a way to do this using the slice command? slice() lets you index rows by their (integer) locations.

```
Expand to see solution

dd6 <- dd %>%
    slice(match(colnames(ds), .$VARNAME))

slice: no rows removed

all.equal(dd6$VARNAME, colnames(ds))

[1] TRUE
```

12.9 Question: use select?

Question: Is there a way to do this by transposing and then using select?

12.10 Question: use row names

Question: What about using row names?

"While a tibble can have row names (e.g., when converting from a regular data frame), they are removed when subsetting with the [operator. A warning will be raised when attempting to assign non-NULL row names to a tibble. Generally, it is best to avoid row names, because they are basically a character column with different semantics than every other column."

From: https://tibble.tidyverse.org/reference/rownames.html

13 R Exploratory Data Analysis Exercise

13.1 Load Libraries

```
library(tidyverse)
library(tidylog)
library(DataExplorer)
library(GGally)
```

13.2 Explore Project 1 data

Let's explore the Project 1 data set:

```
load("data/project1.RData", verbose = TRUE)

Loading objects:
    ds
    dd

          ds = data set
          dd = data dictionary
```

13.3 Dimensions

• What are the dimensions of our data?

13.4 Dimensions

Task: Examine the dimensions of our data and data dictionary.

13.4.1 Data ds

```
dim(ds)
[1] 191 24
  names(ds)
 [1] "sample_id"
                                 "Sample_trimester"
 [3] "Gestationalage_sample"
                                 "subject_id"
 [5] "strata"
                                 "race"
 [7] "maternal_age_delivery"
                                 "case_control_status"
 [9] "prepregnancy_weight"
                                 "height"
                                 "gravidity"
[11] "prepregnancy_BMI"
[13] "parity"
                                 "gestationalage_delivery"
[15] "average_SBP_lt20weeks"
                                 "average_DBP_lt20weeks"
[17] "average_SBP_labor"
                                 "average_DBP_labor"
                                 "baby_birthweight"
[19] "smoke_lifetime"
[21] "baby_sex"
                                 "baby_birthweight_centile"
[23] "baby_SGA"
                                 "placental_pathology"
```

13.4.2 Data dictionay dd

```
dim(dd)
[1] 27 5

names(dd)

[1] "Original.Variable.Name" "R21.Variable.Name" "Description"
[4] "Variable.Units" "Variable.Coding"
```

13.5 Arrangement

- How are the data arranged?
 - Is it in tidy format?
 - Is it one row per sample or per subject?
 - Were subjects sampled more than once?

13.5.1 Samples or subjects

Is it one row per sample or per subject?

Question: How would you figure out the answer to this question?

```
Expand to see solution

sum(duplicated(ds$sample_id))

[1] 72

length(unique(ds$sample_id))

[1] 119

length(unique(ds$subject_id))

[1] 54
```

13.5.2 Unique values

Question: How can we figure out the number of unique values in each column of our ds data frame?

```
? Expand to see solution
  sapply(ds, function(x) {
       length(unique(x))
  }) %>%
       kable()
                                                        \mathbf{X}
                           sample\_id
                                                      119
                           Sample_trimester
                                                        4
                           Gestationalage_sample
                                                      189
                           subject_id
                                                       54
                                                       21
                           strata
```

```
race
                             3
maternal_age_delivery
                            54
case_control_status
                             2
prepregnancy_weight
                            51
height
                            42
                            54
prepregnancy BMI
gravidity
                             5
                             4
parity
gestationalage_delivery
                            54
average\_SBP\_lt20weeks
                            19
average\_DBP\_lt20weeks
                            16
average\_SBP\_labor
                            23
average DBP labor
                            27
smoke\_lifetime
                             2
baby_birthweight
                            30
                             2
baby_sex
baby\_birthweight\_centile
                            52
baby_SGA
                             1
placental pathology
                             2
```

13.5.3 Subject-level data set

Task: Construct a subject-level data set

How would you construct a subject-level data set?

```
Expand to see solution

ds.subj <- ds %>%
    select(-sample_id, -Sample_trimester, -Gestationalage_sample) %>%
    distinct()

select: dropped 3 variables (sample_id, Sample_trimester, Gestationalage_sample)
distinct: removed 136 rows (71%), 55 rows remaining

sum(duplicated(ds.subj$subject_id))

[1] 1
```

```
ds.subj %>%
      group_by(subject_id) %>%
      filter(n() > 1)
group_by: one grouping variable (subject_id)
filter (grouped): removed 53 rows (96%), 2 rows remaining
# A tibble: 2 x 21
            subject_id [1]
# Groups:
  subject_id strata race matern~1 case_~2 prepr~3 height prepr~4 gravi~5 parity
  <chr>
              <dbl> <chr>
                             <dbl>
                                     <dbl>
                                             <dbl> <dbl>
                                                             <dbl>
                                                                     <dbl>
1 SUBJ20
                 35 W
                              29.4
                                         1
                                              244.
                                                      63.1
                                                              44.0
                                                                         1
                                                                                0
2 SUBJ20
                 35 White
                              29.4
                                         1
                                              244.
                                                      63.1
                                                              44.0
                                                                         1
                                                                                0
# ... with 11 more variables: gestationalage_delivery <dbl>,
    average_SBP_lt20weeks <dbl>, average_DBP_lt20weeks <dbl>,
    average_SBP_labor <dbl>, average_DBP_labor <dbl>, smoke_lifetime <chr>,
    baby_birthweight <dbl>, baby_sex <chr>, baby_birthweight_centile <dbl>,
    baby_SGA <chr>, placental_pathology <chr>, and abbreviated variable names
    1: maternal_age_delivery, 2: case_control_status, 3: prepregnancy_weight,
    4: prepregnancy_BMI, 5: gravidity
  ds.subj <- ds.subj %>%
      filter(race != "White")
filter: removed one row (2%), 54 rows remaining
  sum(duplicated(ds.subj$subject_id))
[1] 0
```

13.6 Coding

- How are the data coded?
 - Are they coded correctly?
 - Which are categorical and which are continuous?
 - Are they coded consistently with the data dictionary?
 - Is there a data dictionary?

- Do we need to skip rows when reading the data in?

13.6.1 Recode for understandability

Let's recode case_control_status from 0 and 1 into a new PE_status variable coded as control and case.

```
dd %>%
    filter(R21.Variable.Name == "case_control_status") %>%
    pull(Variable.Coding)

filter: removed 26 rows (96%), one row remaining

[1] "0: normotensive control; 1: preeclampsia case"
```

Task: recode case_control_status from 0 and 1 into a new PE_status variable coded as control and case.

13.7 Missing data

- What is the pattern of missing data?
 - How are missing data coded?

- Is there a single missing data code?

Here we use some plotting commands from the DataExplorer R package.

https://boxuancui.github.io/DataExplorer/index.html

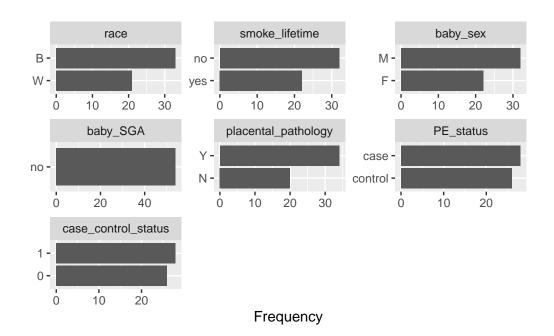


13.8 Distribution

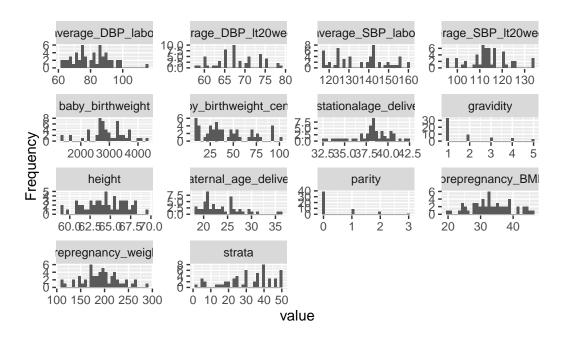
- What is the distribution of each of our phenotypes?
 - Are data skewed?
 - What is the range of values?
 - Is the range of values realistic?

```
plot_bar(ds.subj)
```

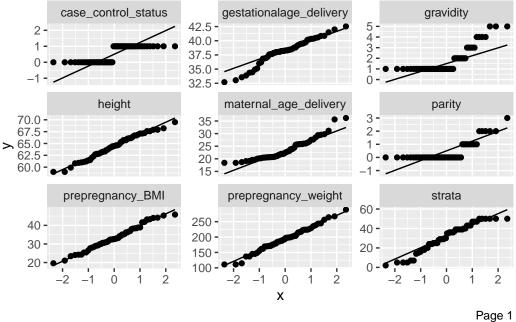
1 columns ignored with more than 50 categories. subject_id: 54 categories

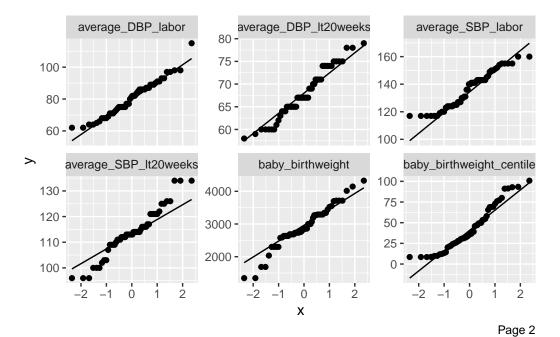


plot_histogram(ds.subj)



plot_qq(ds.subj)





13.9 Variation

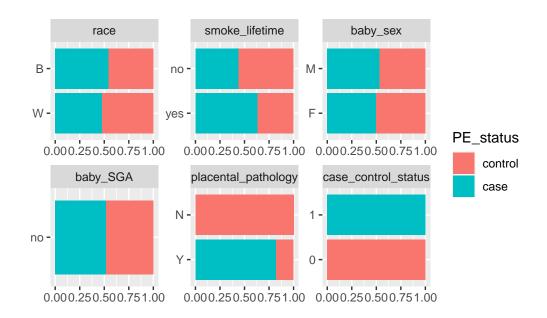
• How do our data vary and co-vary?

- Do multiple measures agree with each other?
- Are there sex-specific or age-specific differences?

13.9.1 Bar plots

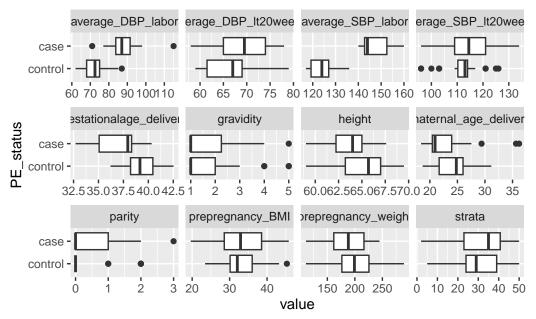
```
plot_bar(ds.subj, by = "PE_status")
```

1 columns ignored with more than 50 categories. subject_id: 54 categories



13.9.2 Box plots

```
plot_boxplot(ds.subj, by = "PE_status")
```



Page 1

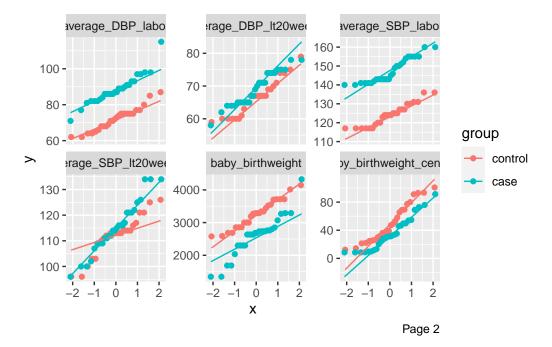


Page 2

13.9.3 QQ plots

plot_qq(ds.subj, by = "PE_status")



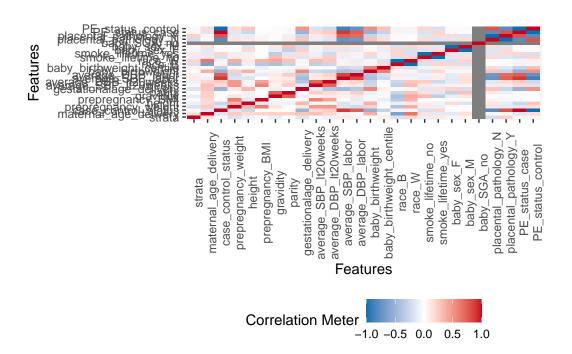


13.9.4 Correlation

```
plot_correlation(ds.subj)
```

1 features with more than 20 categories ignored! subject_id: 54 categories

Warning in cor(x = structure(list(strata = c(29, 39, 29, 36, 40, 39, 40, : the standard deviation is zero

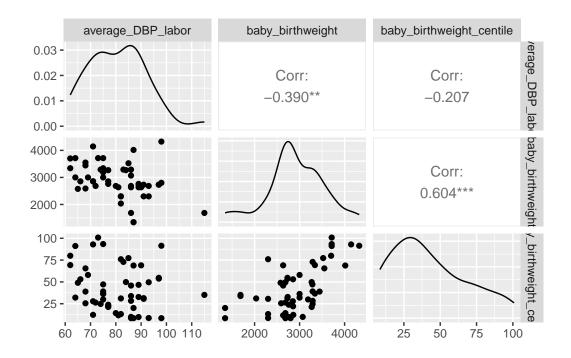


13.9.5 ggpairs from the GGally R package.

Use ggpairs from the GGally R package.

```
# Pull out numeric columns
ds1 <- ds.subj[, sapply(ds.subj, is.numeric)]

ggpairs(ds1[, c(13:15)])</pre>
```



ggpairs - color by ggplot2 aes

ggpairs(ds.subj, columns = c(15, 17, 19), ggplot2::aes(color = PE_status))



```
ggcorr(ds1[, c(13:15)], label = TRUE)
```

baby_birthweight_cen



13.10 DataExplorer

We can quickly create a report using the ${\tt create_report}$ function from the ${\tt DataExplorer}$ R package

create_report(ds.subj)

14 Basic Shell Commands

14.1 Acknowledgment and License

This chapter is a derivative of the Basic Shell Commands cheat sheet from the DEPRECATED-boot-camps/shell/shell_cheatsheet.md file created by Software Carpentry and is used under the Creative Commons - Attribution license CC BY 3.0

Minor section numbering and formatting changes were made here.

This chapter is licensed under the CC BY 3.0 license by Daniel E. Weeks.

14.2 Shell Basics:

Command	Definition
•	a single period refers to the current directory
	a double period refers to the directory immediately above the current directory
~	refers to your home directory. Note: this command does NOT work on
	Windows machines (Mac and Linux are okay)
cd	changes the current directory to the directory dirname
./dirname	
ls -F	tells you what files and directories are in the current directory
pwd	tells you what directory you are in (pwd stands for print working directory)
history	lists previous commands you have entered. history less lets you page
J	through the list.
$\mathtt{man}\ cmd$	displays the manual page for a command.

14.3 Creating Things:

14.3.1 How to create new files and directories...

Commandinition

mkdir makes a new directory called dirname below the current directory. *Note:* Windows ./dirnamers will need to use \ instead of / for the path separator nano if filename does not exist, nano creates it and opens the nano text editor. If the file

filename does not exist, nano creates it and opens the nano text editor. If the file filenameists, nano opens it. Note: (i) You can use a different text editor if you like. In gnome Linux, gedit works really well too. (ii) nano (or gedit) create text files. It doesn't matter what the file extension is (or if there is one)

14.3.2 How to delete files and directories...

14.3.2.1 Remember that deleting is forever. There is NO going back

Command	Definition
rm	deletes a file called filename from the current directory
./filename	
rmdir	deletes the directory dirname from the current directory. Note: dirname
./dirname	must be empty for rmdir to run.

14.3.3 How to copy and rename files and directories...

Command Definition

mv moves the file filename from the directory tmp to the current directory. Note: (i) tmp/filentalme original filename in tmp is deleted. (ii) mv can also be used to rename files . (e.g., mv filename newname

cp copies the file filename from the directory tmp to the current directory. Note: (i) tmp/filename original file is still there

14.4 Pipes and Filters

14.4.1 How to use wildcards to match filenames...

Wildcards are a shell feature that makes the command line much more powerful than any GUI file managers. Wildcards are particularly useful when you are looking for directories, files, or file content that can vary along a given dimension. These wildcards can be used with any command that accepts file names or text strings as arguments.

14.4.1.1 Table of commonly used wildcards

Wildcard	Matches
*	zero or more characters
?	exactly one character
[abcde]	exactly one of the characters listed
[a-e]	exactly one character in the given range
[!abcde]	any character not listed
[!a-e]	any character that is not in the given range
{software,carpentry}	exactly one entire word from the options given

See the cheatsheet on regular expressions on the second page of this PDF cheatsheet for more "wildcard" shortcuts.

14.4.2 How to redirect to a file and get input from a file ...

Redirection operators can be used to redirect the output from a program from the display screen to a file where it is saved (or many other places too, like your printer or to another program where it can be used as input).

Commandescription

- > write stdout to a new file; overwrites any file with that name (e.g., ls *.md >
 mardkownfiles.txt)
- >> append stdout to a previously existing file; if the file does not exist, it is created (e.g., ls *.md >> markdownfiles.txt)
- < assigns the information in a file to a variable, loop, etc (e.g., n <
 markdownfiles.md)</pre>

14.4.2.1 How to use the output of one command as the input to another with a pipe...

A special kind of redirection is called a pipe and is denoted by |.

Commandescription

Output from one command line program can be used as input to another one (e.g. ls *.md | head gives you the first 5 *.md files in your directory)

14.4.2.1.1 Example:

```
ls *.md | head | sed -i `s/markdown/software/g`
```

changes all the instances of the word markdown to software in the first 5 *.md files in your current directory.

14.5 How to repeat operations using a loop...

Loops assign a value in a list or counter to a variable that takes on a different value each time through the loop. There are 2 primary kinds of loops: for loops and while loops.

14.5.1 For loop

For loops loop through variables in a list

```
for varname in list
do
    command1 $varname
    command2 $varname
done
```

where,

- for, in, do, and done are keywords
- list contains a list of values separated by spaces. e.g. list can be replaced by 1 2 3 4 5 6 or by Bob Mary Sue Greg. list can also be a variable:
- varname is assigned a value without using a \$ and the value is retrieved using \$varname

```
list[0]=Sam
list[1]=Lynne
list[2]=Dhavide
list[3]=Trevor
.
.
.
.
list[n]=Mark
```

which is referenced in the loop by:

```
for varname in ${list[@]}
do
          command1 $varname
          command2 $varname
done
```

Note: Bash is zero indexed, so counting always starts at 0, not 1.

14.5.2 While Loop

While loops loop through the commands until a condition is met. For example

```
COUNTER=0
while [ ${COUNTER} -lt 10 ]; do
    command 1
    command 2
    COUNTER=`expr ${COUNTER} + 1`
done
```

continues the loop as long as the value in the variable COUNTER is less than 10 (incremented by 1 on each iteration of the loop).

• while, do, and done are keywords

14.5.2.1 Commonly used conditional operators

Operator	Definition
-eq	is equal to
-ne	is not equal to
-gt	greater than
-ge	greater than or equal to
-lt	less than
-le	less than or equal to

Use man bash or man test to learn about other operators you can use.

14.6 Finding Things

14.6.1 How to select lines matching patterns in text files...

To find information within files, you use a command called grep.

Example command	Description
grep [options] day haiku.txt	finds every instance of the string day in the file haiku.txt and pipes it to standard output

14.6.1.1 Commonly used grep options

grep options

- -E tells grep you will be using a regular expression. Enclose the regular expression in quotes. *Note:* the power of grep comes from using regular expressions. Please see the regular expressions sheet for examples
- -i makes matching case-insensitive
- -n limits the number of lines that match to the first n matches
- -v shows lines that do not match the pattern (inverts the match)
- -w outputs instances where the pattern is a whole word

14.6.2 How to find files with certain properties...

To find file and directory names, you use a command called find

Example	
com-	
mand	Description
find .	find recursively descends the directory tree for each path listed to match the expression given in the command line with file or directory names in the search path

14.6.2.1 Commonly used find options

find options -type d lists directories; f lists files [df] -maxdepthfind automatically searches subdirectories. If you don't want that, specify the n number of levels below the working directory you would like to search -mindepthstarts find's search n levels below the working directory n

15 Summary

In summary, this book is a work in progress.

16 WebR - R in the web browser

This is a WebR-enabled code cell in a Quarto HTML document. As the WebR documentation states: "WebR makes it possible to run R code in the browser without the need for an R server to execute the code: the R interpreter runs directly on the user's machine."

```
# Edit/add code here
fit = lm(mpg ~ am, data = mtcars)
summary(fit)
```

Link: WebR.

17 Technical Details

17.1 Quarto

This book was build using Quarto.

17.1.1 Callout blocks

To hide a solution that then can be clicked to view, we use a .callout-tip collapse="true" callout block.

Here are some examples from the Quarto documentation:

Note

Note that there are five types of callouts, including: note, tip, warning, caution, and important.

⚠ Warning

Callouts provide a simple way to attract attention, for example, to this warning.

I This is Important

Danger, callouts will really improve your writing.

Tip With Title

This is an example of a callout with a title.

Expand To Learn About Collapse

This is an example of a 'collapsed' caution callout that can be expanded by the user. You can use collapse="true" to collapse it by default or collapse="false" to make a collapsible callout that is expanded by default.

17.1.2 Adding a chapter

To add a new chapter to the book, make a Quarto file containing the chapter text and code. It should have only one top-level header at the beginning which will be the title of the chapter.

Then add it to the list of chapters in the _quarto.yml file.

17.2 Previewing the book

Type quarto preview in the Terminal window.

17.3 Deploying the book to GitHub Pages

Type quarto publish in the Terminal window.

17.4 Deploying the book to Netlify

Type quarto publish netlify in the Terminal window.

17.5 WebR: R in the browser

This Quarto book uses this WebR Quarto extension

https://github.com/coatless/quarto-webr

WebR makes installs a version of R that runs within the browser, and the Quarto extension makes it interactively available in web-r chunks.

To get this to work, the _quarto.yml had to be modified.

We added a 'resources' directive to copy over the java script files, which places them next to the 'index.html' file during deployment of the book:

```
project:
   type: book
   resources:
        - "webr-serviceworker.js"
        - "webr-worker.js"
```

We also enabled the webr filter:

filters:

- webr

References