

HuGen2071 book

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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 and Jonathan Chernus

Websites:

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

<https://www.sph.pitt.edu/directory/jonathan-chernus>

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 and Jonathan Chernus

Websites:

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

<https://www.sph.pitt.edu/directory/jonathan-chernus>

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 and genetic/genomic data
- Describe dbGaP, an important genomic data repository

4.1.2 Required Reading

Mailman MD, Feolo M, Jin Y, Kimura M, Tryka K, Bagoutdinov R, Hao L, Kiang A, Paschall J, Phan L, Popova N, Pretel S, Ziyabari L, Lee M, Shao Y, Wang ZY, Sirotkin K, Ward M, Kholodov M, Zbicz K, Beck J, Kimelman M, Shevelev S, Preuss D, Yaschenko E, Graeff A, Ostell J, Sherry ST. The NCBI dbGaP database of genotypes and phenotypes. Nat Genet. 2007 Oct;39(10):1181-6. doi: 10.1038/ng1007-1181. PMID: 17898773; PMCID: PMC2031016. <https://pubmed.ncbi.nlm.nih.gov/17898773/>

4.1.3 Suggested 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 Online Lecture

GitHub Introduction: <https://danieleweeks.github.io/HuGen2071/gitIntro.html>

4.2.3 Active Learning

Version Control with git and GitHub (Sections 4.1 - 4.4): <https://learning.nceas.ucsb.edu/2020-11-RRCourse/session-4-version-control-with-git-and-github.html>

4.2.4 Required Readings

GitHub Classroom Guide for Students

To set up GitHub Classroom, please follow the steps to set up RStudio, R, and git in this detailed guide: <https://github.com/jfiksel/github-classroom-for-students>

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

Be sure to generate an SSH key so you don't need to enter your password every time you interact with GitHub.

Warning

Do not clone your repository onto a OneDrive or other cloud folder, as git does not work properly on cloud drives. Cloud drive systems typically maintain their own backup copies and this confuses git.

4.2.5 Suggested 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>

Version Control with Git: <https://swcarpentry.github.io/git-novice/>

Using Git from RStudio: <https://ucsbcarpentry.github.io/2020-08-10-Summer-GitBash/24-supplemental-rstudio/index.html>

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 Online Lectures

R Basics: <https://danieleweeks.github.io/HuGen2071/RBasicsLecture.html>

4.3.3 Active Learning:

<https://datacarpentry.org/genomics-r-intro/01-r-basics.html>

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

4.4.2 Online Lecture

R: factors, subscripting: <https://danieleweeks.github.io/HuGen2071/RFactors.html>

4.4.3 Active Learning:

Subsetting: <https://swcarpentry.github.io/r-novice-gapminder/06-data-subsetting.html>. This uses the gapminder data from [here](#).

Factors: <https://swcarpentry.github.io/r-novice-inflammation/12-supp-factors.html>. This uses data from this [Zip file](#).

4.4.4 Suggested 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_vlebooks_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 Active Learning

Regular expressions: <https://csiro-data-school.github.io/regex/08-r-regexs/index.html>

4.5.3 Required Readings

Read the chapter on “Strings” in “R for Data Science”: <https://r4ds.hadley.nz/strings>

4.5.4 Suggested Readings

See the “String manipulation with stringr cheatsheet” at <https://rstudio.github.io/cheatsheets/html/strings.html>

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

Read the ‘Working with Strings’ section at the end of the “Working with and Visualizing Data in R” section.

https://pitt.primo.exlibrisgroup.com/permalink/01PITT_INST/i25aoe/cdi_askewsholts_vlebooks_9781449367510

Read the chapter on “Strings” in “R for Data Science”: <https://r4ds.hadley.nz/strings>

Read the chapter on “Regular expressions” in “R for Data Science”: <https://r4ds.hadley.nz/regular-expressions>

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 Online Lectures

Loops in R: <https://danieleweeks.github.io/HuGen2071/RLoops.html>

4.6.3 Active Learning:

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

Loops in R, Part I: <https://danieleweeks.github.io/HuGen2071/loops.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 Suggested 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://datacarpentry.org/genomics-r-intro/05-dplyr.html>

The data file used in this is the `combined_tidy_vcf.csv` file that can be downloaded from [here](#).

4.8.3 Suggested 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:

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

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 Suggested Readings

Supplementary Reading: Spector (2008) Chapters 8 & 9

4.10 R: 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

`continents.RDA` data set used near the end of this Active Learning exercise: <https://mikoontz.github.io/data-carpentry-week/data/continents.RDA>

4.10.3 Required Reading

Tidy Animated Verbs <https://www.garrickadenbuie.com/project/tidyexplain/>

4.10.4 Suggested Readings

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

Data visualization with ggplot2: <https://datacarpentry.org/R-ecology-lesson/04-visualization-with-ggplot2.html>

To create the required data for this “Data visualization with ggplot2” exercise, run this code:

```
library(tidyverse)
download.file(url = "https://ndownloader.figshare.com/files/2292169",
              destfile = "portal_data_joined.csv")
surveys <- read_csv("portal_data_joined.csv")
surveys_complete <- surveys %>%
  filter(!is.na(weight),           # remove missing weight
         !is.na(hindfoot_length),  # remove missing hindfoot_length
         !is.na(sex))
```

4.11.3 Suggested 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 Active Learning

Exploratory analysis of RNAseq count data https://tavareshugo.github.io/data-carpentry-rnaseq/02_rnaseq_exploratory.html

4.12.3 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: Genomic Ranges; Interactive Graphics

4.13.1 Learning Objectives - Genomic Ranges

- To learn about Genomic Ranges
- To learn to use Genomic Ranges to annotate SNPs of interest

4.13.2 Preparation - Genomic Ranges

Before class, install these BioConductor packages: (1) `TxDb.Hsapiens.UCSC.hg19.knownGene`, and (2) `org.Hs.eg.db`

To install these, use these commands:

```
if (!requireNamespace("BiocManager", quietly = TRUE))
  install.packages("BiocManager")

BiocManager::install("TxDb.Hsapiens.UCSC.hg19.knownGene")

BiocManager::install("org.Hs.eg.db")
```

4.13.3 Required Reading - Genomic Ranges

An Introduction to Bioconductor's Packages for Working with Range Data

<https://github.com/vsbuffalo/genomicroranges-intro/blob/master/notes.md>

4.13.4 Active Learning - Genomic Ranges

Working with genomics ranges

<https://carpentries-incubator.github.io/bioc-project/07-genomic-ranges.html>

4.13.5 Suggested Readings - Genomic Ranges

In “Bioinformatics Data Skills”, see Chapter 9 “Working with Range Data”

Bioinformatics Data Skills

Editor: Vince Buffalo

Publisher: O'Reilly

Web access: [link](#)

Hello Ranges: An Introduction to Analyzing Genomic Ranges in R.

[link](#)

4.13.6 Learning Objectives - Interactive Graphics

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

4.13.7 Required Reading - Interactive Graphics

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

4.14 Suggested Reading - Interactive Graphics

Wickham (2009) Chapters 2 & 3

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

4.16.1 Learning Objectives - Unix: Basics

- To learn basic Unix commands

4.16.2 Preparation - Unix: Basics

- Do the Active Learning before class - the lecture will assume you have; otherwise you will have difficulty with the in-class exercises
- Make sure you've done the Unix setup homework assignment; for the in-class exercises you will need to connect to htc and use git there

4.16.3 Active Learning - Unix: Basics

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

4.16.4 Required Reading - Unix: Basics

See Active Learning.

4.16.5 Suggested Reading - Unix: Basics

[Buffalo \(2015\)](#) Chapter 2. Setting up and managing a bioinformatics project.

[Buffalo \(2015\)](#) Chapter 3. Remedial Unix Shell (beginning of chapter up to and not including “working with streams and redirection”)

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

“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

4.17 Unix: Streams, Pipes, Scripts

4.17.1 Learning Objectives - Unix: Streams, Pipes, Scripts

- To learn how streams operate in Unix
- To learn how to pass streamed data from program to program in Unix
- To learn how to interact with running processes
- To learn how to write a script that can run in Unix
- To learn about the cluster and how to submit jobs there

4.17.2 Preparation - Unix: Streams, Pipes, Scripts

- Do the Active Learning before class - the lecture will assume you have; otherwise you will have difficulty with the in-class exercises

4.17.3 Active Learning - Unix: Streams, Pipes, Scripts

Software Carpentry Unix Shell intro parts 4 and 6 <https://swcarpentry.github.io/shell-novice/>

4.17.4 Required Reading - Unix: Streams, Pipes, Scripts

See Active Learning.

4.17.5 Suggested Reading - Unix: Streams, Pipes, Scripts

[Buffalo \(2015\)](#) Chapter 3. Remedial Unix Shell (from “working with streams and redirection” to and not including “command substitution”)

4.18 Genetic Data Structures

4.18.1 Learning Objectives

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

4.18.2 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.

4.19 PLINK I

4.19.1 Learning Objectives

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

4.19.2 Readings

Marees AT, de Kluiver H, Stringer S, Vorspan F, Curis E, Marie-Claire C, Derkx 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/

4.20 PLINK II

4.20.1 Learning Objectives

- To learn how to use PLINK to manipulate data files

4.21 PLINK Computer Lab

4.21.1 Learning Objectives

- To practice using PLINK to manipulate data files

4.22 Unix: Data Manipulation

4.22.1 Learning Objectives - Unix: Data Manipulation

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

4.22.2 Preparation - Unix: Data Manipulation

See Required Reading.

4.22.3 Active Learning - Unix: Data Manipulation

See Required Reading.

4.22.4 Required Reading - Unix: Data Manipulation

[Buffalo \(2015\)](#) Chapter 7. Unix Data Tools (Beginning of chapter up to and including “Finding Unique values in Uniq”)

4.22.5 Suggested Reading - Unix: Data Manipulation

None.

4.23 Unix: Miscellaneous

4.23.1 Learning Objectives - Unix: Miscellaneous

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

4.23.2 Preparation - Unix: Miscellaneous

See Required Reading.

4.23.3 Active Learning - Unix: Miscellaneous

See Required Reading.

4.23.4 Required Reading - Unix: Miscellaneous

[Buffalo \(2015\)](#) Chapter 7. Unix Data Tools (“Join” through the end of the chapter)

4.23.5 Suggested Reading - Unix: Miscellaneous

None.

4.24 Unix: Scripting

4.24.1 Learning Objectives - Unix: Scripting

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

4.24.2 Preparation - Unix: Scripting

- Do the Active Learning before class - the lecture will assume you have; otherwise you will have difficulty with the in-class exercises

4.24.3 Active Learning - Unix: Scripting

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

4.24.4 Required Reading - Unix: Scripting

See Active Learning.

4.24.5 Suggested Reading - Unix: Scripting

[Buffalo \(2015\)](#) Chapter 3. Remedial Unix Shell (“command substitution” through the end of the chapter.)

[Buffalo \(2015\)](#) Chapter 12. Bioinformatics Shell Scripting (entire chapter)

4.25 VCF, bcftools, vcftools

4.25.1 Learning Objectives

- To learn about VCF data format
- To learn about bcftools and vcftools 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 - Genetic Data in R, GDS

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

4.27.2 Preparation - Genetic Data in R, GDS

See Required Reading.

4.27.3 Active Learning - Genetic Data in R, GDS

None. See Required Reading.

4.27.4 Required Reading - Genetic Data in R, GDS

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/>

4.27.5 Suggested Reading - Genetic Data in R, GDS

None

5 GitHub

5.1 GitHub Introduction lecture

Here's a recording of this lecture (32 minutes 8 seconds):

[Recording](#)

5.2 GitHub Introduction slides

[PDF slide set](#)

6 Git Commands

6.1 git - best practices

pull - work - commit - pull - push

- `git pull`
- Make changes
- `git commit` your changes to your local repository
- `git pull` the latest remote changes to your local repository
- `git push` your changes.

Pay attention to any error messages.

6.2 Outline of essential Git commands

Here's an outline of essential Git commands, initially created by ChatGPT:

6.2.1 Initialization and Configuration

- `git init`: Initializes a new Git repository in the current directory.
- `git config`: Configure Git settings.

6.2.2 Basic Workflow

- `git add`: Stage changes.
- `git commit -m "message"`: Commits staged changes with a descriptive message.

6.2.3 Remote Repositories

- `git clone`: Clones a remote repository to your local machine.
- `git push`: Send local changes to remote repository.
- `git pull`: Retrieve changes from remote.
- `git remote`: Manage remote repositories.

6.2.4 Status and Changes

- `git status`: Shows the current state of your working directory.
- `git diff`: Displays changes between working directory and the last commit.

6.2.5 History and Logs

- `git log`: View commit history.
- `git log --oneline`: Compact commit history.

6.2.6 Ignoring Files

- Create `.gitignore` file.

6.2.7 Branching

- `git branch`: List/create branches.
- `git checkout`: Switch branches.
- `git merge`: Merge branches.

6.2.8 Undoing Changes

- `git reset`: Unstage or reset changes.
- `git revert`: Create undoing commits.

6.2.9 Tagging

- `git tag`: Create and manage tags.

6.2.10 Stashing

- `git stash`: Temporarily store changes.

7 Lecture: R Basics

7.1 R Basics lecture

Here's a recording of this lecture (48 minutes 14 seconds):

[Recording](#)

7.2 R Basics slides

[PDF slide set](#)

8 R Basics Group Exercise

8.1 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
4	4	8	12

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

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

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.

8.2 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 <- data.frame(n = 1:4)  
dim(a)
```

```
[1] 4 1
```

```
a
```

```
n  
1 1  
2 2  
3 3  
4 4
```

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 <- data.frame(n = 1:4))  
# Edit/add R code here
```

💡 Tip

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  
3 3      1  
4 4      2
```

8.3 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.

8.4 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 <- 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
a
```

```
n rowNum1 rowNum6
1 1      1      1
2 2      2      2
3 3      1      1
4 4      2      2
```

8.5 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))
```

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

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
```



Tip

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
2	2	2	2
3	3	1	1
4	4	2	2

8.6 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"
```

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

  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 1      1      1      1
2 2      2      2      2
3 3      1      1      1
4 4      2      2      2
```

8.7 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  
  }  
}  
a  
  
n rowNum1 rowNum6 rowNum2 rowNum3 rowNum4  
1 1 1 1 1 1  
2 2 2 2 2 2  
3 3 1 1 1 1  
4 4 2 2 2 2
```

8.8 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)
a

n rowNum1 rowNum6 rowNum2 rowNum3 rowNum4 rowNum5
1 1      1      1      1      1      1      1
2 2      2      2      2      2      2      2
3 3      1      1      1      1      1      1
4 4      2      2      2      2      2      2
```

8.9 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
4 4      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
```

8.10 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"
# Adjust the read.table options to read the file correctly as desired.
b <- read.table(infile)
b
```

```
V1 V2 V3
1 1 1 1
2 2 2 2
3 3 NA 99
4 4 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 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.txt"
download.file(srcfile, infile)
# Adjust the read.table options to read the file correctly as desired.
b <- read.table(infile)
b
```



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
1 1 1 1
2 2 2 2
3 3 NA NA
4 4 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          V2          V3    
Min. :1.000   Min. :1.000   Min. :1.000
```

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 :1	NA's :1	NA's :1

9 Lecture: R: factors, subscripting

9.1 R: factors, subscripting lecture

Here's a recording of this lecture (43 minutes 25 seconds):

[Recording](#)

9.2 R: factors, subscripting slides

[PDF slide set](#)

10 R Character Exercise

10.1 Load Libraries

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

10.2 Useful RStudio cheatsheet

See the “String manipulation with stringr cheatsheet” at
<https://rstudio.github.io/cheatsheets/html/strings.html>

10.3 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)
```

ID	height
1	152
104	172
2112	180
2543	163

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

ID	SBP
SG0001	120
SG0104	111
SG2112	125
SG2543	119

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

Sample	rs1212
TaqMan-SG0001-190601	G/C
TaqMan-SG0104-190602	G/G
TaqMan-SG2112-190603	C/C
TaqMan-Sg2543-190603	C/G

10.4 Discussion Questions

10.4.1 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 10.4: 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.

10.4.1.1 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")
root_srcfile <- "https://raw.githubusercontent.com/DanieleWeeks/HuGen2071/main/"
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)
kable(clin)
lab <- read.table(file = "data/lab_data.txt", header = TRUE)
kable(lab)
geno <- read.table(file = "data/genotype_data.txt", header = TRUE)
kable(geno)
# Edit/add R code here
```

10.4.2 Answer 1

💡 Expand to see solution

```
clin$SUBJECT_ID <- paste0("SG", formatC(clin$ID, width = 4, flag = "0000"))
kable(clin)



| ID   | height | SUBJECT_ID |
|------|--------|------------|
| 1    | 152    | SG0001     |
| 104  | 172    | SG0104     |
| 2112 | 180    | SG2112     |
| 2543 | 163    | SG2543     |



# Or here's an alternative using the 'sub' command:
sub("00", "SG", formatC(clin$ID, flag="0000", width=6))

[1] "SG0001" "SG0104" "SG2112" "SG2543"

# Or can be done using a `case_when`:
case_when(
  clin$ID < 10 ~ paste0("SG000", clin$ID),
  clin$ID < 100 ~ paste0("SG00", clin$ID),
  clin$ID < 1000 ~ paste0("SG0", clin$ID),
  clin$ID < 10000 ~ paste0("SG", clin$ID)
)

[1] "SG0001" "SG0104" "SG2112" "SG2543"
```

10.4.3 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 10.6: The `lab` data frame

ID	SBP
SG0001	120

ID	SBP
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
```

10.4.4 Answer 2

 Expand to see solution

```
lab$ID2 <- as.numeric(gsub("SG","",lab$ID))
kable(lab)
```

ID	SBP	ID2
SG0001	120	1
SG0104	111	104
SG2112	125	2112
SG2543	119	2543

```

lab$ID2 <- NA
lab$ID2 <- str_replace_all(lab$ID, pattern = "SG", replacement = "") %>% as.numeric()
kable(lab)

```

ID	SBP	ID2
SG0001	120	1
SG0104	111	104
SG2112	125	2112
SG2543	119	2543

10.4.5 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 10.9: The `geno` data frame

Sample	rs1212
TaqMan-SG0001-190601	G/C
TaqMan-SG0104-190602	G/G
TaqMan-SG2112-190603	C/C
TaqMan-Sg2543-190603	C/G

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

```

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

```

10.4.6 Answer 3

💡 Expand to see solution

```
a <- str_split_fixed(geno$Sample, pattern = "-", n=3)
a

[,1]      [,2]      [,3]
[1,] "TaqMan" "SG0001" "190601"
[2,] "TaqMan" "SG0104" "190602"
[3,] "TaqMan" "SG2112" "190603"
[4,] "TaqMan" "Sg2543" "190603"

geno$ID <- toupper(a[,2])
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 `tidyverse` 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
```

10.5 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)
```

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

10.5.1 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 10.12: 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
```

10.5.2 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)
```

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)
```

	ID	SBP
	RP5002	121
	RP5012	118
	RP5113	112
	RP5213	142

11 Lecture: Loops in R

11.1 Loops in R lecture

Here's a recording of this lecture (8 minutes 11 seconds):

[Recording](#)

11.2 Loops in R slides

[PDF slide set](#)

12 Loops in R, Part I

12.1 Acknowledgment/License

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The material presented here has been modified from the original source.

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12.2 Source code

If you'd like to work within R Studio using the source code of this chapter, you can obtain it from [here](#).

12.3 Basic for loop

- Loops are the fundamental structure for repetition in programming
- `for` loops perform the same action for each item in a list of things

```
for (item in list_of_items) {  
  do_something(item)  
}
```

- To see an example of this let's calculate masses from volumes using a loop

- Need `print()` to display values inside a loop or function

```
volumes = c(1.6, 3, 8)
for (volume in volumes){
  mass <- 2.65 * volume ^ 0.9
  print(mass)
}
```

- Code in the loop will run once for each value in `volumes`
- Everything between the curly brackets is executed each time through the loop
- Code takes the first value from `volumes` and assigns it to `volume` and does the calculation and prints it
- Then it takes the second value from `volumes` and assigns it to `volume` and does the calculation and prints it
- And so on
- So, this loop does the same exact thing as

```
volume <- volumes[1]
mass <- 2.65 * volume ^ 0.9
print(mass)
volume <- volumes[2]
mass <- 2.65 * volume ^ 0.9
print(mass)
volume <- volumes[3]
mass <- 2.65 * volume ^ 0.9
print(mass)
```

! Do Tasks 1 & 2 in Basic For Loops

1. The code below prints the numbers 1 through 5 one line at a time. Modify it to print each of these numbers multiplied by 3.

```
numbers <- c(1, 2, 3, 4, 5)
for (number in numbers){
  print(number)
}
```

2. Write a for loop that loops over the following vector and prints out the mass in kilograms (`mass_kg = 2.2 * mass_lb`)

```
(mass_lbs <- c(2.2, 3.5, 9.6, 1.2))
# Edit/add/try out R code here
```

12.4 Looping with an index & storing results

- R loops iterate over a series of values in a vector or other list like object
- When we use that value directly this is called looping by value
- But there is another way to loop, which is called looping by index
- Looping by index loops over a list of integer index values, typically starting at 1
- These integers are then used to access values in one or more vectors at the position indicated by the index
- If we modified our previous loop to use an index it would look like this
- We often use `i` to stand for “index” as the variable we update with each step through the loop

```
volumes = c(1.6, 3, 8)
for (i ...)
```

- We then create a vector of position values starting at 1 (for the first value) and ending with the length of the object we are looping over

```
volumes = c(1.6, 3, 8)
for (i in 1:3)
```

- We don’t want to have to know the length of the vector and it might change in the future, so we’ll look it up using the `length()` function

```
volumes = c(1.6, 3, 8)
for (i in 1:length(volumes)){
}
```

- Then inside the loop instead of doing the calculation on the index (which is just a number between 1 and 3 in our case)
- We use square brackets and the index to get the appropriate value out of our vector

```
volumes = c(1.6, 3, 8)
for (i in 1:length(volumes)){
```

```
    mass <- 2.65 * volumes[i] ^ 0.9
    print(mass)
}
```

- This gives us the same result, but it's more complicated to understand
- So why would we loop by index?
- The advantage to looping by index is that it lets us do more complicated things
- One of the most common things we use this for are storing the results we calculated in the loop
- To do this we start by creating an empty object the same length as the results will be before the loop starts
- To store results in a vector we use the function `vector` to create an empty vector of the right length
- `mode` is the type of data we are going to store
- `length` is the length of the vector

```
masses <- vector(mode = "numeric", length = length(volumes))
masses
```

- Then add each result in the right position in this vector
- For each trip through the loop put the output into the empty vector at the `i`th position

```
for (i in 1:length(volumes)){
  mass <- 2.65 * volumes[i] ^ 0.9
  masses[i] <- mass
}
masses
```

! Do Tasks 3-4 in Basic For Loops.

3. Complete the code below so that it prints out the name of each bird one line at a time.

```
birds = c('robin', 'woodpecker', 'blue jay', 'sparrow')
for (i in 1:length(______)){
  print(birds[__])
}
```

4. Complete the code below so that it stores one area for each radius.

```

radius <- c(1.3, 2.1, 3.5)
areas <- vector(____ = "numeric", length = _____)
for (_ in 1:length(____)){
  areas[_] <- pi * radius[i] ^ 2
}
areas

```

12.5 Looping over multiple values

- Looping with an index also allows us to access values from multiple vectors

```

as <- c(2.65, 1.28, 3.29)
bs <- c(0.9, 1.1, 1.2)
volumes = c(1.6, 3, 8)
masses <- vector(mode="numeric", length=length(volumes))
for (i in 1:length(volumes)){
  mass <- as[i] * volumes[i] ^ bs[i]
  masses[i] <- mass
}
masses

```

! Do Task 5 in Basic For Loops.

5. Complete the code below to calculate an area for each pair of `lengths` and `widths`, store the areas in a vector, and after they are all calculated print them out:

```

lengths = c(1.1, 2.2, 1.6)
widths = c(3.5, 2.4, 2.8)
areas <- vector(length = _____)
for (i in _____) {
  areas[_] <- lengths[_] * widths[_]
}
areas

```

13 Conditionals in R

13.1 Acknowledgment/License

The original source for this chapter was from the web site

<https://datacarpentry.org/semester-biology/>

which was built using this underlying code

<https://github.com/datacarpentry/semester-biology>

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The material presented here has been modified from the original source.

Accordingly this chapter is made available under the same license terms.

13.2 Source code

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13.3 Conditionals

- Conditional statements are when we check to see if some condition is true or not
- We used these for filtering data in `dplyr`

```
weight <- 65
species <- "DM"
weight > 50
species == "DM"
```

- These statements generate a value is of type "logical"
- The value is TRUE if the condition is satisfied
- The value is FALSE if the condition is not satisfied
- These aren't the strings "TRUE" and "FALSE"
- They are a special type of value
- Conditional statements are made with a range of operators
- We've seen
 - == for equals
 - != for not equals
 - <, > for less than and greater than
 - <=, >= for less than or equal to and greater than or equal to
 - is.na() for is this value null
- There are others, including %in%, which checks to see if a value is present in a vector of possible values

```
10 >= 5
is.na(5)
"DM" %in% c("DM", "DO", "DS")
"PP" %in% c("DM", "DO", "DS")
```

- We can combine conditions using "and" and "or"
- We use the & for "and"
- Which means if both conditions are TRUE return TRUE
- If one of the conditions is FALSE then return FALSE

```
5 > 2 & 6 >=10
```

- We use the | for "or"
- Which means if either or both of the conditions are TRUE return TRUE

```
5 > 2 | 6 >=10
```

- Vectors of values compared to a single value return one logical per value

```
c(1, 1, 2, 3, 1) == 1
```

- Checks each value to see if equal to 1
- This is what subsetting approaches use to subset

- They keep the values where the value in this condition vector is equal to TRUE
- Let's look at an example where we have a vector of sites and a vector of the states they occur in

```
(site = c('a', 'b', 'c', 'd'))
(state = c('FL', 'FL', 'GA', 'AL'))
```

- A conditional statement checking if the state is 'FL' returns a vector of TRUE's and FALSEs

```
state == 'FL'
```

- So when we filter the site vector to only return values where the state is equal to 'FL'

```
site[state == 'FL']
```

- It is the same as passing a vector of TRUE and FALSE values inside the square brackets

```
site[c(TRUE, TRUE, FALSE, FALSE)]
```

- This keeps the first and second values in site because the values in the vector are TRUE
- This is how dplyr::filter() and other methods for subsetting data work

13.3.1 Tasks: Choice Operators

! Important

Do Tasks 1-4 in Choice Operators

Create the following variables.

```
(w <- 10.2)
(x <- 1.3)
(y <- 2.8)
(z <- 17.5)
(colors <- c("red", "blue", "green"))
(masses <- c(45.2, 36.1, 27.8, 81.6, 42.4))
```

Use them to print whether or not the following statements are TRUE or FALSE.

1. w is greater than 10
2. "green" is in colors

3. x is greater than y
4. Each value in `masses` is greater than 40.

```
# Edit/add/try out R code here
```

13.4 if statements

- Conditional statements generate logical values to filter inputs.
- `if` statements use conditional statements to control flow of the program.

```
if (the conditional statement is TRUE ) {
  do something
}
```

- Example

```
x = 6
if (x > 5){
  x = x^2
}
x
```

- `x > 5` is TRUE, so the code in the `if` runs
- `x` is now 6^2 or 36
- Change `x` to 4

```
x = 4
if (x > 5){
  x = x^2
}
x
```

- `x > 5` is FALSE, so the code in the `if` doesn't run
- `x` is still 4
- This is *not* a function, so everything that happens in the `if` statement influences the global environment
- Different mass calculations for different vegetation types

```
veg_type <- "shrub"
volume <- 16.08
if (veg_type == "shrub") {
  mass <- 2.65 * volume^0.9
}
mass
```

13.4.1 Task 1: Basic If Statements

! Important

Do Task 1 in Basic If Statements

1. Complete (i.e., copy into your code and then modify) the following `if` statement so that if `age_class` is equal to “sapling” it sets `y <- 10`.

```
age_class = "sapling"
if (){

}
y
```

- Often want to chose one of several options
- Can add more conditions and associated actions with `else if`

```
veg_type <- "grass"
volume <- 16.08
if (veg_type == "shrub") {
  mass <- 2.65 * volume^0.9
} else if (veg_type == "grass") {
  mass <- 0.65 * volume^1.2
}
mass
```

- Checks the first condition
- If TRUE runs that condition’s code and skips the rest
- If not it checks the next one until it runs out of conditions
- Can specify what to do if none of the conditions is TRUE using `else` on its own

```
veg_type <- "tree"
volume <- 16.08
if (veg_type == "shrub") {
  mass <- 2.65 * volume^0.9
} else if (veg_type == "grass") {
  mass <- 0.65 * volume^1.2
} else {
  mass <- NA
}
mass
```

13.4.2 Tasks 2-3: Basic If Statements

! Important

Do Tasks 2-3 in Basic If Statements

2. Complete the following `if` statement so that if `age_class` is equal to “sapling” it sets `y <- 10` and if `age_class` is equal to “seedling” it sets `y <- 5`.

```
age_class = "seedling"
if (){

}
y
```

3. Complete the following `if` statement so that if `age_class` is equal to “sapling” it sets `y <- 10` and if `age_class` is equal to “seedling” it sets `y <- 5` and if `age_class` is something else then it sets the value of `y <- 0`.

```
age_class = "adult"
if (){

}
y
```

13.5 Multiple ifs vs else if

- Multiple ifs check each conditional separately
- Executes code of all conditions that are TRUE

```
x <- 5
if (x > 2){
  x * 2
}
if (x > 4){
  x * 4
}
x
```

- `else if` checks each condition sequentially
- Executes code for the first condition that is TRUE

```
x <- 5
if (x > 2){
  x * 2
} else if (x > 4){
  x * 4
}
x
```

13.6 Using Conditionals Inside Functions

- We've used a conditional to estimate mass differently for different types of vegetation
- This is the kind of code we are going to want to reuse, so let's move it into a function
- We do this by placing the same code inside of a function
- And making sure that the function takes all required variables as input

```
est_mass <- function(volume, veg_type){
  if (veg_type == "shrub") {
    mass <- 2.65 * volume^0.9
  } else if (veg_type == "grass") {
    mass <- 0.65 * volume^1.2
  } else {
    mass <- NA
  }
  return(mass)
}
```

- We can then run this function with different vegetation types and get different estimates for mass

```
est_mass(1.6, "shrub")
est_mass(1.6, "grass")
est_mass(1.6, "tree")
```

- Let's walk through how this code executes using the debugger
- When we call the function the first thing that happens is that 1.6 gets assigned to `volume` and "tree" gets assigned to `veg_type`
- The code then checks to see if `veg_type` is equal to "shrub"
- It isn't so the code then checks to see if `veg_type` is equal to "grass"
- It isn't so the code then hits the `else` statement and executes the code in the `else` block
- It assigns `NA` to `mass`
- It then finishes the if/else if/else statement and returns the value for `mass`, which is `NA` to the global environment

13.6.1 Task: Size Estimates by Name

! Important

Do Size Estimates by Name

13.6.1.1 Part I

The length of an organism is typically strongly correlated with its body mass. This is useful because it allows us to estimate the mass of an organism even if we only know its length. This relationship generally takes the form:

$$\text{mass} = a * \text{length}^b$$

Where the parameters `a` and `b` vary among groups. This allometric approach is regularly used to estimate the mass of dinosaurs since we cannot weigh something that is only preserved as bones.

The following function estimates the mass of an organism in kg based on its length in meters for a particular set of parameter values, those for *Theropoda* (where `a` has been estimated as 0.73 and `b` has been estimated as 3.63; [Seebacher 2001](#)).

```
get_mass_from_length_theropoda <- function(length){
  mass <- 0.73 * length ^ 3.63
  return(mass)
}
```

1. Use this function to print out the mass of a Theropoda that is 16 m long based on its reassembled skeleton.

```
# Edit/add/try out R code here
```

2. Create a new version of this function called `get_mass_from_length()` that takes `length`, `a` and `b` as arguments and uses the following code to estimate the mass `mass <- a * length ^ b`. Use this function to estimate the mass of a Sauropoda (`a = 214.44`, `b = 1.46`) that is 26 m long.

```
# Edit/add/try out R code here
```

13.6.1.2 Part II

To make it even easier to work with your dinosaur size estimation functions you decide to create a function that lets you specify which dinosaur group you need to estimate the size of by name and then have the function automatically choose the right parameters. Create a new function `get_mass_from_length_by_name()` that takes two arguments, the `length` and the name of the dinosaur group. Inside this function use `if/else if/else` statements to check to see if the name is one of the following values and if so use the associated `a` and `b` values to estimate the species mass.

- *Stegosauria*: `a = 10.95` and `b = 2.64` ([Seebacher 2001](#)).
- *Theropoda*: `a = 0.73` and `b = 3.63` ([Seebacher 2001](#)).
- *Sauropoda*: `a = 214.44` and `b = 1.46` ([Seebacher 2001](#)).

If the name is not any of these values the function should return `NA`.

Run the function for: 1. A *Stegosauria* that is 10 meters long. 2. A *Theropoda* that is 8 meters long. 3. A *Sauropoda* that is 12 meters long. 4. A *Ankylosauria* that is 13 meters long.

```
# Edit/add/try out R code here
```

Challenge (optional): If the name is not one of values that have `a` and `b` values print out a message that it doesn't know how to convert that group that includes that groups name in a message like "No known estimation for Ankylosauria". (the function `paste()` will be helpful here). Doing this successfully will modify your answer to (4), which is fine.

```
# Edit/add/try out R code here
```

Challenge (optional): Change your function so that it uses two different values of `a` and `b` for *Stegosauria*. When *Stegosauria* is greater than 8 meters long use the equation above.

When it is less than 8 meters long use $a = 8.5$ and $b = 2.8$. Run the function for a *Stegosauria* that is 6 meters long.

```
# Edit/add/try out R code here
```

Challenge (optional): Rewrite your function so that instead of calculating mass directly it sets the values of a and b to the values for the species (or to NA if the species doesn't have an equation) and then calls another function to do the basic $\text{mass} = a * \text{length}^b$ calculation.

```
# Edit/add/try out R code here
```

13.7 Automatically extracting functions

- Can pull code out into functions
- Highlight the code
- Code -> Extract Function
- Provide a name for the function

13.8 Nested conditionals

- Sometimes decisions are more complicated
- For example we might have different equations for some vegetation types based on the age of the plant
- Can “nest” conditionals inside of one another

```
est_mass <- function(volume, veg_type, age){  
  if (veg_type == "shrub") {  
    if (age < 5) {  
      mass <- 1.6 * volume^0.8  
    } else {  
      mass <- 2.65 * volume^0.9  
    }  
  } else if (veg_type == "grass" | veg_type == "sedge") {  
    mass <- 0.65 * volume^1.2  
  } else {  
    mass <- NA  
  }  
}
```

```
    return(mass)
}

est_mass(1.6, "shrub", age = 2)
est_mass(1.6, "shrub", age = 6)
```

- First checks if the vegetation type is “shrub”
- If it is checks to see if it is < 5 years old
- If so does one calculation, if not does another
- But nesting can be difficult to follow so try to minimize it

13.8.1 Task 4: Basic If Statements

! Important

Do Task 4 in Basic If Statements

4. Convert your conditional statement from Task 3 in Section 13.4.2 into a function that takes `age_class` as an argument and returns `y`. Call this function 5 times, once with each of the following values for `age_class`: “sapling”, “seedling”, “adult”, “mature”, “established”.

```
# Edit/add/try out R code here
```

14 Loops in R, Part II

14.1 Acknowledgment/License

The original source for this chapter was from the web site

<https://datacarpentry.org/semester-biology/>

which was built using this underlying code

<https://github.com/datacarpentry/semester-biology>

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The material presented here has been modified from the original source.

Accordingly this chapter is made available under the same license terms.

14.2 Source code

If you'd like to work within R Studio using the source code of this chapter, you can obtain it from [here](#).

14.3 Looping with functions

- It is common to combine loops with functions by calling one or more functions as a step in our loop
- For example, let's take the non-vectorized version of our `est_mass` function that returns an estimated mass if the `volume > 5` and `NA` if it's not.

```
est_mass <- function(volume, a, b){  
  if (volume > 5) {  
    mass <- a * volume ^ b  
  } else {  
    mass <- NA  
  }  
  return(mass)  
}
```

```

    } else {
      mass <- NA
    }
    return(mass)
}
class(est_mass)

```

- We can't pass the vector to the function and get back a vector of results because of the `if` statements
- So let's loop over the values
- First we'll create an empty vector to store the results
- And then loop by index, calling the function for each value of `volumes`

```

as <- c(2.65, 1.28, 3.29)
bs <- c(0.9, 1.1, 1.2)
volumes = c(1.6, 3, 8)
masses <- vector(mode="numeric", length=length(volumes))
for (i in 1:length(volumes)){
  mass <- est_mass(volumes[i], as[i], bs[i])
  masses[i] <- mass
}
masses

```

- This is the for loop equivalent of an `mapply` statement

```
(masses_apply <- mapply(est_mass, volumes, as, bs))
```

! Do Size Estimates By Name Loop.

If `dinosaur_lengths.csv` is not already in your working directory download a copy of the [data on dinosaur lengths with species names](#). Load it into R.

Write a function `mass_from_length()` that uses the equation `mass <- a * length^b` to estimate the size of a dinosaur from its length. This function should take two arguments, `length` and `species`. For each of the following inputs for `species`, use the given values of `a` and `b` for the calculation:

- For Stegosauria: `a = 10.95` and `b = 2.64` ([Seebacher 2001](#)).
 - For Theropoda: `a = 0.73` and `b = 3.63` ([Seebacher 2001](#)).
 - For Sauropoda: `a = 214.44` and `b = 1.46` ([Seebacher 2001](#)).
 - For any other value of `species`: `a = 25.37` and `b = 2.49`.
1. Use this function and a for loop to calculate the estimated mass for each dinosaur,

store the masses in a vector, and after all of the calculations are complete show the first few items in the vector using `head()`.

```
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"  
download.file(paste0(root_srcfile,"data/dinosaur_lengths.csv"),  
              "dinosaur_lengths.csv")  
list.files(pattern = "dinosaur")  
# Edit/add/try out R code here
```

2. Add the results in the vector back to the original data frame. Show the first few rows of the data frame using `head()`.

```
# Edit/add/try out R code here
```

3. Calculate the mean mass for each `species` using `dplyr`.

```
# Edit/add/try out R code here
```

14.4 Looping over files

- Repeat same actions on many similar files
- Let's download some simulated satellite collar data

```
# Download files within the WebR environment  
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"  
download.file(paste0(root_srcfile,"data/locations.zip"),  
              "locations.zip")  
unzip("locations.zip")
```

- Now we need to get the names of each of the files we want to loop over
- We do this using `list.files()`
- If we run it without arguments it will give us the names of all files in the directory

```
list.files()
```

- But we just want the data files so we'll add the optional `pattern` argument to only get the files that start with "locations-"

```
(data_files = list.files(pattern = "locations-"))
```

- Once we have this list we can loop over it count the number of observations in each file
- First create an empty vector to store those counts

```
(n_files = length(data_files))
(results <- integer(n_files))
```

- Then write our loop

```
for (i in 1:n_files){
  filename <- data_files[i]
  data <- read.csv(filename)
  count <- nrow(data)
  results[i] <- count
}
results
```

! Do Task 1 of Multiple-file Analysis.

Exercise uses different collar data

You have a satellite collars on a number of different individuals and want to be able to quickly look at all of their recent movements at once. The data is posted daily to a zip file that contains one csv file for each individual: [data/individual_collar_data.zip](#)

Start your solution by:

- If `individual_collar_data.zip` is not already in your working directory download [the zip file](#) using `download.file()`
 - Unzip it using `unzip()`
 - Obtain a list of all of the files with file names matching the pattern "`collar-data-.*.txt`" (using `list.files()`)
- Use a loop to load each of these files into R and make a line plot (using `geom_path()`) for each file with `long` on the x axis and `lat` on the y axis. Graphs, like other types of output, won't display inside a loop unless you explicitly display them, so you need put your `ggplot()` command inside a `print()` statement.

Include the name of the file in the graph as the graph title using `labs()`.

```

root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
download.file(paste0(root_srcfile,"data/individual_collar_data.zip"),
              "individual_collar_data.zip")
unzip("individual_collar_data.zip")
list.files()
# Edit/add/try out R code here

```

14.5 Storing loop results in a data frame

- We often want to calculate multiple pieces of information in a loop making it useful to store results in things other than vectors
- We can store them in a data frame instead by creating an empty data frame and storing the results in the *i*th row of the appropriate column
- Associate the file name with the count
- Also store the minimum latitude
- Start by creating an empty data frame
- Use the `data.frame` function
- Provide one argument for each column
- “Column Name” = “an empty vector of the correct type”

```
(results <- data.frame(file_name = character(n_files),
                      count = integer(n_files),
                      min_lat = numeric(n_files)))
```

- Now let's modify our loop from last time
- Instead of storing `count` in `results[i]` we need to first specify the `count` column using the `$: results$count[i]`
- We also want to store the filename, which is `data_files[i]`

```

for (i in 1:n_files){
  filename <- data_files[i]
  data <- read.csv(filename)
  count <- nrow(data)
  min_lat = min(data$lat)
  results$file_name[i] <- filename
  results$count[i] <- count
  results$min_lat[i] <- min_lat
}

```

```
results
```

! Do Task 2 of Multiple-file Analysis.

Exercise uses different collar data

2. Add code to the loop to calculate the minimum and maximum latitude in the file, and store these values, along with the name of the file, in a data frame. Show the data frame as output.

```
# Edit/add/try out R code here
```

If you're interested in seeing another application of for loops, check out the code below used to simulate the data for this exercise using for loops.

```
individuals = paste(c('A', 'B', 'C', 'D', 'E', 'F', 'G', 'H', 'I', 'J'), c(1:10), sep = "
for (individual in individuals) {
  lat = vector("numeric", 24)
  long = vector("numeric", 24)
  lat[1] = rnorm(1, mean = 26, sd = 2)
  long[1] = rnorm(1, mean = -35, sd = 3)
  for (i in 2:24) {
    lat[i] = lat[i - 1] + rnorm(1, mean = 0, sd = 1)
    long[i] = long[i - 1] + rnorm(1, mean = 0, sd = 1)
  }
  times = seq(from=as.POSIXct("2016-02-26 00:00", tz="UTC"),
              to=as.POSIXct("2016-02-26 23:00", tz="UTC"),
              by="hour")
  df = data.frame(date = "2016-02-26",
                  collar = individual,
                  time = times,
                  lat = lat,
                  long = long)
  write.csv(df, paste("collar-data-", individual, "-2016-02-26.txt", sep = ""))
}
zip("data/individual_collar_data.zip", list.files(pattern = "collar-data-[A-Z] [0-9]+.*"))
```

14.6 Subsetting Data

- Loops can subset in ways that are difficult with things like `group_by`

- Look at some data on trees from the National Ecological Observatory Network

```
library(ggplot2)
library(dplyr)

root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
download.file(paste0(root_srcfile,"data/harv_034subplt.csv"),
               "harv_034subplt.csv")
neon_trees <- read.csv('harv_034subplt.csv')
head(neon_trees)
ggplot(neon_trees, aes(x = easting, y = northing)) +
  geom_point()
```

- Look at a north-south gradient in number of trees
- Need to know number of trees in each band of y values
- Start by defining the size of the window we want to use
 - Use the grid lines which are 2.5 m

```
(window_size <- 2.5)
```

- Then figure out the edges for each window

```
(south_edges <- seq(4713095, 4713117.5, by = window_size))
(north_edges <- south_edges + window_size)
```

- But we don't want to go all the way to the far edge

```
(south_edges <- seq(4713095, 4713117.5 - window_size, by = window_size))
(north_edges <- south_edges + window_size)
```

- Set up an empty data frame to store the output

```
(counts <- vector(mode = "numeric", length = length(south_edges)))
```

- Look over the left edges and subset the data occurring within each window

```
for (i in 1:length(south_edges)) {
  data_in_window <- filter(neon_trees, northing >= south_edges[i], northing < north_edges[i])
  counts[i] <- nrow(data_in_window)
}
counts
```

```

yedges <- unique(c(south_edges, north_edges))
ggplot(neon_trees, aes(x = easting, y = northing)) +
  geom_point() +
  geom_hline(yintercept = yedges) +
  scale_y_reverse()

```

14.7 Nested Loops

- Sometimes need to loop over multiple things in a coordinate fashion
- Pass a window over some spatial data
- Look at full spatial pattern not just east-west gradient
- Basic nested loops work by putting one loop inside another one

```

for (i in 1:3) {
  for (j in 1:2) {
    print(paste("i = " , i, "; j = ", j))
  }
}

```

- Loop over x and y coordinates to create boxes
- Need top and bottom edges

```
(east_edges <- seq(731752.5, 731772.5 - window_size, by = window_size))
(west_edges <- east_edges + window_size)
```

- Redefine out storage

```
(output <- matrix(nrow = length(south_edges), ncol = length(east_edges)))
```

```

for (i in 1:length(south_edges)) {
  for (j in 1:length(east_edges)) {
    data_in_window <- filter(neon_trees,
      northing >= south_edges[i], northing < north_edges[i],
      easting >= east_edges[j], easting < west_edges[j])
    output[i, j] <- nrow(data_in_window)
  }
}

```

```
output

xedges <- unique(c(east_edges, west_edges))
yedges <- unique(c(south_edges, north_edges))
ggplot(neon_trees, aes(x = easting, y = northing)) +
  geom_point() +
  geom_vline(xintercept=xedges) +
  geom_hline(yintercept = yedges) +
  scale_y_reverse()
```

14.8 Sequence along

- `seq_along()` generates a vector of numbers from 1 to `length(volumes)`

```
1:length(east_edges)
seq_along(east_edges)
```

15 Functions

15.1 Acknowledgment/License

The original source for this chapter was from the web site

<https://datacarpentry.org/semester-biology/>

which was built using this underlying code

<https://github.com/datacarpentry/semester-biology>

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The material presented here has been modified from the original source.

Accordingly this chapter is made available under the same license terms.

15.2 Source code

If you'd like to work within R Studio using the source code of this chapter, you can obtain it from [here](#).

15.3 Understandable and reusable code

- Write code in understandable chunks.
- Write reusable code.

15.4 Understandable chunks

- Human brain can only hold limited number of things in memory
- Write programs that don't require remembering all of the details at once
- Treat functions as a single conceptual chunk.

15.5 Reuse

- Want to do the same thing repeatedly?
 - Inefficient & error prone to copy code
 - If it occurs in more than one place, it will eventually be wrong somewhere.
- Functions are written to be reusable.

15.6 Function basics

```
function_name <- function(inputs) {  
  output_value <- do_something(inputs)  
  return(output_value)  
}
```

- The braces indicate that the lines of code are a group that gets run together

```
{a = 2  
b = 3  
a + b}
```

- Pressing run anywhere in this group runs all the lines in that group
- A function runs all of the lines of code in the braces
- Using the arguments provided
- And then returns the output

```
calc_shrub_vol <- function(length, width, height) {  
  area <- length * width  
  volume <- area * height  
  return(volume)  
}  
class(calc_shrub_vol)
```

- Creating a function doesn't run it.
- Call the function with some arguments.

```
calc_shrub_vol(0.8, 1.6, 2.0)
```

- Store the output to use it later in the program

```
(shrub_vol <- calc_shrub_vol(0.8, 1.6, 2.0))
```

! Do Writing Functions

Edit the following function to replace the _____ with variables names for the input and output.

```
convert_pounds_to_grams <- function(_____) {
  grams = 453.6 * pounds
  return(_____)
}
```

Use the function to calculate how many grams there are in 3.75 pounds.

```
# Edit/add/try out R code here
```

- Treat functions like a black box
 - *Draw a box on board showing inputs->function->outputs*
 - The only things the function knows about are the inputs we pass it
 - The only thing the program knows about the function is the output it produces

! Do Function Execution

- Walk through function execution (using debugger)
 - Call function
 - Assign 0.8 to length, 1.6 to width, and 2.0 to height inside function
 - Calculate the area and assign it to `area`
 - Calculate volume and assign it to `volume`
 - Send `volume` back as output
 - Store it in `shrub_vol`

```
# Edit/add/try out R code here
```

Solution

```
shrubVol <- function(length=0.8, width=1.6, height=2.0) {  
  area <- length * width  
  volume <- area * height  
  return(volume)  
}  
shrubVol()
```

- Treat functions like a black box.
 - Can't access a variable that was created in a function
 - * > volume
 - * Error: object 'volume' not found
 - Or an argument by name
 - * > width
 - * Error: object 'width' not found
 - ‘Global’ variables can influence function, but should not.
 - * Very confusing and error prone to use a variable that isn’t passed in as an argument

Do Use and Modify.

The length of an organism is typically strongly correlated with its body mass. This is useful because it allows us to estimate the mass of an organism even if we only know its length. This relationship generally takes the form:

$$\text{mass} = a * \text{length}^b$$

Where the parameters **a** and **b** vary among groups. This allometric approach is regularly used to estimate the mass of dinosaurs since we cannot weigh something that is only preserved as bones.

The following function estimates the mass of an organism in kg based on its length in meters for a particular set of parameter values, those for *Theropoda* (where **a** has been estimated as 0.73 and **b** has been estimated as 3.63; [Seebacher 2001](#)).

```
get_mass_from_length_theropoda <- function(length){  
  mass <- 0.73 * length ^ 3.63  
  return(mass)  
}  
class(get_mass_from_length_theropoda)
```

1. Use this function to print out the mass of a Theropoda that is 16 m long based on its reassembled skeleton.

```
# Edit/add/try out R code here
```

2. Create a new version of this function called `get_mass_from_length()` that takes `length`, `a` and `b` as arguments and uses the following code to estimate the mass
`mass <- a * length ^ b.`

```
# Edit/add/try out R code here
```

Use this function to estimate the mass of a Sauropoda ($a = 214.44$, $b = 1.46$) that is 26 m long.

```
# Edit/add/try out R code here
```

15.7 Default arguments

- Defaults can be set for common inputs.
- For example, many of our shrubs are the same height so for those shrubs we only measure the `length` and `width`.
- So we want a default value for the `height` for cases where we don't measure it

```
calc_shrub_vol <- function(length, width, height = 1) {  
  area <- length * width  
  volume <- area * height  
  return(volume)  
}  
  
calc_shrub_vol(0.8, 1.6)  
calc_shrub_vol(0.8, 1.6, 2.0)
```

```
calc_shrub_vol(length = 0.8, width = 1.6, height = 2.0)
```

! Do Default Arguments.

This is a follow up to the Use and Modify exercise above.

Allowing `a` and `b` to be passed as arguments to `get_mass_from_length()` made the function more flexible, but for some types of dinosaurs we don't have specific values of `a` and `b` and so we have to use general values that can be applied to a number of different species.

Rewrite your `get_mass_from_length()` function from Use and Modify so that its arguments have default values of `a = 39.9` and `b = 2.6` (the average values from [Seebacher 2001](#)).

```
# Edit/add/try out R code here
```

1. Use this function to estimate the mass of a Sauropoda (`a = 214.44`, `b = 1.46`) that is 22 m long (by setting `a` and `b` when calling the function).

```
# Edit/add/try out R code here
```

2. Use this function to estimate the mass of a dinosaur from an unknown taxonomic group that is 16m long. Only pass the function `length`, not `a` and `b`, so that the default values are used.

```
# Edit/add/try out R code here
```

Discuss why passing `a` and `b` in is more useful than having them fixed

15.8 Named vs unnamed arguments

- When to use or not use argument names

```
calc_shrub_vol(length = 0.8, width = 1.6, height = 2.0)
```

Or

```
calc_shrub_vol(0.8, 1.6, 2.0)
```

- You can always use names
 - Value gets assigned to variable of that name
- If not using names then order determines naming
 - First value is `length`, second value is `width`, third value is `height`
 - If order is hard to remember use names
- In many cases there are *a lot* of optional arguments
 - Convention to always name optional argument
- So, in our case, the most common approach would be

```
calc_shrub_vol(0.8, 1.6, height = 2.0)
```

15.9 Combining Functions

- Each function should be single conceptual chunk of code
- Functions can be combined to do larger tasks in two ways
- Calling multiple functions in a row

```
est_shrub_mass <- function(volume){
  mass <- 2.65 * volume^0.9
}

(shrub_volume <- calc_shrub_vol(0.8, 1.6, 2.0))
(shrub_mass <- est_shrub_mass(shrub_volume))
```

- We can also use pipes with our own functions
- The output from the first function becomes the first argument for the second function

```
library(dplyr)
(shrub_mass <- calc_shrub_vol(0.8, 1.6, 2.0) %>%
  est_shrub_mass())
```

! Do Combining Functions.

This is a follow up to the Default Argument exercise above.

Measuring things using the metric system is the standard approach for scientists, but when communicating your results more broadly it may be useful to use different units (at

least in some countries). Write a function called `convert_kg_to_pounds` that converts kilograms into pounds (`pounds = 2.205 * kg`).

```
# Edit/add/try out R code here
```

Use that function and your `get_mass_from_length()` function from Default Arguments to estimate the weight, in pounds, of a 12 m long Stegosaurus with $a = 10.95$ and $b = 2.64$ (The estimated a and b values for *Stegosauria* from [Seebacher 2001](#)).

```
# Edit/add/try out R code here
```

- We can nest functions

```
(shrub_mass <- est_shrub_mass(calc_shrub_vol(0.8, 1.6, 2.0)))
```

- But we careful with this because it can make code difficult to read
- Don't nest more than two functions
- Can also call functions from inside other functions
- Allows organizing function calls into logical groups

```
est_shrub_mass_dim <- function(length, width, height){  
  volume = calc_shrub_vol(length, width, height)  
  mass <- est_shrub_mass(volume)  
  return(mass)  
}
```

```
est_shrub_mass_dim(0.8, 1.6, 2.0)
```

- We **don't** need to pass the function name into the function
- That's the one violation of the black box rule

15.10 Using dplyr & ggplot in functions

- There is an extra step we need to take when working with functions from dplyr and ggplot that work with “data variables”, i.e., names of columns that are not in quotes
- These functions use tidy evaluation, a special type of non-standard evaluation
- This basically means they do fancy things under the surface to make them easier to work with

- But it means they don't work if we just pass things to functions in the most natural way

```
library(ggplot2)

make_plot <- function(df, column, label) {
  ggplot(data = df, mapping = aes(x = column)) +
    geom_histogram() +
    xlab(label)
}

root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
download.file(paste0(root_srcfile,"data/surveys.csv"),
               "surveys.csv")
list.files(pattern = "surveys")

surveys <- read.csv("surveys.csv")
make_plot(surveys, hindfoot_length, "Hindfoot Length [mm]")
```

- To fix this we have to tell our code which inputs/arguments are this special type of data variable
- We do this by “embracing” them in double braces

```
library(ggplot2)

make_plot <- function(df, column, label) {
  ggplot(data = df, mapping = aes(x = {{ column }})) +
    geom_histogram() +
    xlab(label)
}

surveys <- read.csv("surveys.csv")
make_plot(surveys, hindfoot_length, "Hindfoot Length [mm]")
make_plot(surveys, weight, "Weight [g]")
```

15.11 Code design with functions

- Functions let us break code up into logical chunks that can be understood in isolation
- Write functions at the top of your code then call them at the bottom
- The functions hold the details
- The function calls show you the outline of the code execution

```

clean_data <- function(data){
  do_stuff(data)
}

process_data <- function(cleaned_data){
  do_dplyr_stuff(cleaned_data)
}

make_graph <- function(processed_data){
  do_ggplot_stuff(processed_data)
}

raw_data <- read.csv('mydata.csv')
cleaned_data <- clean_data(raw_data)
processed_data <- process_data(cleaned_data)
make_graph(processed_data)

```

15.12 Documentation & Comments

- Documentation
 - How to use code
 - Use Roxygen comments for functions
- Comments
 - Why & how code works
 - Only if it code is confusing to read

15.13 Working with functions in RStudio

- It is possible to find and jump between functions
- Click on list of functions at bottom of editor and select
- Can be helpful to clearly see what is a function
- Can have RStudio highlight them
- Global Options -> Code -> Display -> Highlight R function calls

16 R Functions Excercise

16.1 Load Libraries

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

16.2 Data set creation code

```
i <- 6
for (i in 1:10) {
  f1 <- data.frame(name=rep(paste0("name",i),26))
  b <- data.frame(name = rep(NA, 26))
  b$name <- paste0(f1$name,"_",letters)
  b$trait <- rnorm(26)
  write_tsv(b,paste0("data/dataset",i,".txt"))
}
```

16.3 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)
f11 <- read.table("data/dataset1.txt",sep="\t",header=TRUE)
results$dataset[1] <- "dataset1"
results$N <- nrow(f11)
```

```

results$mean[1] <- mean(f11$trait)
results$median[1] <- median(f11$trait)
results$var[1] <- var(f11$trait)
results

      dataset   N     mean    median     var
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)
results$dataset[2] <- "dataset2"
results$N <- nrow(f12)
results$mean[2] <- mean(f12$trait)
results$median[2] <- median(f12$trait)
results$var[2] <- var(f12$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          <NA> 26           NA           NA           NA

f13 <- read.table("data/dataset3.txt",sep="\t",header=TRUE)
results$dataset[3] <- "dataset3"
results$N <- nrow(f13)
results$mean[3] <- mean(f13$trait)
results$median[3] <- median(f13$trait)
results$var[3] <- var(f13$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

```

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.

16.3.1 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:

```
f1s <- list.files(path="data", pattern="dataset*")  
f1s
```

```
[1] "dataset1.txt" "dataset2.txt" "dataset3.txt" "dataset4.txt" "dataset5.txt"  
[6] "dataset6.txt"
```

16.3.2 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

16.3.3 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 `f1s`
- Count the number of input files `N`
- Set up an empty results table with `N` rows
- For each file in our file name list `f1s`

- Read the file
- Compute the statistics
- Insert the information into the correct row of the results table
- Return the filled-in results table

16.3.4 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.

 Expand to see solution

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) {
  f11 <- read.table(paste0("data/dataset",n,".txt"),sep="\t",header=TRUE)
  results$dataset[n] <- paste0("dataset",n,".txt")
  results$N <- nrow(f11)
  results$mean[n] <- mean(f11$trait)
  results$median[n] <- median(f11$trait)
  results$var[n] <- var(f11$trait)
  invisible(results)
}
```

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)  
}
```

16.3.5 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.

16.3.6 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*")

loop_over_dataset <- function(fls) {
  # Input: the list of file names
  # Output: the 'results' table
  # Count the number of data set file names in fls
  n_datasets <- length(fls)
  # Set up a results data frame 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)
  }
  return(results)
}

loop_over_dataset(fls = fls)

      dataset   N     mean     median     var
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
```

16.3.7 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) {
  f11 <- read.table(paste0("data/dataset",n,".txt"),sep="\t",header=TRUE)
  results$dataset[n] <- paste0("dataset",n,".txt")
  results$N <- nrow(f11)
  results$mean[n] <- mean(f11$trait)
  results$median[n] <- median(f11$trait)
  results$var[n] <- var(f11$trait)
  invisible(results)
```

}

💡 Expand to see solution

If `N` varies across the data sets, then this line will not do the right thing:

```
results$N <- nrow(f11)

results <- data.frame(dataset=rep(NA,6), N=NA, mean=NA, median=NA, var=NA)
read_data_file <- function(n=1, results) {
  f11 <- read.table(paste0("data/dataset",n,".txt"), sep="\t", header=TRUE)
  results$dataset[n] <- paste0("dataset",n,".txt")
  results$N[n] <- nrow(f11)
  results$mean[n] <- mean(f11$trait)
  results$median[n] <- median(f11$trait)
  results$var[n] <- var(f11$trait)
  invisible(results)
}
```

17 Tidyverse

17.1 Acknowledgment/License

The original source for this chapter was from the web site

<https://github.com/UoMResearchIT/r-day-workshop/>

which was used to build this web page:

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

and is used under the

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The material presented here has been modified from the original source.

Accordingly this chapter is made available under the same license terms.

17.2 Load gapminder data

In the previous episode we used the `readr` package to load the gapminder data into a tibble within R.

```
library(tidyverse)

# Download files within the WebR environment
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
download.file(paste0(root_srcfile,"data/gapminder-FiveYearData.csv"),
              "gapminder-FiveYearData.csv")
gapminder <- read_csv("gapminder-FiveYearData.csv")
dim(gapminder)
head(gapminder)
```

In this episode we'll use the `dplyr` package to manipulate the data we loaded, and calculate some summary statistics. We'll also introduce the concept of “pipes”.

17.3 Manipulating tibbles

Manipulation of tibbles means many things to many researchers. We often select only certain observations (rows) or variables (columns). We often group the data by a certain variable(s), or calculate summary statistics.

17.4 The dplyr package

The `dplyr` package is part of the tidyverse. It provides a number of very useful functions for manipulating tibbles (and their base-R cousin, the `data.frame`) in a way that will reduce repetition, reduce the probability of making errors, and probably even save you some typing.

We will cover:

1. selecting variables with `select()`
2. subsetting observations with `filter()`
3. grouping observations with `group_by()`
4. generating summary statistics using `summarize()`
5. generating new variables using `mutate()`
6. Sorting tibbles using `arrange()`
7. chaining operations together using pipes `%>%`

17.5 Using `select()`

If, for example, we wanted to move forward with only a few of the variables in our tibble we use the `select()` function. This will keep only the variables you select.

```
year_country_gdp <- select(gapminder, year, country, gdpPercap)
print(year_country_gdp)
```

Select will select *columns* of data. What if we want to select rows that meet certain criteria?

17.6 Other ways of selecting

Instead of saying what columns we *do* want, we can tell R which columns we don't want by prefixing the column name with a `-`. For example to select everything except year we would use `select(gapminder, -year)`.

There are also other ways of selecting columns based on parts of their names (such as `starts_with()` and `ends_with()`) - see `?select_helpers` for more information.

17.7 Using `filter()`

The `filter()` function is used to select rows of data. For example, to select only countries in Europe:

```
gapminder_Europe <- filter(gapminder, continent=="Europe")
print(gapminder_Europe)
```

Only rows of the data where the condition (i.e. `continent=="Europe"`) is TRUE are kept.

17.8 Using pipes and `dplyr`

We've now seen how to choose certain columns of data (using `select()`) and certain rows of data (using `filter()`). In an analysis we often want to do both of these things (and many other things, like calculating summary statistics, which we'll come to shortly). How do we combine these?

There are several ways of doing this; the method we will learn about today is using *pipes*.

The pipe operator `%>%` lets us pipe the output of one command into the next. This allows us to build up a data-processing pipeline. This approach has several advantages:

- We can build the pipeline piecemeal - building the pipeline step-by-step is easier than trying to perform a complex series of operations in one go
- It is easy to modify and reuse the pipeline
- We don't have to make temporary tibbles as the analysis progresses

Note

Note that R now has a native pipe operator `|>` which is very similar (but not identical) to the pipe operator `%>%` used here. The pipe operator `%>%` is defined by the `magrittr` R package, which is loaded when we load `dplyr` or `tidyverse`.

17.9 Pipelines and the shell

If you're familiar with the Unix shell, you may already have used pipes to pass the output from one command to the next. The concept is the same, except the shell uses the `|` character rather than R's pipe operator `%>%`

17.10 Keyboard shortcuts and getting help

The pipe operator can be tedious to type. In Rstudio pressing Ctrl + Shift+M under Windows / Linux will insert the pipe operator. On the mac, use ⌘ + Shift+M.

We can use tab completion to complete variable names when entering commands. This saves typing and reduces the risk of error.

RStudio includes a helpful “cheat sheet”, which summarises the main functionality and syntax of `dplyr`. This can be accessed via the help menu → cheatsheets → data transformation with `dplyr`.

Let's rewrite the select command example using the pipe operator:

```
year_country_gdp <- gapminder %>% select(year, country, gdpPercap)
print(year_country_gdp)
```

To help you understand why we wrote that in that way, let's walk through it step by step. First we summon the gapminder tibble and pass it on, using the pipe symbol `%>%`, to the next step, which is the `select()` function. In this case we don't specify which data object we use in the `select()` function since it gets that from the previous pipe.

What if we wanted to combine this with the filter example? I.e. we want to select year, country and GDP per capita, but only for countries in Europe? We can join these two operations using a pipe; feeding the output of one command directly into the next:

```
year_country_gdp_euro <- gapminder %>%
  filter(continent == "Europe") %>%
  select(year, country, gdpPercap)
print(year_country_gdp_euro)
```

Note that the order of these operations matters; if we reversed the order of the `select()` and `filter()` functions, the `continent` variable wouldn't exist in the data-set when we came to apply the filter.

What about if we wanted to match more than one item? To do this we use the `%in%` operator:

```
gapminder_scandinavia <- gapminder %>%
  filter(country %in% c("Denmark",
                        "Norway",
                        "Sweden"))
print(gapminder_scandinavia)
```

17.11 Another way of thinking about pipes

It might be useful to think of the statement

```
gapminder %>%  
  filter(continent=="Europe") %>%  
  select(year,country,gdpPercap)
```

as a sentence, which we can read as “take the gapminder data *and then* filter records where continent == Europe *and then* select the year, country and gdpPercap

We can think of the `filter()` and `select()` functions as verbs in the sentence; they do things to the data flowing through the pipeline.

17.12 Splitting your commands over multiple lines

It’s generally a good idea to put one command per line when writing your analyses. This makes them easier to read. When doing this, it’s important that the `%>%` goes at the *end* of the line, as in the example above. If we put it at the beginning of a line, e.g.:

```
gapminder_benelux <- gapminder  
%>% filter(country %in% c("Belgium", "Netherlands", "France"))
```

the first line makes a valid R command. R will then treat the next line as a new command, which won’t work.

! Challenge 1

Write a single command (which can span multiple lines and includes pipes) that will produce a tibble that has the values of `lifeExp`, `country` and `year`, for the countries in Africa, but not for other Continents. How many rows does your tibble have? (You can use the `nrow()` function to find out how many rows are in a tibble.)

```
# Edit/add/try out R code here
```

🔥 Solution to Challenge 1

```
year_country_lifeExp_Africa <- gapminder %>%
  filter(continent=="Africa") %>%
  select(year,country,lifeExp)
nrow(year_country_lifeExp_Africa)
```

As with last time, first we pass the gapminder tibble to the `filter()` function, then we pass the filtered version of the gapminder tibble to the `select()` function. **Note:** The order of operations is very important in this case. If we used ‘`select`’ first, `filter` would not be able to find the variable `continent` since we would have removed it in the previous step.

17.13 Sorting tibbles

The `arrange()` function will sort a tibble by one or more of the variables in it:

```
gapminder %>%
  filter(continent == "Europe", year == 2007) %>%
  arrange(pop)
```

We can use the `desc()` function to sort a variable in reverse order:

```
gapminder %>%
  filter(continent == "Europe", year == 2007) %>%
  arrange(desc(pop))
```

17.14 Generating new variables

The `mutate()` function lets us add new variables to our tibble. It will often be the case that these are variables we *derive* from existing variables in the data-frame.

As an example, the gapminder data contains the population of each country, and its GDP per capita. We can use this to calculate the total GDP of each country:

```
gapminder_totalgdp <- gapminder %>%
  mutate(gdp = gdpPercap * pop)
print(gapminder_totalgdp)
```

We can also use functions within `mutate` to generate new variables. For example, to take the log of `gdpPercap` we could use:

```
gapminder %>%
  mutate(logGdpPercap = log(gdpPercap))
```

The `dplyr` cheat sheet contains many useful functions which can be used with `dplyr`. This can be found in the help menu of RStudio. You will use one of these functions in the next challenge.

! Challenge 2

Create a tibble containing each country in Europe, its life expectancy in 2007 and the rank of the country's life expectancy. (note that ranking the countries *will not* sort the table; the row order will be unchanged. You can use the `arrange()` function to sort the table).

Hint: First `filter()` to get the rows you want, and then use `mutate()` to create a new variable with the rank in it. The cheat-sheet contains useful functions you can use when you make new variables (the cheat-sheets can be found in the help menu in RStudio). There are several functions for ranking observations, which handle tied values differently. For this exercise it doesn't matter which function you choose.

Can you reverse the ranking order so that the country with the longest life expectancy gets the lowest rank? Hint: This is similar to sorting in reverse order

```
# Edit/add/try out R code here
```

🔥 Solution to challenge 2

```
europeLifeExp <- gapminder %>%
  filter(continent == "Europe", year == 2007) %>%
  select(country, lifeExp) %>%
  mutate(rank = min_rank(lifeExp))
print(europeLifeExp, n=100)
```

To reverse the order of the ranking, use the `desc` function, i.e. `mutate(rank = min_rank(desc(lifeExp)))`

There are several functions for calculating ranks; you may have used, e.g. `dense_rank()`. The functions handle ties differently. The help file for `dplyr`'s ranking functions explains the differences, and can be accessed with `?ranking`

17.15 Calculating summary statistics

We often wish to calculate a summary statistic (the mean, standard deviation, etc.) for a variable. We frequently want to calculate a separate summary statistic for several groups of data (e.g. the experiment and control group). We can calculate a summary statistic for the whole data-set using the dplyr's `summarise()` function:

```
gapminder %>%
  filter(year == 2007) %>%
  summarise(meanlife = mean(lifeExp))
```

To generate summary statistics for each value of another variable we use the `group_by()` function:

```
gapminder %>%
  filter(year == 2007) %>%
  group_by(continent) %>%
  summarise(meanlife = mean(lifeExp))
```

17.16 Aside

In the examples above it would be preferable to calculate the weighted mean (to reflect the different populations of the countries). R can calculate this for us using `weighted.mean(lifeExp, pop)`. For simplicity I've used the regular mean in the above examples.

17.17 Statistics revision

If you need to revise or learn about statistical concepts, the University Library's "My Learning Essentials" team have produced a site [Start to Finish:Statistics](#) which covers important statistical concepts.

! Challenge 3

For each combination of continent and year, calculate the average life expectancy.

```
# Edit/add/try out R code here
```

Solution to Challenge 3

```
lifeExp_bycontinentyear <- gapminder %>%
  group_by(continent, year) %>%
  summarise(mean_lifeExp = mean(lifeExp))
print(lifeExp_bycontinentyear)
```

17.18 count() and n()

A very common operation is to count the number of observations for each group. The `dplyr` package comes with two related functions that help with this.

If we need to use the number of observations in calculations, the `n()` function is useful. For instance, if we wanted to get the standard error of the life expectancy per continent:

```
gapminder %>%
  filter(year == 2002) %>%
  group_by(continent) %>%
  summarize(se_pop = sd(lifeExp)/sqrt(n()))
```

Although we could use the `group_by()`, `n()` and `summarize()` functions to calculate the number of observations in each group, `dplyr` provides the `count()` function which automatically groups the data, calculates the totals and then ungroups it.

For instance, if we wanted to check the number of countries included in the dataset for the year 2002, we can use:

```
gapminder %>%
  filter(year == 2002) %>%
  count(continent, sort = TRUE)
```

We can optionally sort the results in descending order by adding `sort=TRUE`:

17.19 Equivalent functions in base R

In this course we've taught the tidyverse. You are likely come across code written others in base R. You can find a guide to some base R functions and their tidyverse equivalents [here](#), which may be useful when reading their code.

17.20 Other great resources

- [Data Wrangling tutorial](#) - an excellent four part tutorial covering selecting data, filtering data, summarising and transforming your data.
- [R for Data Science](#)
- [Data Wrangling Cheat sheet](#) - you can also access this from the help menu in RStudio (in newer versions of RStudio it has been replaced with “Data Transformation with dplyr”)
- [Introduction to dplyr](#) - this is the package vignette. It can be viewed within R using `vignette(package="dplyr", "dplyr")`
- [Data wrangling with R and RStudio](#) - 55 minute webinar from RStudio

18 R Tidyverse Exercise

18.1 Load Libraries

Load the `tidyverse` packages

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

18.2 Untidy data

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

```
who <- tidyverse::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
4 Afghanistan AF    AFG    2000      52       228
5 Afghanistan AF    AFG    2001     129       379
6 Afghanistan AF    AFG    2002      90       476
```

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.

18.3 Tidy data

Tidy data: Have each variable in a column.

Question: Are these data tidy?

💡 Expand to see solution

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
  <chr>      <chr> <chr> <dbl>        <dbl>
1 Afghanistan AF    AFG    1998       30
```

 Expand to see solution

We would replace the `new_sp_m014` with the following four columns:

```
type  sex   age   n
sp     m     014   30
```

This would place each variable in its own column.

18.4 Gather

```
stocks <- tibble(
  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       X     Y     Z
  <date>   <dbl> <dbl> <dbl>
1 2009-01-01  1.04  2.24  4.81
2 2009-01-02 -0.757 -1.09  1.71
3 2009-01-03  0.882  0.953 -5.61
4 2009-01-04 -1.96   2.95 -3.99
5 2009-01-05 -2.11  -3.01 -5.81
6 2009-01-06 -0.245 -5.92 -0.619

stocks %>% gather("stock", "price", -time) %>% head()

# A tibble: 6 x 3
  time     stock price
  <date>   <chr> <dbl>
1 2009-01-01 X      1.04
2 2009-01-02 X     -0.757
3 2009-01-03 X      0.882
4 2009-01-04 X     -1.96
```

```
5 2009-01-05 X      -2.11
6 2009-01-06 X     -0.245
```

18.5 Pivot_longer

```
stocks %>% pivot_longer(c(X,Y,Z), names_to= "stock", values_to = "price") %>%
  head()

# A tibble: 6 x 3
  time      stock  price
  <date>    <chr>  <dbl>
1 2009-01-01 X      1.04
2 2009-01-01 Y      2.24
3 2009-01-01 Z      4.81
4 2009-01-02 X     -0.757
5 2009-01-02 Y     -1.09
6 2009-01-02 Z      1.71
```

18.6 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
  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
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   iso3   year demo      n
  <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    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    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      n
  <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    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    1997 new_sp_m5564     2
```

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?

 Expand to see solution

```
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
  country     iso2   iso3   year type sex   age_range     n
  <chr>      <chr>  <chr>  <dbl> <chr> <chr> <chr>       <dbl>
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
```

18.7 Conclusion

Now our untidy data are tidy.

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

```
# A tibble: 6 x 8
  country     iso2   iso3   year type sex   age_range     n
  <chr>      <chr>  <chr>  <dbl> <chr> <chr> <chr>       <dbl>
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
```

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

18.8 Acknowledgment

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

https://people.duke.edu/~ccc14/cfar-data-workshop-2018/CFAR_R_Workshop_2018_Exercises.html

19 R Recoding Reshaping Exercise

19.1 Key points

Here are some key points regarding recoding and reshaping data in R:

- Count the number of times ID2 is duplicated
 - `sum(duplicated(b$ID2))`
- List all rows with a duplicated c1 value
 - `f %>% group_by(c1) %>% filter(n()>1)`
- Recode data using `left_join`
- Pivot data from long to wide
 - `pivot_wider`
- Pivot data from wide to long
 - `pivot_longer`
- Useful table commands
 - `table()`
 - `addmargins(table())`
 - `prop.table(table(), margin)`

19.2 Load Libraries

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

19.3 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.

```
# Download file within the WebR environment
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
download.file(paste0(root_srcfile,"data/exercise.RData"),
               "exercise.RData")
load("exercise.RData", verbose = TRUE)
dim(ds)
names(ds)
dim(dd)
names(dd)
```

19.4 Exercise 1: duplicated values

Skill: Checking for duplicated IDs

```
ds %>% select(subject_id, sample_id, height) %>% head(n=10)
```

Check if there are any duplicated `sample_id`'s using the `duplicated` command. If so, count how many duplicated `sample_id`'s there are.

```
# Edit/add/try out R code here
```

 Expand to see solution

```
sum(duplicated(ds$sample_id))
```

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

```
# Edit/add/try out R code here
```

 Expand to see solution

Note that it is important to be aware of missing IDs. So when constructing tables of counts using the `table` command, the `useNA` argument controls if the table includes counts of NA values.

```
# Count how many times each sample_id occurs
# ignoring NA's
tail(table(ds$sample_id))
# Count how many times each sample_id occurs
# including NA's
tail(table(ds$sample_id, useNA="always"))
# Table of the different number of times sample_id's are repeated
table(table(ds$sample_id))
# But why do we get differing numbers here?
sum(duplicated(ds$sample_id))
35+13*2+2*3+1*4
sum(duplicated(ds$sample_id, incomparables = NA))
```

How many `sample_id`'s are NA's?

```
sum(is.na(ds$sample_id))

table(table(ds$sample_id, useNA="always"))
36+13*2+2*3+1*4
```

Check if there are any duplicated `subject_ids`

```
# Edit/add/try out R code here
```

 Expand to see solution

We can check if there are any duplicated `subject_id`'s by counting how many duplicates there are.

```
sum(duplicated(ds$subject_id))
```

19.5 Checking for duplicates

How do we return every row that contains a duplicate?

This approach only does not return every row that contains a duplicated ID:

```
f <- data.frame(ID=c(1,1,2),c2=c(1,2,3))
f
f[duplicated(f$ID),]
```

19.6 Counting the number of occurrences of the ID

```
f %>% group_by(ID) %>% summarise(n=n())
f %>% group_by(ID) %>% count()
```

19.7 Count sample_id duplicates

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

```
# Edit/add/try out R code here
```

 Expand to see solution

```
ds %>% group_by(sample_id) %>%
  summarise(n=n()) %>%
  filter(n>1) %>%
  arrange(desc(n)) %>%
  head()

ds %>% group_by(sample_id) %>%
  summarise(n = n()) %>% filter(n > 1) %>%
  arrange(desc(n)) %>% pull(n) %>% table()
```

19.8 Checking for duplicates

Here we list all of the rows containing a duplicated ‘ID’ value using functions from the ‘tidyverse’ package:

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

19.8.1 How to list all duplicates

Use Tidyverse commands to list (1) all duplicates for `sample_id` and (2) all duplicates for `subject_id`. Sort the results by the ID.

```
# Edit/add/try out R code here
```

 Expand to see solution

19.8.2 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()
```

19.8.3 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)
```

19.9 Exercise 2: Reshaping data

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”.

```
# Edit/add/try out R code here
```

 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
head(b2)

# Trimester 1 Gestationalage_sample values for SAMP149
glimpse(b2[1,"1"])
```

19.9.1 Comment

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.

19.10 Exercise 3: Aggregating data

Skill: Aggregating data

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

```
# Edit/add/try out R code here
```

 Expand to see solution

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

19.10.1 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.

```
prop.table(table(ds$case_control_status,ds$race),  
          margin = 1)  
  
prop.table(table(ds$case_control_status,ds$race))
```

Construct more readable tables with labels using `xtabs`

```
# Edit/add/try out R code here
```

 Expand to see solution

19.10.2 xtabs table with labels

```
prop.table(xtabs(~ case_control_status + race, data = ds),  
           margin = 1)
```

Create a count cross table using Tidyverse commands

```
# Edit/add/try out R code here
```

 Expand to see solution

```
ds %>%  
  group_by(case_control_status, race)%>%  
  summarize(n=n())%>%  
  spread(race, n)  
  addmargins(xtabs(~ case_control_status + race, data = ds))
```

Create a proportion cross table using Tidyverse commands

```
# Edit/add/try out R code here
```

 Expand to see solution

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

19.11 Exercise 4: Summarizing within groups

Skill: Summarizing within groups

Apply the `summary` command to the “Gestationalage_sample” within each “Sample_trimester” group.

```
# Edit/add/try out R code here
```

 Expand to see solution

```
f <- split(ds[, "Gestationalage_sample"], ds$Sample_trimester)
sapply(f, summary)

# Or 'tapply' can be used:
tapply(ds$Gestationalage_sample, ds$Sample_trimester, summary)
```

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

19.12 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)  
  
# Edit/add/try out R code here
```

 Expand to see solution

```
a5 <- ds  
a5$ID <- DictPer[a5$subject_id]  
a5 %>% select(subject_id, ID) %>% head  
head(DictPer)
```

19.13 Recoding data

Approach 2

- Implement our dictionaries using left joins

19.13.1 Comment

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

```
# Edit/add/try out R code here
```

 Expand to see solution

```
key <- data.frame(SubjectID=names(DictPer), ID=DictPer)  
head(key)  
b5 <- left_join(ds, key, by=c("subject_id" = "SubjectID"))  
b5 %>% select(subject_id, ID) %>% head()
```

19.14 Exercise 6: Filtering rows

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.

Edit/add/try out R code here

💡 Expand to see solution

```
tri1 <- ds %>% filter(Sample_trimester==1)
tri1 %>% select(subject_id, sample_id, Sample_trimester) %>% head()
tri2 <- ds %>% filter(Sample_trimester==2)
tri2 %>% select(subject_id, sample_id, Sample_trimester) %>% head()
```

19.15 Exercise 7

Skill: Selecting columns

Update `tri1` and `tri2` to only contain the three columns “sample_id”, “Sample_trimester”, “Gestationalage_sample”

Edit/add/try out R code here

💡 Expand to see solution

```
tri1 <- tri1 %>% select(sample_id, Sample_trimester, Gestationalage_sample)
head(tri1)
tri2 <- tri2 %>% select(sample_id, Sample_trimester, Gestationalage_sample)
head(tri2)
```

20 R Merging Exercise

20.1 Merging Best Practice

- *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**, before and after the merge, to make sure the merged object has the expected number of rows and columns.
- Always explicitly name the keys you are merging on.

20.2 Load Libraries

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

20.3 Input data

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

```
# Download file within the WebR environment
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
download.file(paste0(root_srcfile,"data/project1.RData"),
```

```
"project1.RData")
load("project1.RData", verbose = TRUE)
dim(ds)
dim(dd)
```

20.4 Select a subset of subject-level fields

Set up a data frame ‘a’ that has these subject-level fields: “subject_id” “maternal_age_delivery” “case_control_status” “pregnancy_BMI”

```
a <- ds %>%
  select("subject_id",
         "maternal_age_delivery",
         "case_control_status",
         "pregnancy_BMI") %>%
  arrange(subject_id)
dim(a)
head(a,10)
tail(a)
```

20.5 Unique records

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

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

```
# Edit/add/try out R code here
```

 Expand to see solution

```
dim(a)
b <- unique(a)
dim(b)
head(b)

b1 <- a %>% distinct()
dim(b1)

all.equal(b,b1)
all.equal(b,b1, check.attributes=FALSE)
head(rownames(b))
head(rownames(b1))
# Reset row names
rownames(b) <- NULL
rownames(b1) <- NULL
all.equal(b,b1)
```

20.5.1 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.

Note that the `dplyr` R package provides the `distinct` command, which keeps only unique/distinct rows from a data frame. It is faster than the `unique` command.

```
(ex1 <- data.frame(ID=c(1,1,1,2),trait=c(10, 9, 9, 11)))
unique(ex1)

ex1 %>% distinct()
```

20.6 Check that the `subject_id`'s are now not duplicated

Are the `subject_id`'s unique?

```
# Edit/add/try out R code here
```

 Expand to see solution

```
sum(duplicated(b$subject_id))
b %>%
  group_by(subject_id) %>%
  filter(n() > 1)
```

20.7 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.

```
# Edit/add/try out R code here
```

 Expand to see solution

```
set.seed(10234)
b$ID <- sample(c(1:nrow(b)), replace = FALSE)
head(b %>% select(subject_id, ID))
sum(duplicated(b$ID))
```

20.8 Merge in new phenotype information

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

```
# Download file within the WebR environment
root_srcfile <- "https://raw.githubusercontent.com/DanielEWeeks/HuGen2071/main/"
download.file(paste0(root_srcfile, "data/newtrait.tsv"),
              "newtrait.tsv")
new <- read_tsv("newtrait.tsv")
head(new)
dim(new)
dim(b)
```

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.

20.9 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`.

20.10 Merge in new phenotype information

Carefully merge in the new data in using tidyverse commands. As this is subject-level information, it should be merged into the subject-level data frame `b` which was created above when from your data frame ‘`a`’ you selected only the unique records.

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.

```
# Edit/add/try out R code here
```

 Expand to see solution

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

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

# Check for missing IDs
sum(is.na(b$subject_id))
sum(is.na(new$subject_id))

# Check the dimensions
dim(b)
dim(new)
b2 <- left_join(b, new, by="subject_id")
head(b2)
dim(b2)
b3 <- full_join(b, new, by="subject_id")
dim(b3)
```

20.11 Further checks

When merging data based on an ID shared in common, it is not only important to check for duplicated IDs, but it is also important to check for overlap of the two ID sets.

Check if the set of `subject_id` IDs in your dataframe `b` fully overlaps the set of `subject_id` IDs in the `new` data set. If there is not full overlap, document which IDs do not overlap.

Hint: Use an `anti_join`.

```
# Edit/add/try out R code here
```

 Expand to see solution

`anti_join()` return all rows from `x` without a match in `y`.

```
# Tally how many of b's subject_id's are in new  
table(b$subject_id %in% new$subject_id)  
# Tally how many of new's subject_id's are in b  
table(new$subject_id %in% b$subject_id)  
# List the b's subject_id's that are not in new  
b$subject_id[!(b$subject_id %in% new$subject_id)]  
# List the new's subject_id's that are not in b  
new$subject_id[!(new$subject_id %in% b$subject_id)]  
  
# Simpler to do this with anti_join's  
anti_join(b, new, by="subject_id")  
anti_join(new, b, by="subject_id")
```

21 R Graphics Exercise

21.1 The Grammar of Graphics

This slide set by Garrick Aden-Buie provides a nice introduction to plotting with ggplot2:

A Gentle Guide to the Grammar of Graphics with ggplot2

Slides: <https://gadenbuie.github.io/gentle-ggplot2>

Source: <https://github.com/gadenbuie/gentle-ggplot2>

21.2 Load Libraries

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

21.3 Exercise 1

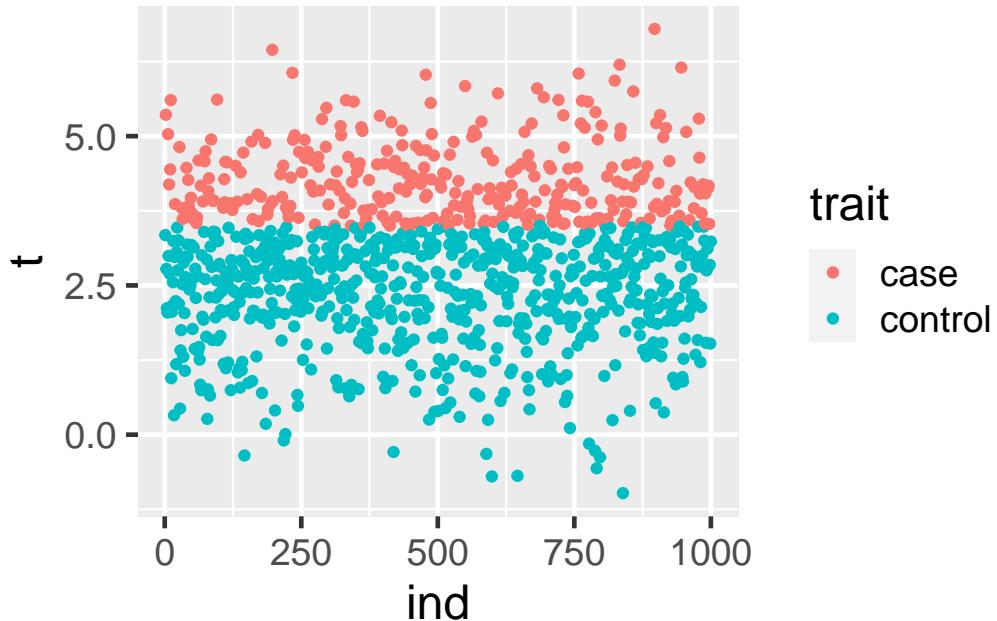
Read in and set up the data set **b**, a cleaned version of our simulated data set:

```
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="/")
```

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()
```

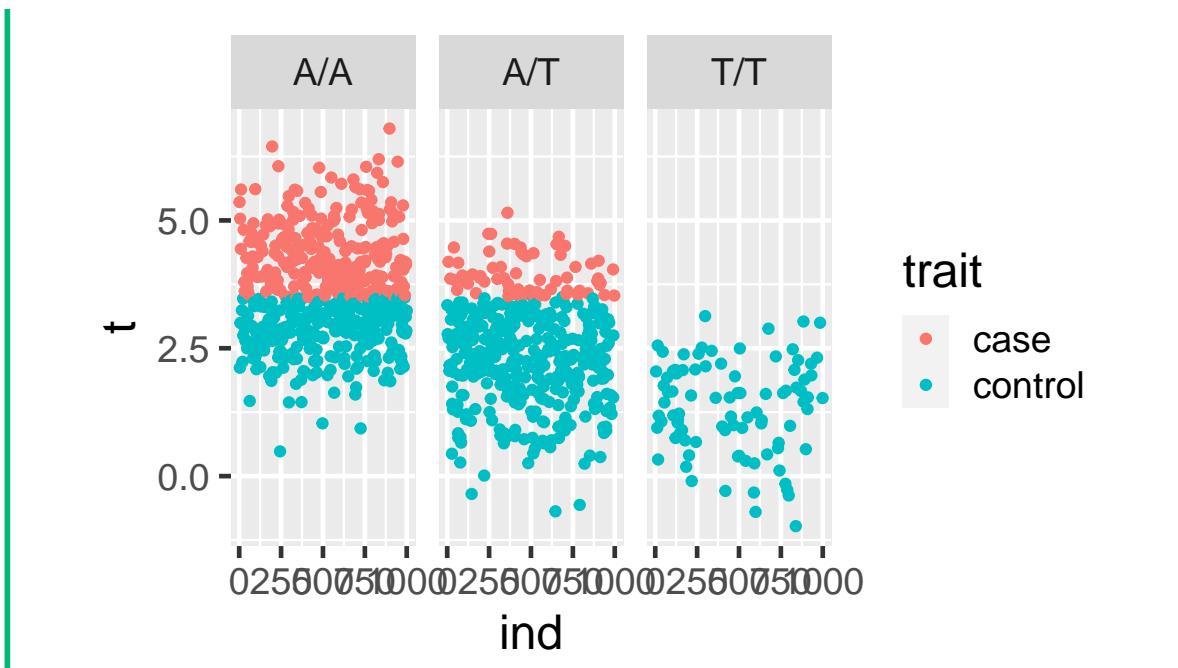


21.4 Exercise 2

Using ggplot, plot `ind` vs. `t`, coloring by case-control status (`trait`) and facetting 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)
```



21.5 Facetting

Facetting can be done using the classic interface, where formula notation is used to indicate rows (before the ~) and columns (after the ~). According to the `facet_grid` documentation

“the dot in the formula is used to indicate there should be no facetting on this dimension (either row or column)”

So this will facet in columns by `geno`:

```
facet_grid(~ geno)
```

This will facet in rows by `geno`:

```
facet_grid(geno ~ .)
```

The current recommended notation for facetting instead clearly names the rows and columns that you’d like to facet on.

This will facet in columns by `geno`:

```
facet_grid(cols = vars(geno))
```

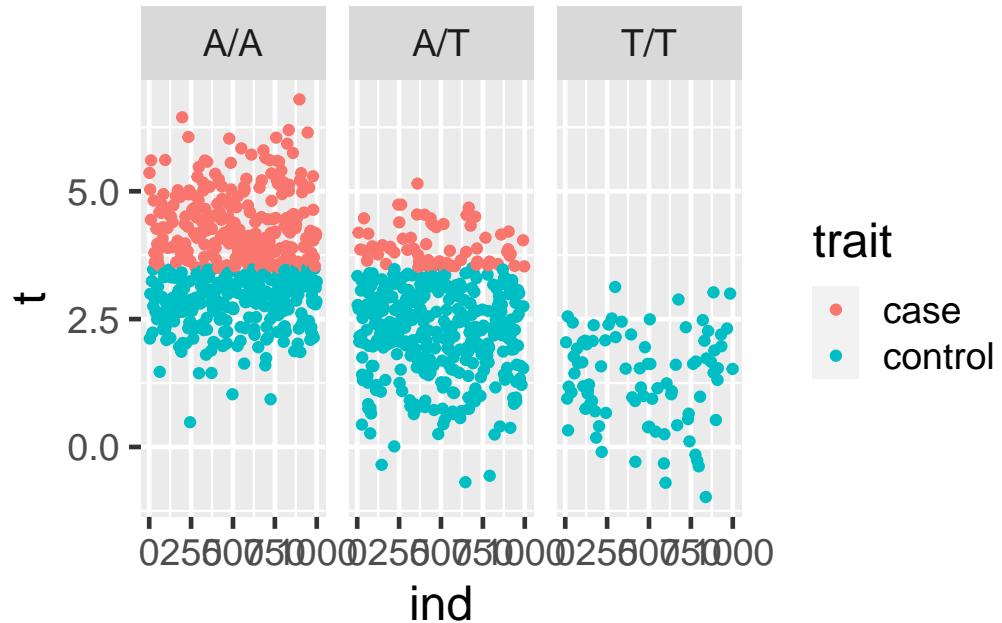
This will facet in rows by geno:

```
facet_grid(rows = vars(geno))
```

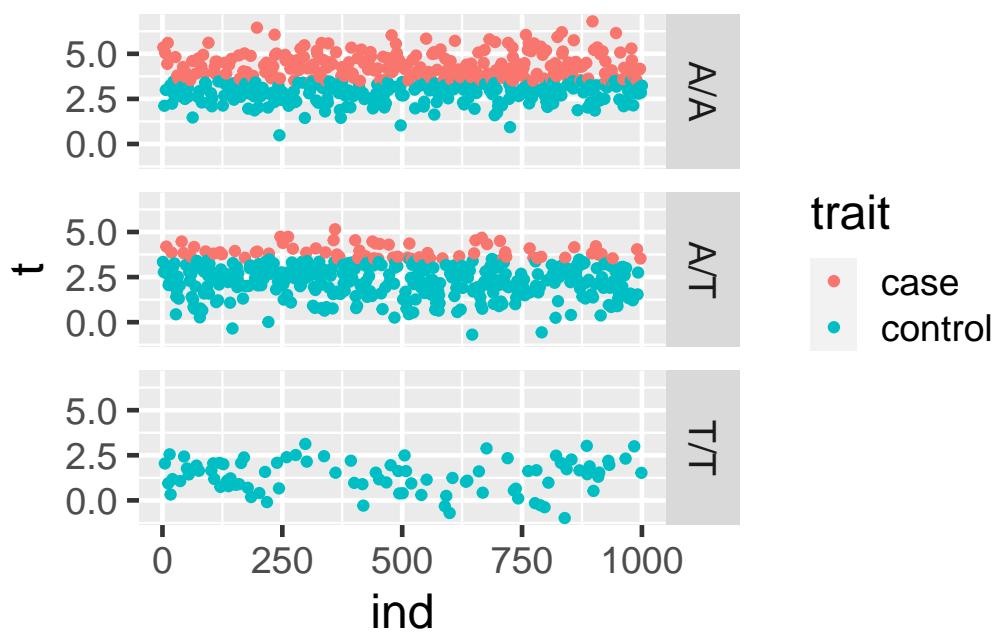
Try these various ways of facetting out.

💡 Expand to see solution

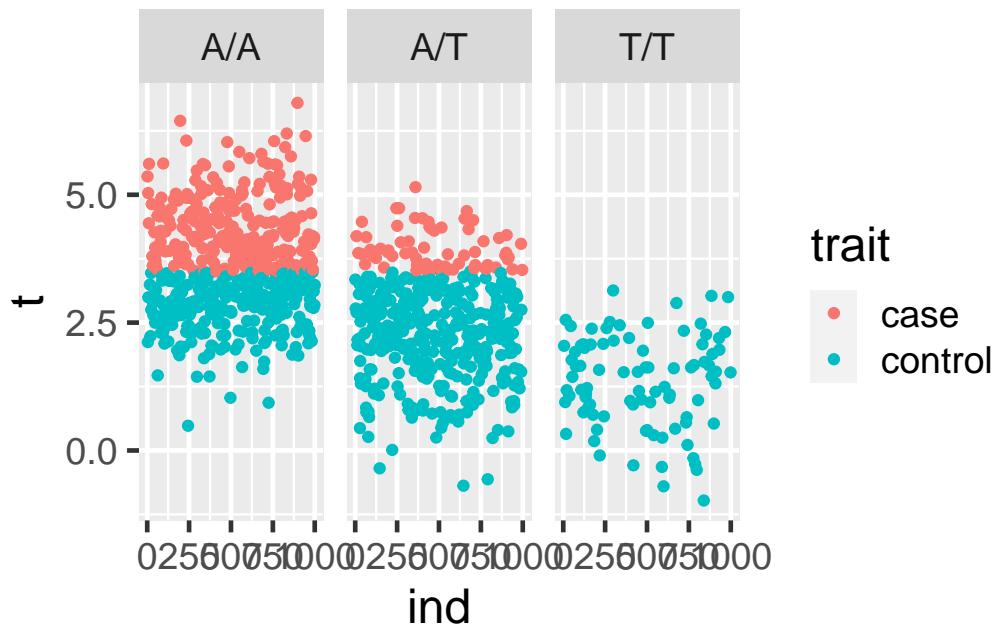
```
g <- ggplot(data=b, aes(x=ind, y=t, color=trait)) +  
  geom_point()  
g +  
  facet_grid(. ~ geno)
```



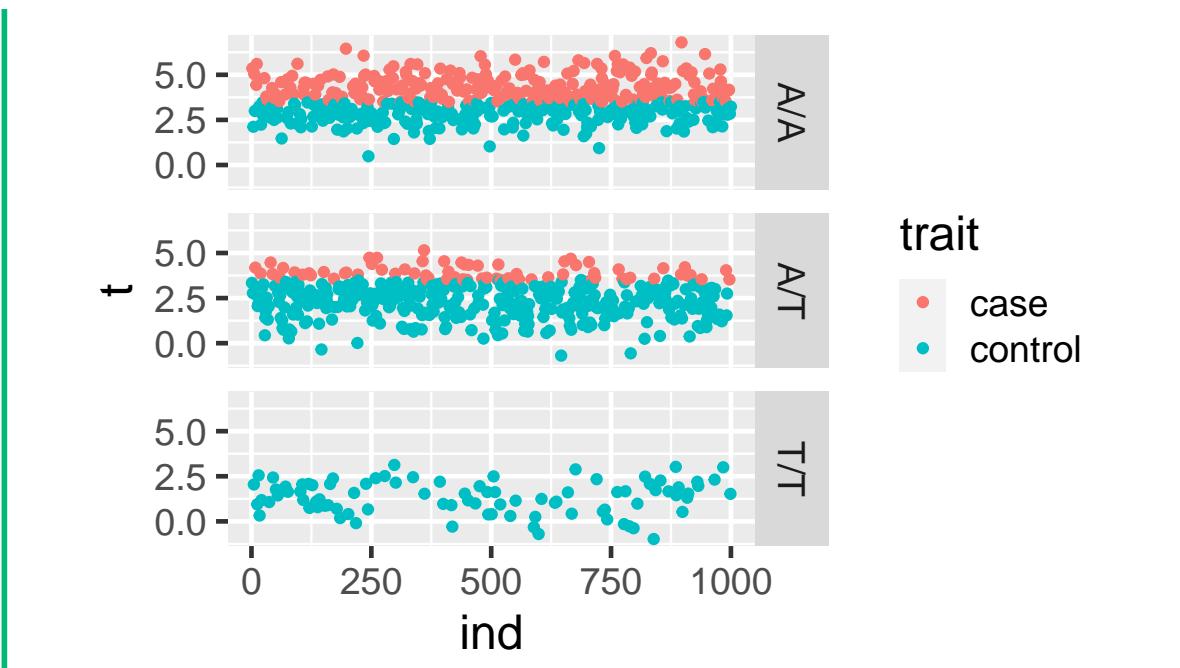
```
g + facet_grid(geno ~ .)
```



```
g + facet_grid(cols = vars(geno))
```



```
g + facet_grid(rows = vars(geno))
```



21.6 Always plot your data

```
library(tidyverse)
d <- read_tsv("data/example.tsv")
```

```
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 <- rep(c("x","y"), 13)
n2 <- c("", "", rep("_", 24))
n3 <- c("", "", c(sort(rep(c(1:12), 2))))
names(d) <- 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"
```

21.7 Similar regression lines

These three data sets have very similar regression lines:

```
summary(lm(x ~ y, data=d)) %>% coef()
```

	Estimate	Std. Error	t value	Pr(> t)
(Intercept)	56.17563819	2.87986960	19.5063131	9.435087e-42
y	-0.03991951	0.05250204	-0.7603419	4.483288e-01

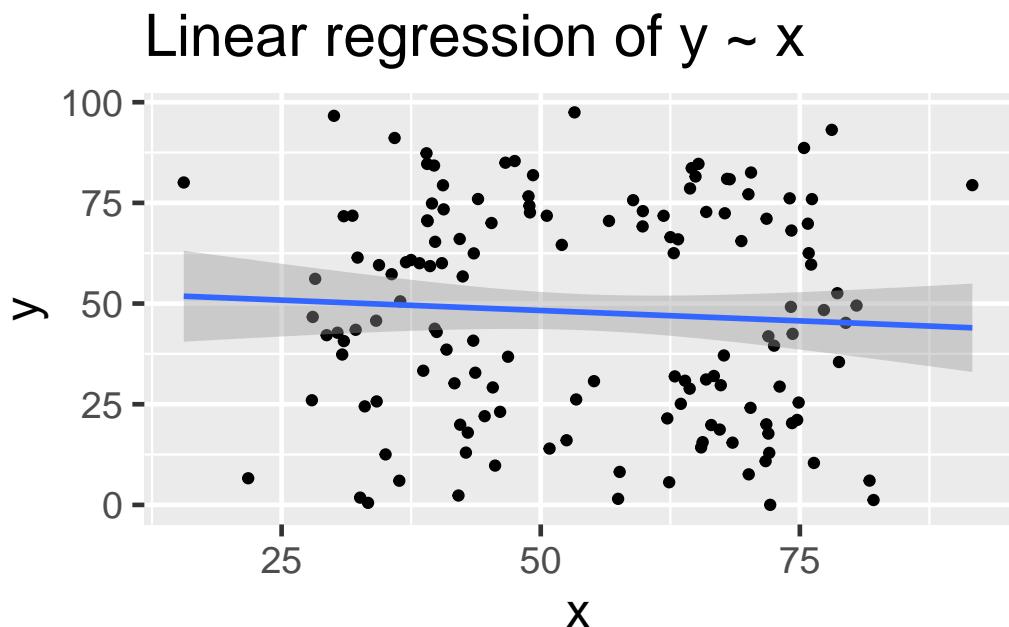
```
summary(lm(x_1 ~ y_1, data=d)) %>% coef()
```

	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")
```



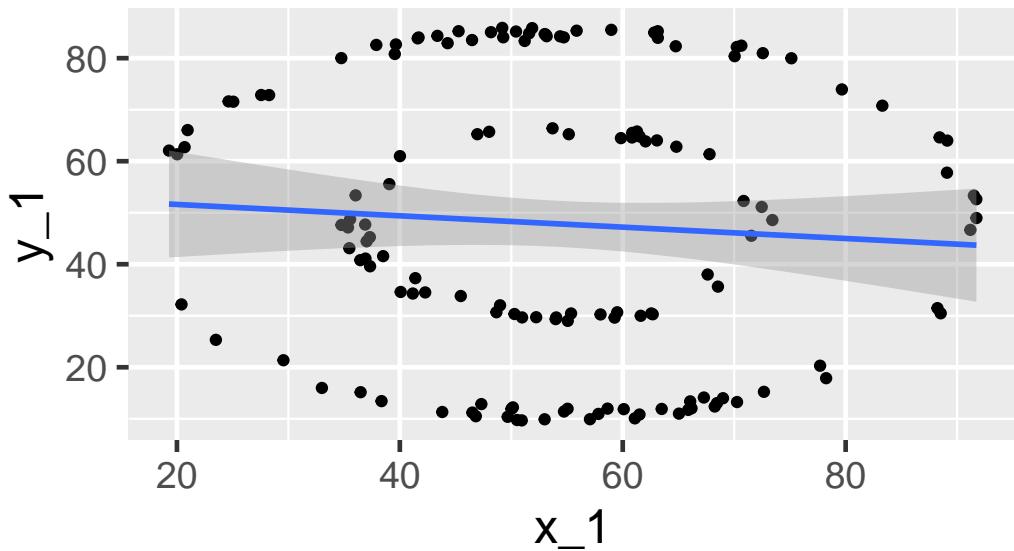
Now try this:

```
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



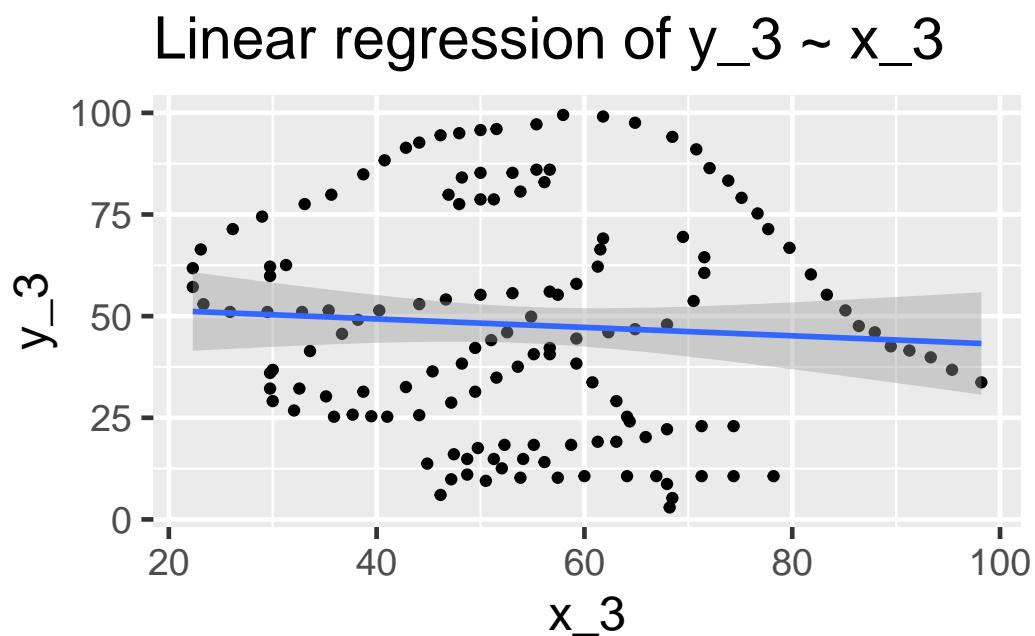
And now try this:

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

 Expand to see solution

21.7.1 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'
```



21.8 Always plot your data

```
f <- read_tsv("data/BoxPlots.tsv")
# Delete the first column
f <- f[,-1]
head(f)

# A tibble: 6 x 5
  left lines normal right split
  <dbl> <dbl> <dbl> <dbl> <dbl>
1 -9.77 -9.77 -9.76 -9.76 -9.77
2 -9.76 -9.74 -9.72 -9.05 -9.77
3 -9.75 -9.77 -9.68 -8.51 -9.77
4 -9.77 -9.77 -9.64 -8.24 -9.77
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
```

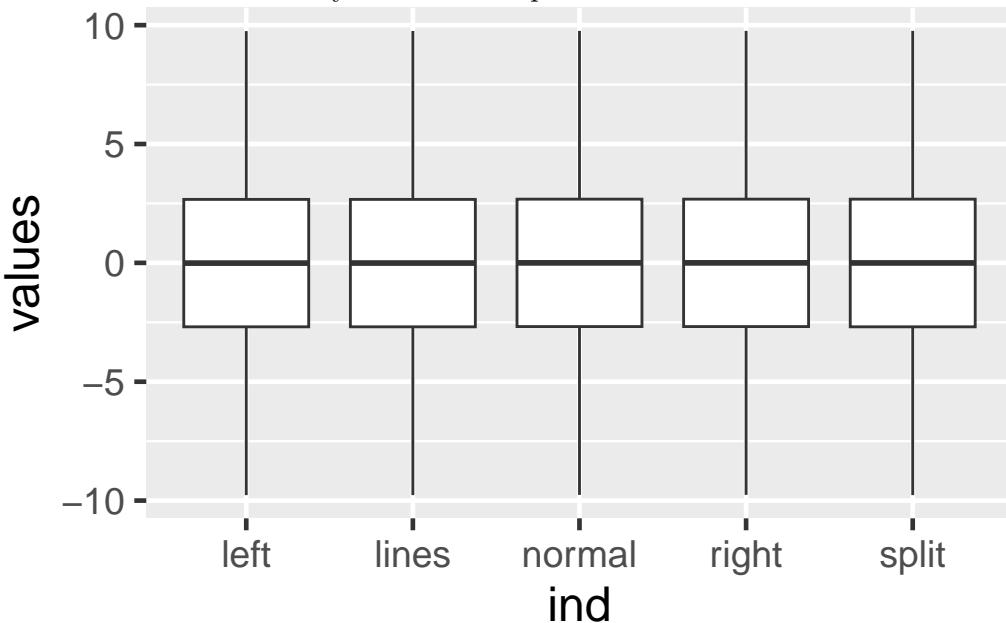
Now try this:

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

 Expand to see solution

21.9 Identical box plots

These data have essentially identical box plots.



21.10 Boxplots

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

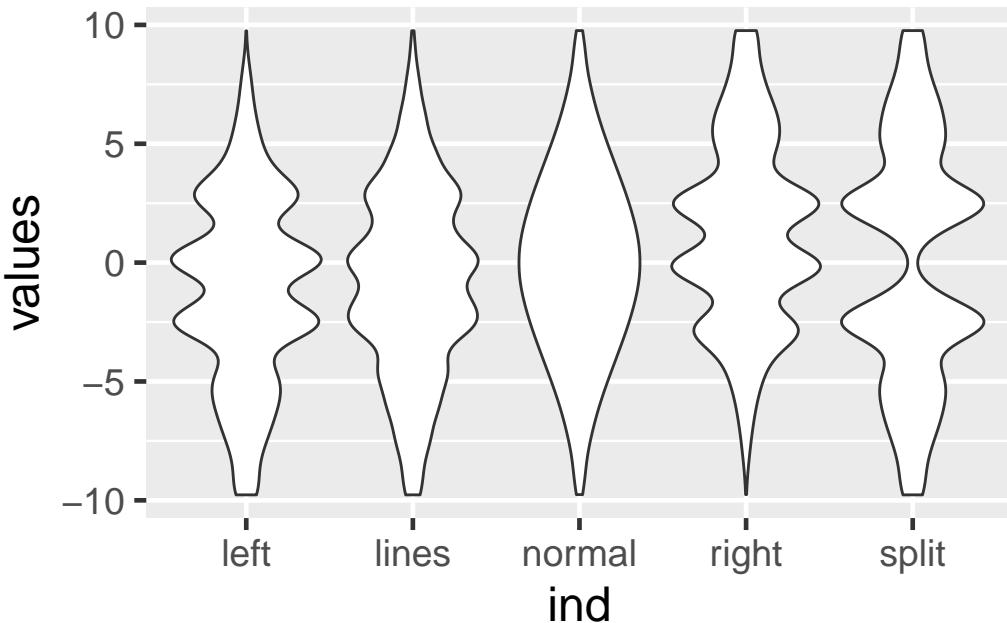
Let's try violin plots instead:

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

A violin plot is a mirrored density plot.

💡 Expand to see solution

21.11 Non-identical violin plots



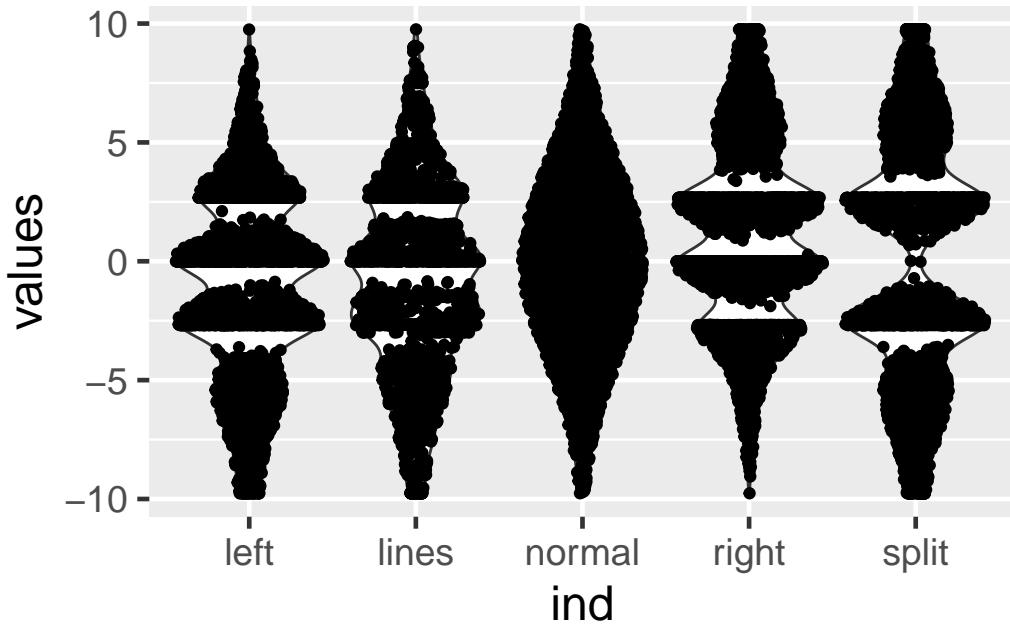
21.12 Sina plots

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()
```

 Expand to see solution

21.13 Sina plots



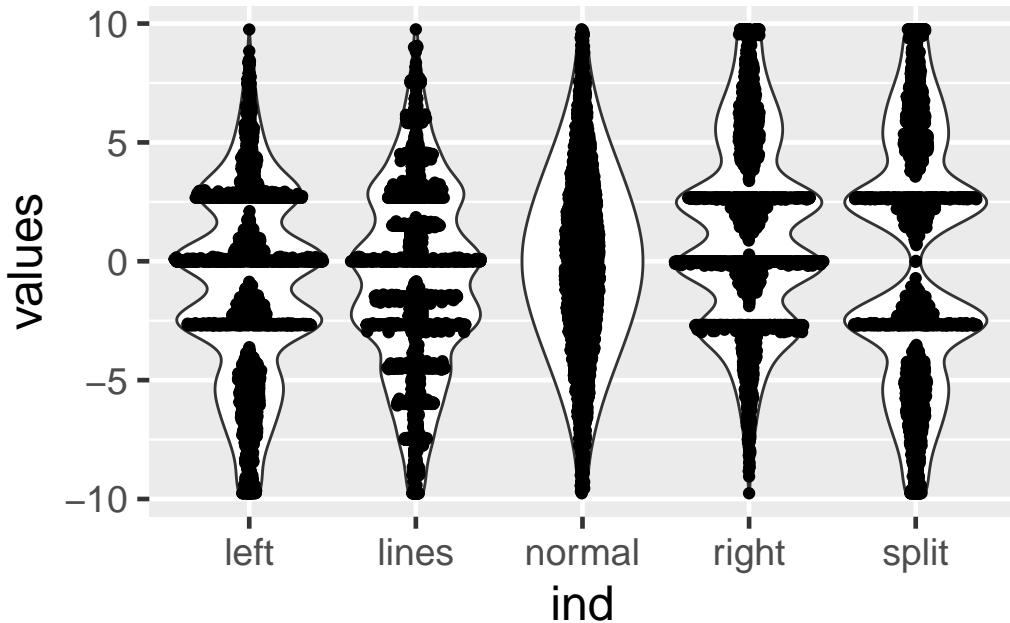
21.14 Sina plots

`method == "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")
```

 Expand to see solution

21.15 Sina plots



21.16 Raincloud plots

Raincloud plots can be created by using the `geom_rain` geometry from the `ggrain` R package.

“These”raincloud plots” can visualize raw data, probability density, and key summary statistics such as median, mean, and relevant confidence intervals in an appealing and flexible format with minimal redundancy.”

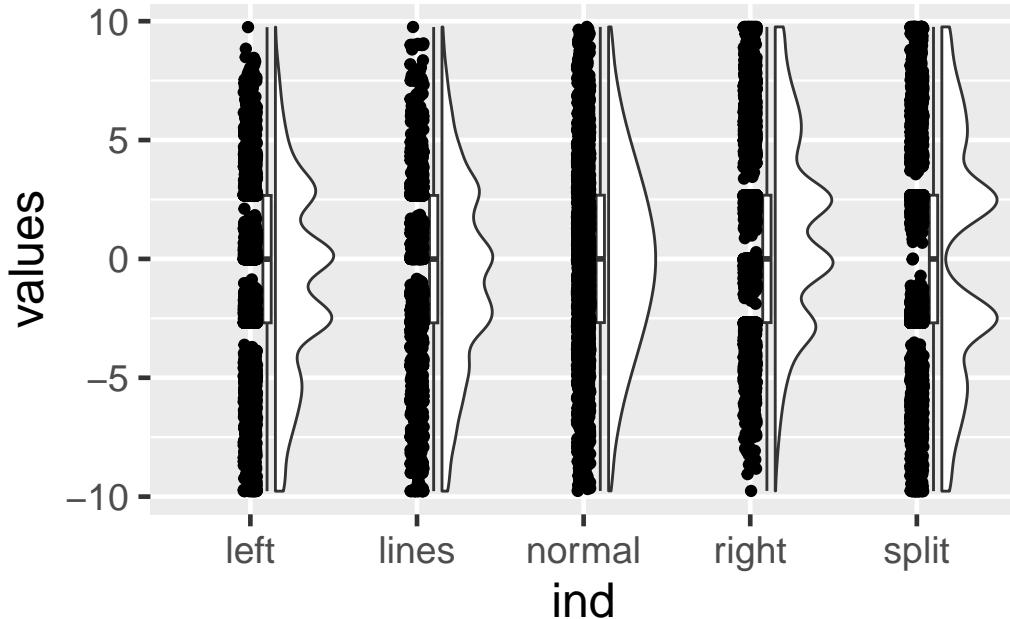
Allen M, Poggiali D, Whitaker K, Marshall TR, Van Langen J, Kievit RA. Raincloud plots: a multi-platform tool for robust data visualization. Wellcome Open Res. 2021 Jan 21;4:63. PMID: 31069261 PMCID: PMC6480976 DOI: <https://doi.org/10.12688/wellcomeopenres.15191.2>

<https://github.com/RainCloudPlots/RainCloudPlots>

Try it out.

 Expand to see solution

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



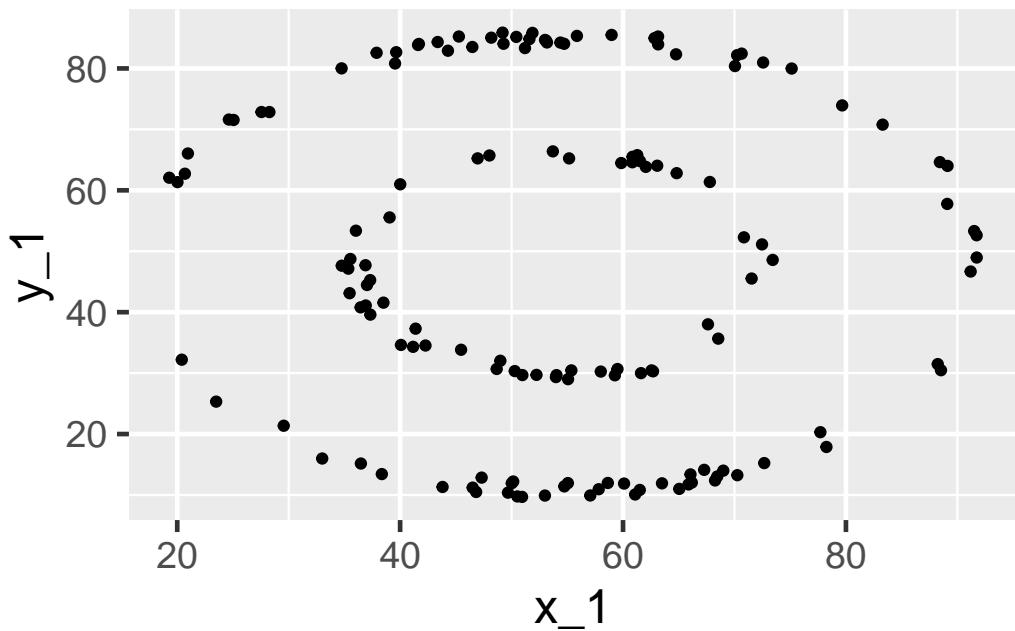
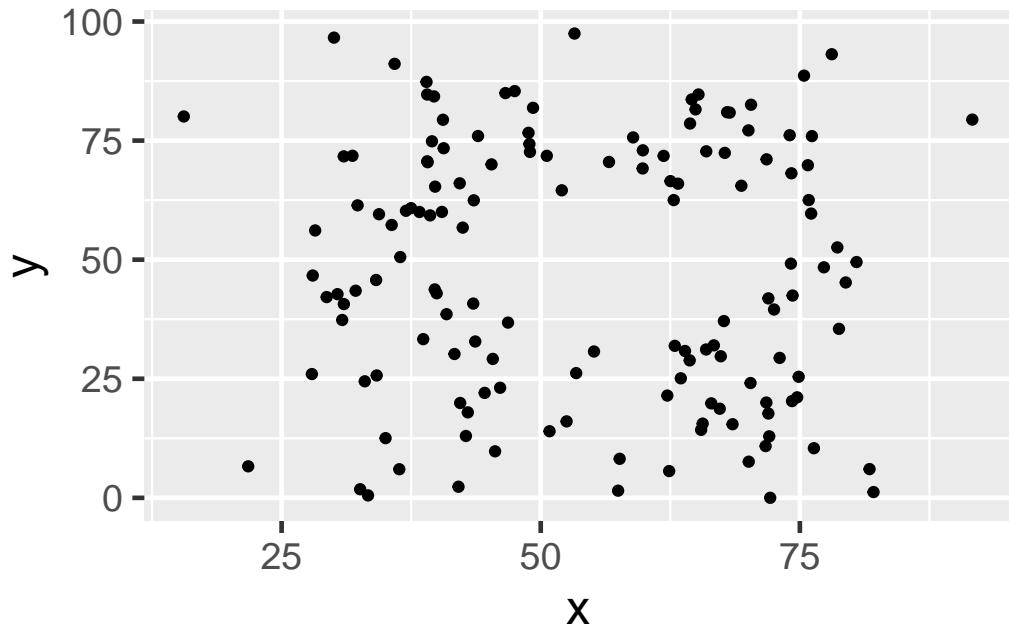
21.17 Drawing multiple graphs

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:

```
x.names <- c("x", "x_1")
y.names <- c("y", "y_1")
for (i in 1:2) {
  x.nam <- sym(x.names[i])
  y.nam <- sym(y.names[i])
  print(ggplot(data=d, aes(x = {{ x.nam }},
```

```
    y = {{ y.nam }})) +  
  geom_point())  
}
```



21.18 Writing ggplot functions

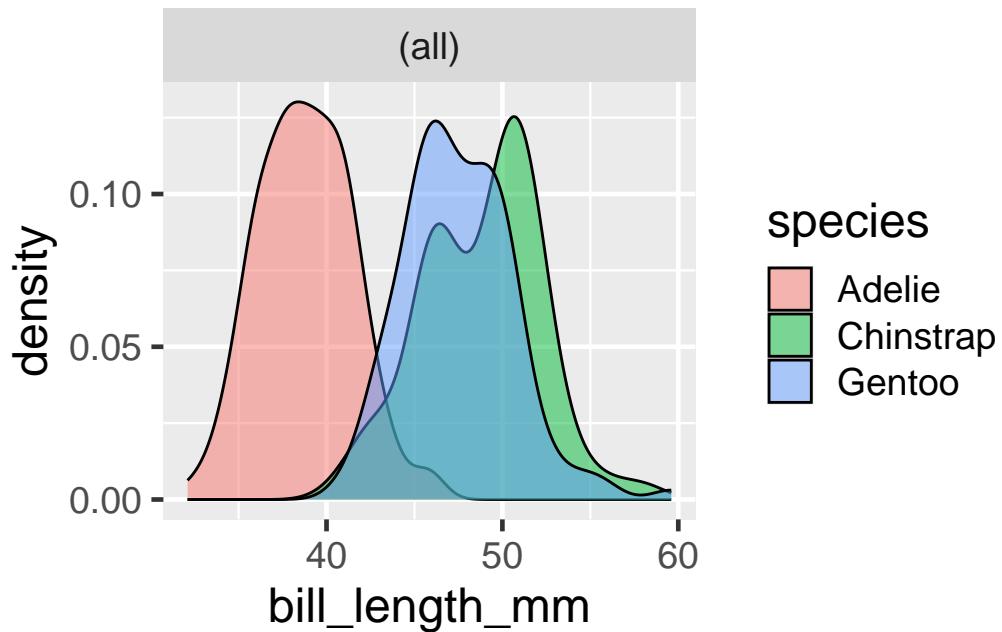
See <https://r4ds.hadley.nz/functions.html#plot-functions>

```
library(palmerpenguins)

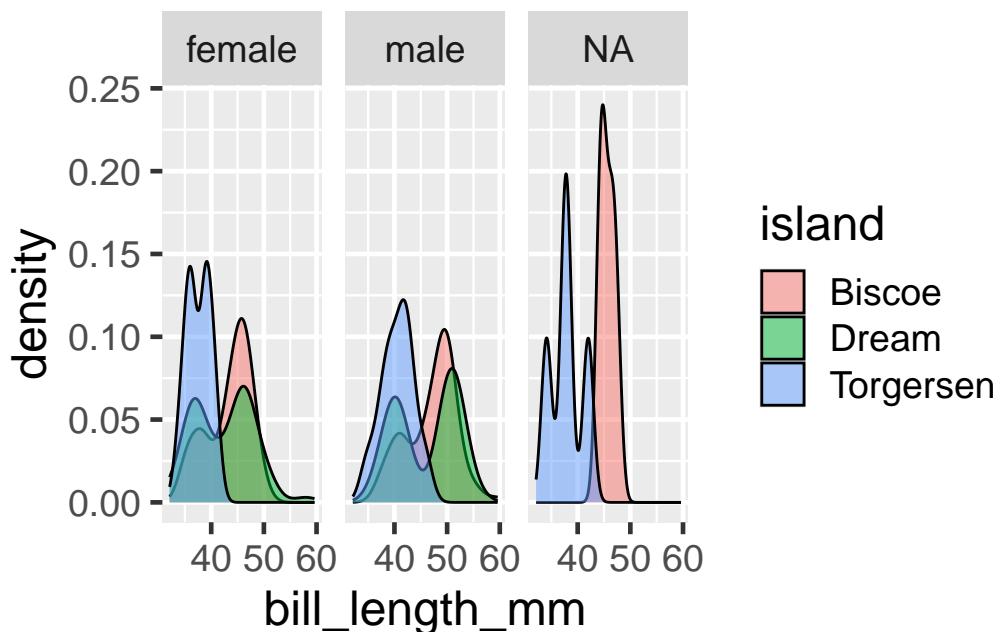
PlDensity <- function(fill, ...) {
  ggplot(penguins %>% filter(!is.na(bill_length_mm)),
         aes(bill_length_mm, fill = {{ fill }})) +
    geom_density(alpha = 0.5) +
    facet_wrap(vars(...))
}
```

Example from: https://twitter.com/yutannihilat_en/status/1574387230025875457?s=20&t=FLbwErwEKQKWtKIGufDLIQ

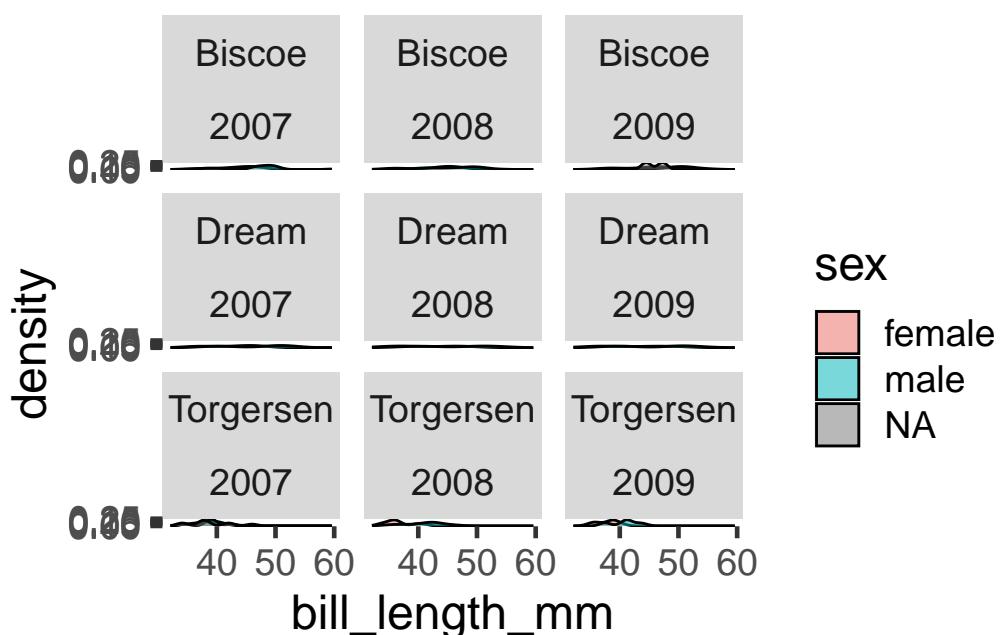
```
PlDensity(species)
```



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



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



21.19 Exercise 3

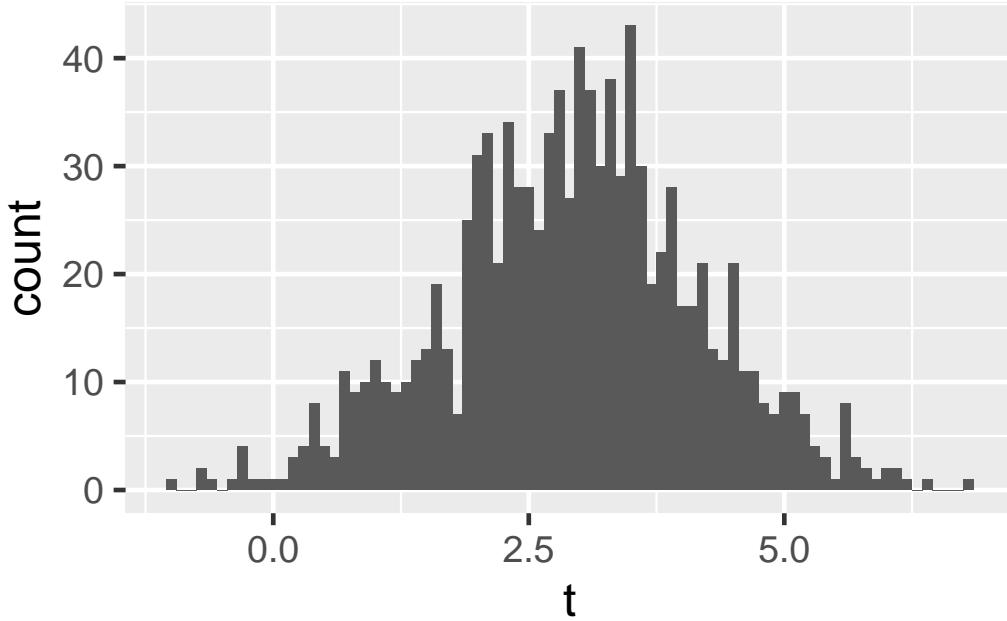
Consider this example code:

```
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)
```



21.19.1 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` faceted by `geno` using the data set `b` that we set up above.

Hints

- See <https://r4ds.hadley.nz/functions.html#plot-functions>
- Use the `vars()` approach

Expand to see solution

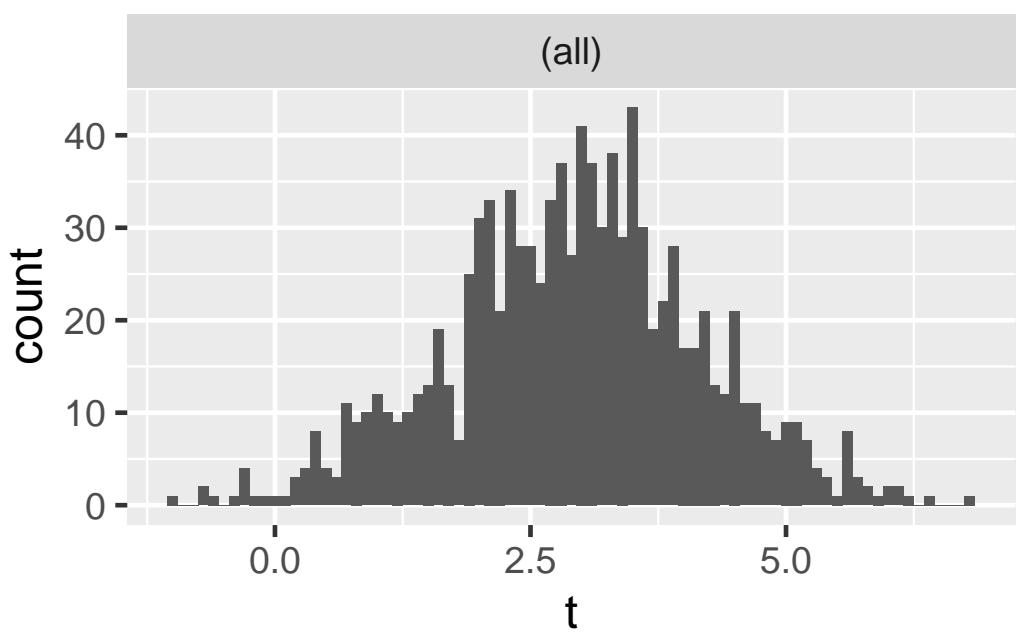
Hadley Wickham states:

You have to use the `vars()` syntax

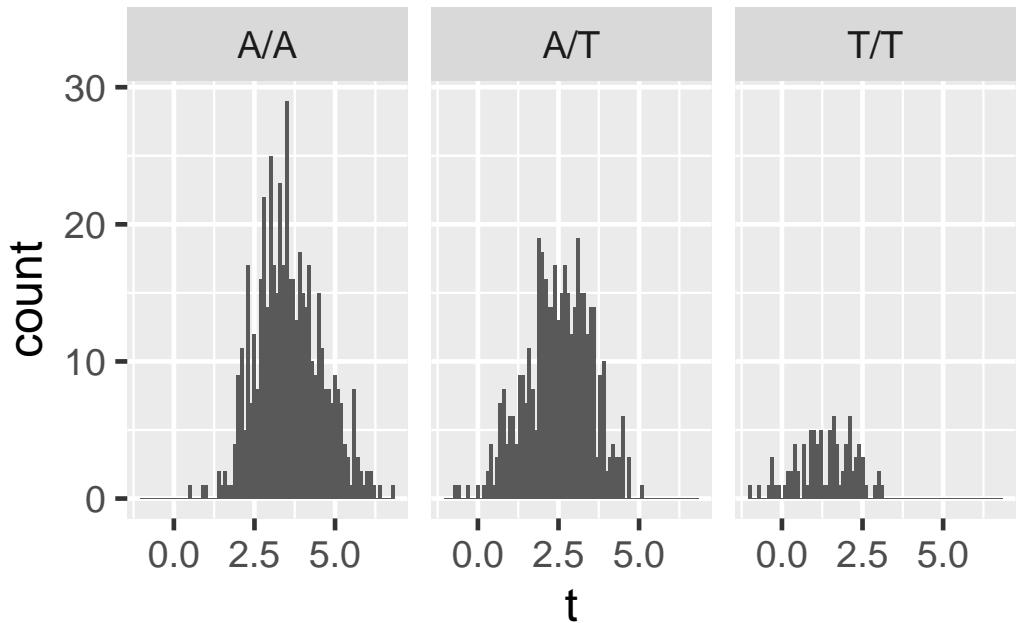
```
foo <- function(x) {  
  ggplot(mtcars) +  
    aes(x = mpg, y = disp) +  
    geom_point() +  
    facet_wrap(vars({{ x }}))  
}
```

Tweet: https://twitter.com/hadleywickham/status/1574380137524887554?s=20&t=F_LbwErwEKQKWtKIGufDLIQ

```
histogram <- function(df, var, binwidth, grp) {  
  df |>  
    ggplot(aes({{ var }})) +  
    geom_histogram(binwidth = binwidth) +  
    facet_wrap(vars({{ grp }}))  
}  
  
histogram(b, t, 0.1)
```



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



21.20 Source of data

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

22 R Reordering Exercise

22.1 Load Libraries

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

22.2 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")
# Set up data
ds <- as.data.frame(t(dd %>%
  arrange(VARNAME)))
names(ds) <- letters
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)
idx <- 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"))
  dim(dd)

[1] 27  2

  head(dd)

  VARNAME      TYPE
1      c numeric
2      m numeric
3      f numeric
4      e numeric
5      a numeric
6      d numeric

  dim(ds)

[1] 2 27

  head(ds[1:3])

  ID                  b                  z
1 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"

```

22.3 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.

22.4 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
```

22.5 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))
```

```
[1] TRUE
```

22.6 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"
```

22.7 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
```

22.8 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
```

22.9 Question: use select?

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

💡 Expand to see solution

```
# Transpose so rows become columns, and then we can use 'select' to rearrange
# those columns, and then transpose back, and rename columns as needed.
dd5 <- dd %>%
  t() %>%
  as_tibble(.name_repair = "unique") %>%
  select(colnames(ds)) %>%
  t() %>%
  as.data.frame() %>%
  rename(VARNAME = "V1", TYPE = "V2")

select: columns reordered (ID, b, z, a, p, ...)
rename: renamed 2 variables (VARNAME, TYPE)

all.equal(dd5\$VARNAME, colnames(ds))

[1] TRUE
```

22.10 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>

23 R Exploratory Data Analysis Exercise

24 Exploratory Data Analysis

- Goal: Understand your data
- Ask questions
 - Understand each phenotype
 - Understand how each phenotype varies
 - Understand how the phenotypes are related to each other
 - Understand how the data are organized
- Plot often, plot everything!

24.1 Load Libraries

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

24.2 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

Project 1 Questions

- Which of the measurements are sample-specific?
- Which are subject-specific?
- How to structure the data for sharing via dbGaP?

24.3 Dimensions

- What are the dimensions of our data?

24.4 Dimensions

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

 Expand to see solution

24.4.1 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] "pregnancy_weight"      "height"
[11] "pregnancy_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"
[21] "baby_sex"              "baby_birthweight_centile"
[23] "baby_SGA"              "placental_pathology"
```

24.4.2 Data dictionary dd

```
dim(dd)
```

```
[1] 27 5
```

```
names(dd)

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

24.5 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?

24.5.1 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
```

24.5.2 Unique values

To figure out which phenotypes vary within subjects, it would be helpful to answer this question:

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

A similar related question is: How would you count the number of subjects who have more than one distinct measure at each of the phenotypes?

💡 Expand to see solution

```
sapply(ds, function(x) {  
  length(unique(x))  
}) %>%  
  sort(decreasing = TRUE) %>%  
  kable()
```

	x
Gestationalage_sample	189
sample_id	119
subject_id	54
maternal_age_delivery	54
pregnancy_BMI	54
gestationalage_delivery	54
baby_birthweight_centile	52
pregnancy_weight	51
height	42
baby_birthweight	30
average_DBP_labor	27
average_SBP_labor	23
strata	21
average_SBP_lt20weeks	19
average_DBP_lt20weeks	16
gravidity	5
Sample_trimester	4
parity	4
race	3
case_control_status	2
smoke_lifetime	2
baby_sex	2
placental_pathology	2
baby_SGA	1

This can also be generated using the `map` function from the `purrr` R package.

```
ds %>%
  map(\(x) length(unique(x))) %>%
  unlist() %>%
  sort(decreasing = TRUE) %>%
  kable()
```

	x
Gestationalage_sample	189
sample_id	119
subject_id	54
maternal_age_delivery	54
pregnancy_BMI	54
gestationalage_delivery	54
baby_birthweight_centile	52
pregnancy_weight	51
height	42
baby_birthweight	30
average_DBP_labor	27
average_SBP_labor	23
strata	21
average_SBP_lt20weeks	19
average_DBP_lt20weeks	16
gravidity	5
Sample_trimester	4
parity	4
race	3
case_control_status	2
smoke_lifetime	2
baby_sex	2
placental_pathology	2
baby_SGA	1

Suppose we wanted to directly count the number of subjects who have more than one distinct measure at each of the phenotypes.

One approach for doing this would be to take the phenotype and group by `subject_id` and count distinct values within those subject-specific groups, and then add up the total number of subjects who have more than one distinct value.

```
subject.N <- function(df.col, subj.ID) {  
  # Count distinct entries when grouped by subj.ID Input: df.col = a  
  # phenotype vector subj.ID = a vector of corresponding subject IDs  
  # Construct a dataframe containing the phenotype and subject IDs  
  df <- bind_cols(df.col = df.col, subj.ID = subj.ID)  
  #  
  suppressMessages(df.n <- df %>%  
    group_by(subj.ID) %>%  
    distinct() %>%  
    mutate(n = n()) %>%  
    select(subj.ID, n) %>%  
    distinct())  
  # Count how many subj.ID's have more than one distinct value  
  sum(df.n$n > 1)  
}  
  
subj.ID <- ds$subject_id  
# Apply our function using 'map'  
ds %>%  
  map(\(x) subject.N(x, subj.ID)) %>%  
  unlist() %>%  
  sort(decreasing = TRUE)
```

Gestationalage_sample	sample_id	Sample_trimester
47	39	34
race	subject_id	strata
1	0	0
maternal_age_delivery	case_control_status	pregnancy_weight
0	0	0
height	pregnancy_BMI	gravidity
0	0	0
parity	gestationalage_delivery	average_SBP_lt20weeks
0	0	0
average_DBP_lt20weeks	average_SBP_labor	average_DBP_labor
0	0	0

smoke_lifetime	baby_birthweight	baby_sex
0	0	0
baby_birthweight_centile	baby_SGA	placental_pathology
0	0	0

The variables with a count of zero here are those where no more than 1 distinct value was observed for each subject. These are likely subject-level variables. Indeed, for the majority of these, the variable names are consistent with them being subject-level variables instead of variables that are measured every time a sample was taken.

24.5.3 Subject-level data set

Task: Construct a subject-level data set `ds.subj`

How would you construct a subject-level data set?

 Expand to see solution

We need to drop the sample-specific measures, retaining only subject-level measures, and then select unique records:

```
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
```

But there is a duplicated record where `race` differs but all other attributes are identical, so we filter one of those two records out:

```
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
# Groups:   subject_id [1]
  subject_id strata race  maternal_age_delivery case_control_status
  <chr>       <dbl> <chr>           <dbl>                  <dbl>
1 SUBJ20      35 W            29.4                 1
2 SUBJ20      35 White        29.4                 1
# i 16 more variables: prepregnancy_weight <dbl>, height <dbl>,
#   prepregnancy_BMI <dbl>, gravidity <dbl>, parity <dbl>,
#   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>

ds.subj <- ds.subj %>%
  filter(race != "White")

filter: removed one row (2%), 54 rows remaining

sum(duplicated(ds.subj$subject_id))

[1] 0

```

24.6 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?

24.6.1 Recode for understandability

Using the subject-level data set `ds.subj`, let's recode `case_control_status` from 0 and 1 into a new `PE_status` variable coded as `control` and `case`.

First, look up the coding used for the `case_control_status` variable in the Data Dictionary dd:

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

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

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

Task: Using the subject-level data set `ds.subj`, recode `case_control_status` from 0 and 1 into a new `PE_status` variable coded as `control` and `case`.

 Expand to see solution

So the data dictionary gives the meaning of the 0 and 1 codes:
“0: normotensive control; 1: preeclampsia case”

```
ds.subj$PE_status <- factor(ds.subj$case_control_status)
levels(ds.subj$PE_status)

[1] "0" "1"

levels(ds.subj$PE_status) <- c("control", "case")

# Check that the recoding was correct:
xtabs(~case_control_status + PE_status, data = ds.subj)

PE_status
case_control_status control case
      0        26     0
      1         0    28
```

Recoding could also be done using Tidyverse function:

```
ds.subj <- ds.subj %>%
  mutate(PE_status = case_when(case_control_status == 0 ~ "control", case_control_status
  1 ~ "case"))

mutate: converted 'PE_status' from factor to character (0 new NA)
```

```
xtabs(~case_control_status + PE_status, data = ds.subj)

PE_status
case_control_status case control
0      0       26
1     28       0
```

24.7 Missing data

- What is the pattern of missing data?
 - How are missing data coded?
 - Is there a single missing data code?

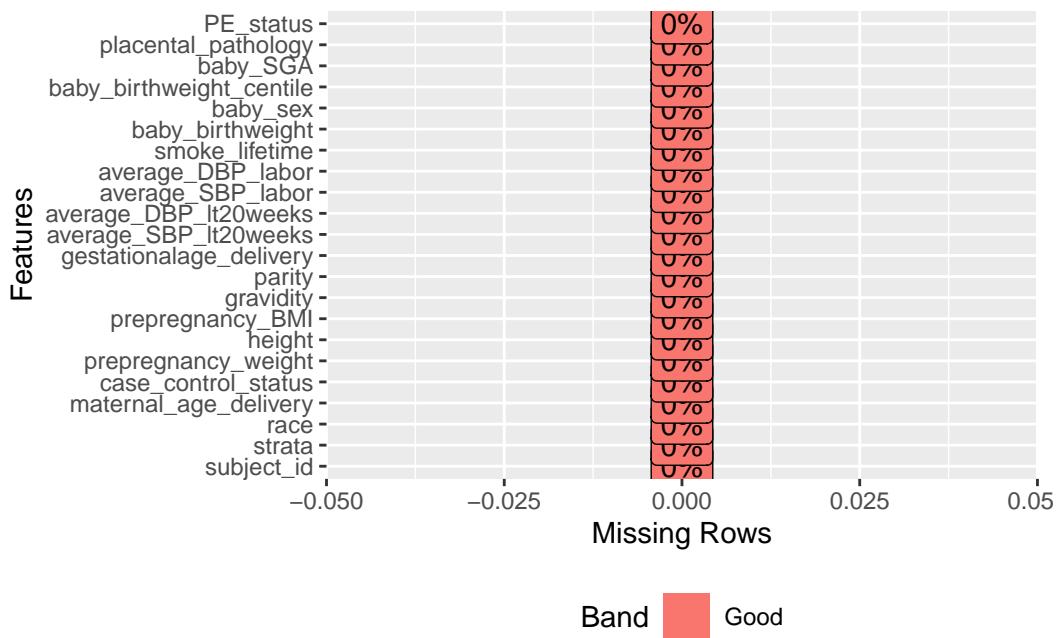
Here we could use `plot_missing` from the `DataExplorer` R package.

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

Task: Try out `plot_missing` on the subject-level data set `ds.subj`.

💡 Expand to see solution

```
plot_missing(ds.subj)
```



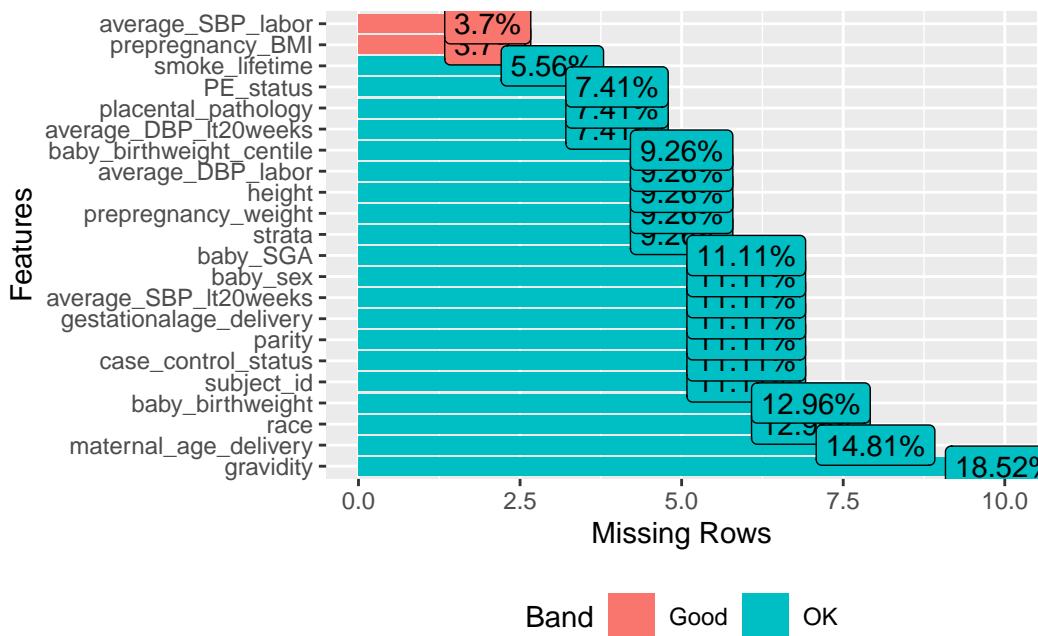
It is kind of unusual to have no missing data in a real data set.

To see what the output might look like when there is some missing data, let's introduce some using the `createNAs` function from this StackOverflow entry:

<https://stackoverflow.com/questions/39513837/add-exact-proportion-of-random-missing-values-to-data-frame>

```
createNAs <- function(x, pctNA = 0.1) {
  n <- nrow(x)
  p <- ncol(x)
  NALoc <- rep(FALSE, n * p)
  NALoc[sample.int(n * p, floor(n * p * pctNA))] <- TRUE
  x[matrix(NALoc, nrow = n, ncol = p)] <- NA
  return(x)
}

df <- ds.subj
df <- createNAs(df)
df <- data.frame(df)
plot_missing(df)
```



```
profile_missing(df)
```

	feature	num_missing	pct_missing
1	subject_id	6	0.11111111
2	strata	5	0.09259259
3	race	7	0.12962963
4	maternal_age_delivery	8	0.14814815
5	case_control_status	6	0.11111111
6	pregnancy_weight	5	0.09259259
7	height	5	0.09259259
8	pregnancy_BMI	2	0.03703704
9	gravidity	10	0.18518519
10	parity	6	0.11111111
11	gestationalage_delivery	6	0.11111111
12	average_SBP_lt20weeks	6	0.11111111
13	average_DBP_lt20weeks	4	0.07407407
14	average_SBP_labor	2	0.03703704
15	average_DBP_labor	5	0.09259259
16	smoke_lifetime	3	0.05555556
17	baby_birthweight	7	0.12962963
18	baby_sex	6	0.11111111
19	baby_birthweight_centile	5	0.09259259

```
20          baby_SGA           6  0.11111111
21  placental_pathology      4  0.07407407
22          PE_status          4  0.07407407
```

When there is some missing data, in addition to applying `plot_missing` and `profile_missing`, you could also apply functions from the ‘VIM’ R package, which has a number of commands that are useful for visualizing missing data patterns.

<https://cran.r-project.org/web/packages/VIM/vignettes/VIM.html>

24.8 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?

Potentially useful `DataExplorer` commands to use in this context include:

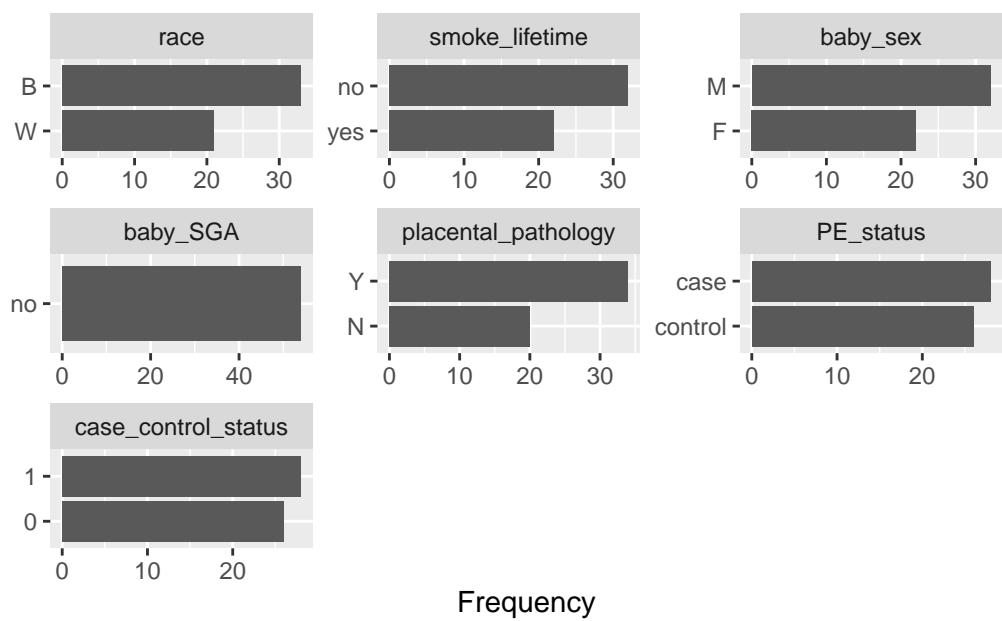
```
plot_bar    Plot bar chart
plot_density  Plot density estimates
plot_histogram Plot histogram
plot_qq Plot QQ plot
```

Task: Try out these commands.

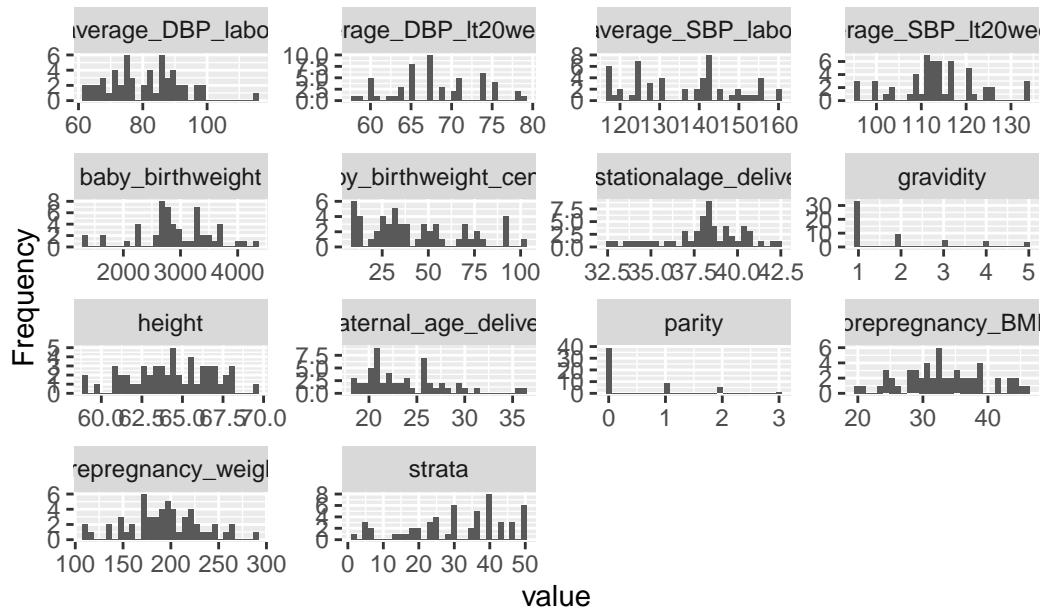
💡 Expand to see solution

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

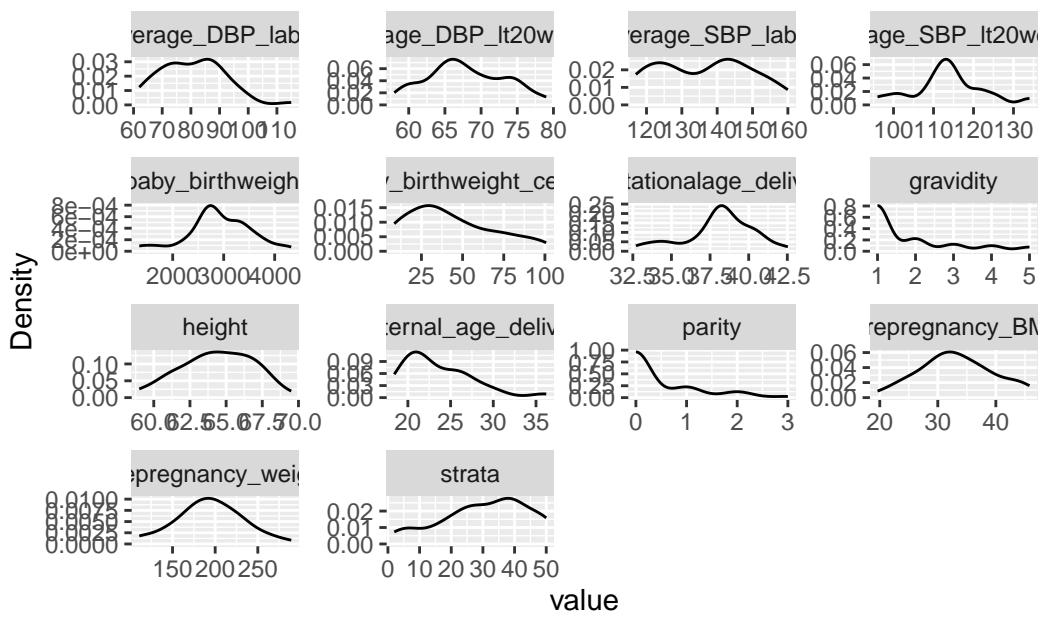
```
1 columns ignored with more than 50 categories.
subject_id: 54 categories
```



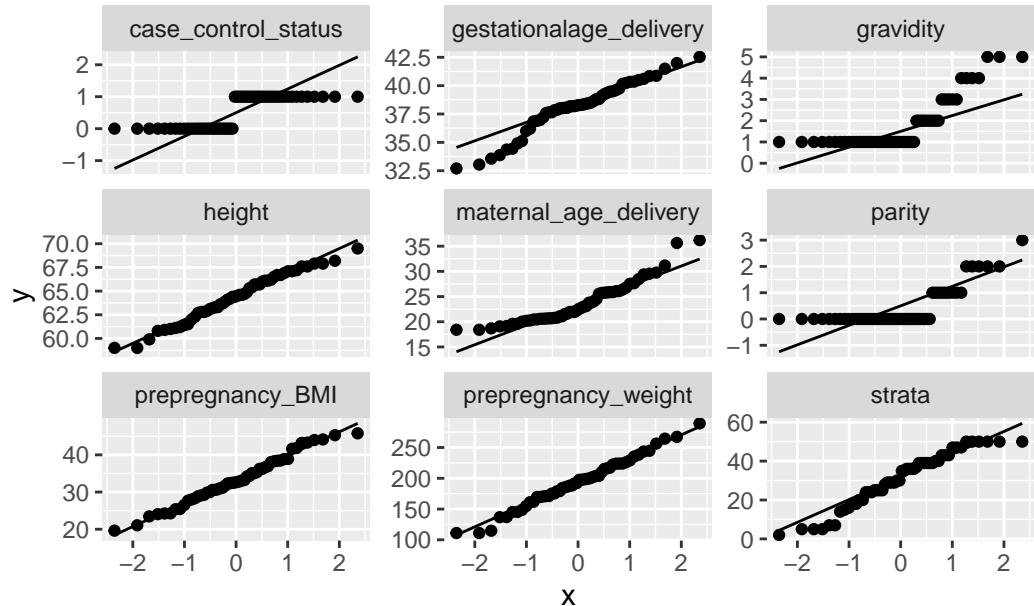
```
plot_histogram(ds.subj)
```

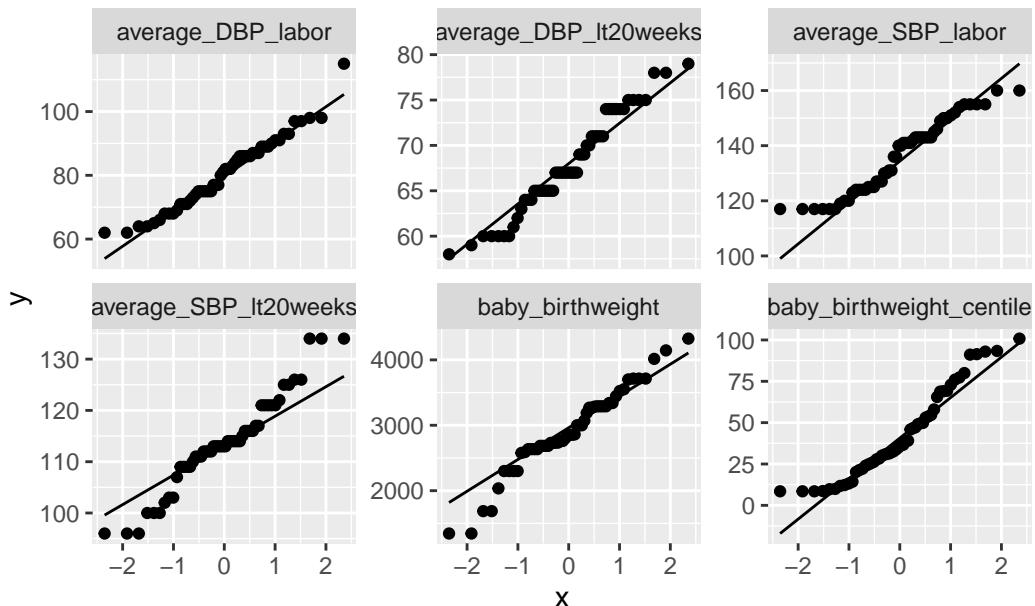


```
plot_density(ds.subj)
```



```
plot_qq(ds.subj)
```





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24.9 Variation

- How do our data vary and co-vary?
 - Do multiple measures agree with each other?
 - Are there sex-specific or age-specific differences?

Task: As it is of interest to examine how our traits vary by pre-eclampsia case/control status, we can explore this by using the `by = "PE_status"` argument within the `DataExplorer` commands to break down the plots drawn in the previous section by `PE_status`.

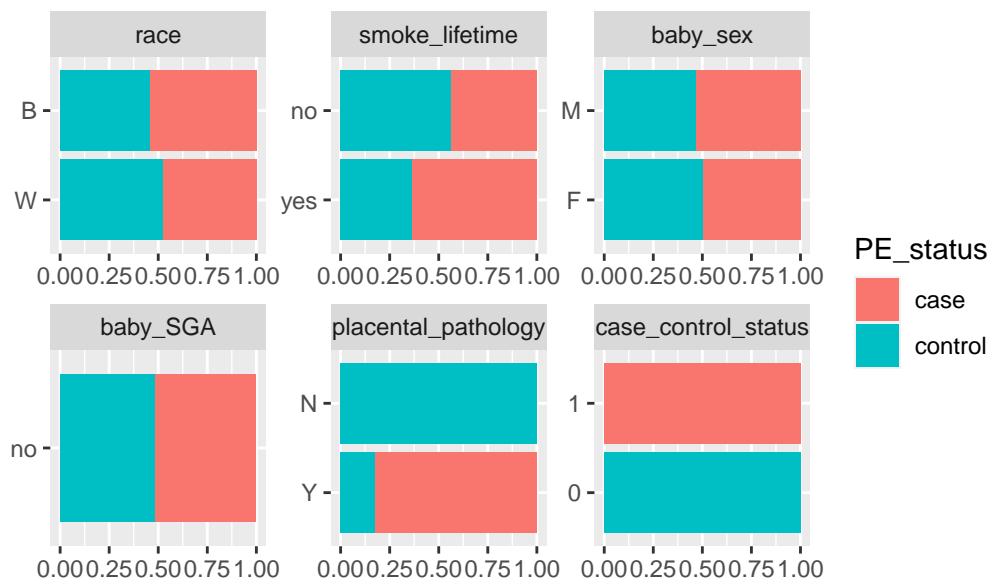
Also try creating boxplots using the `plot_boxplot` command.

Expand to see solution

24.9.1 Bar plots

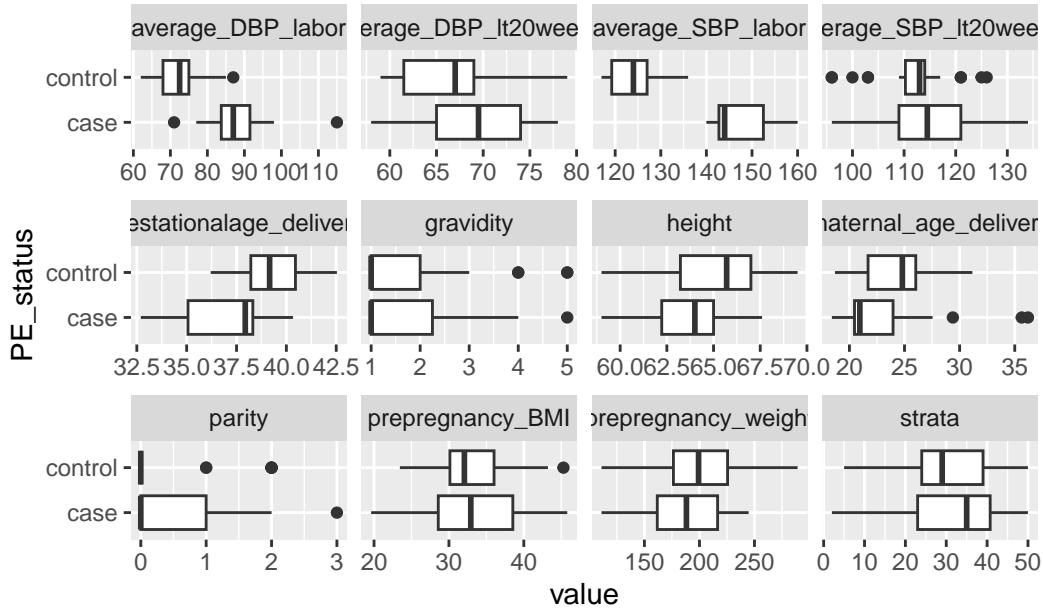
```
plot_bar(ds.subj, by = "PE_status")
```

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

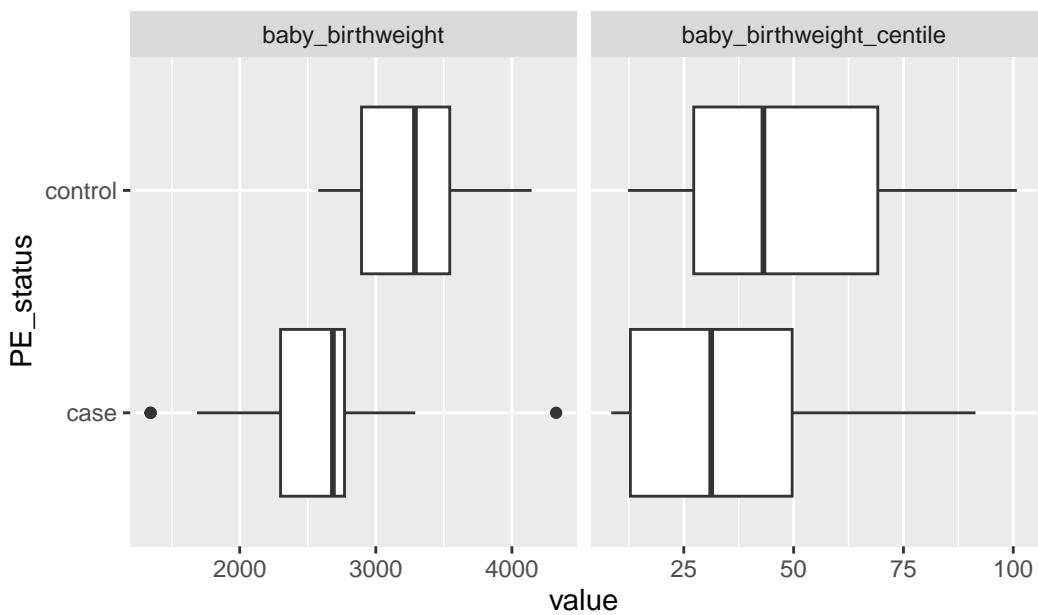


24.9.2 Box plots

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



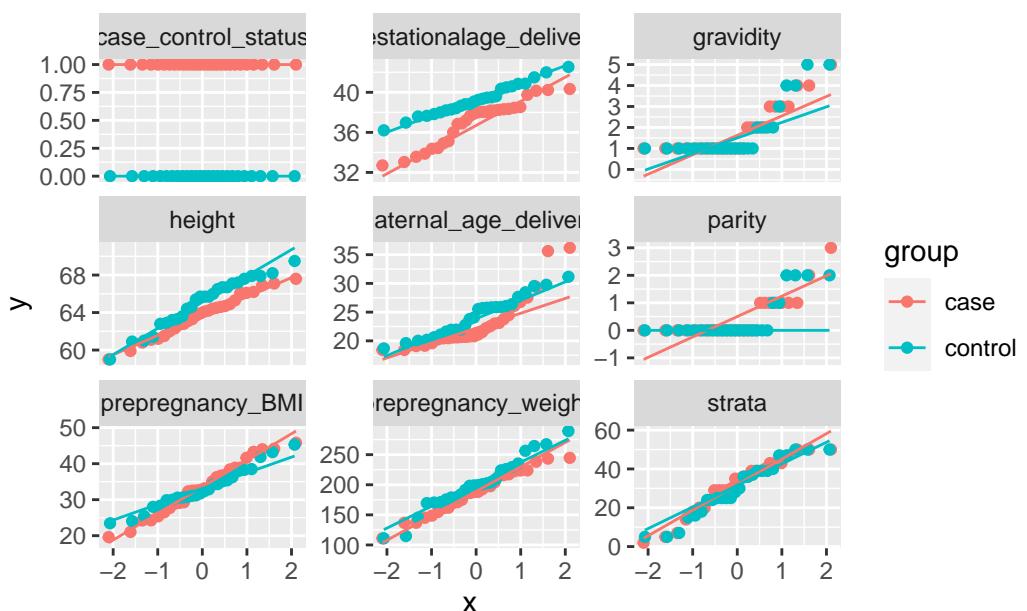
Page 1



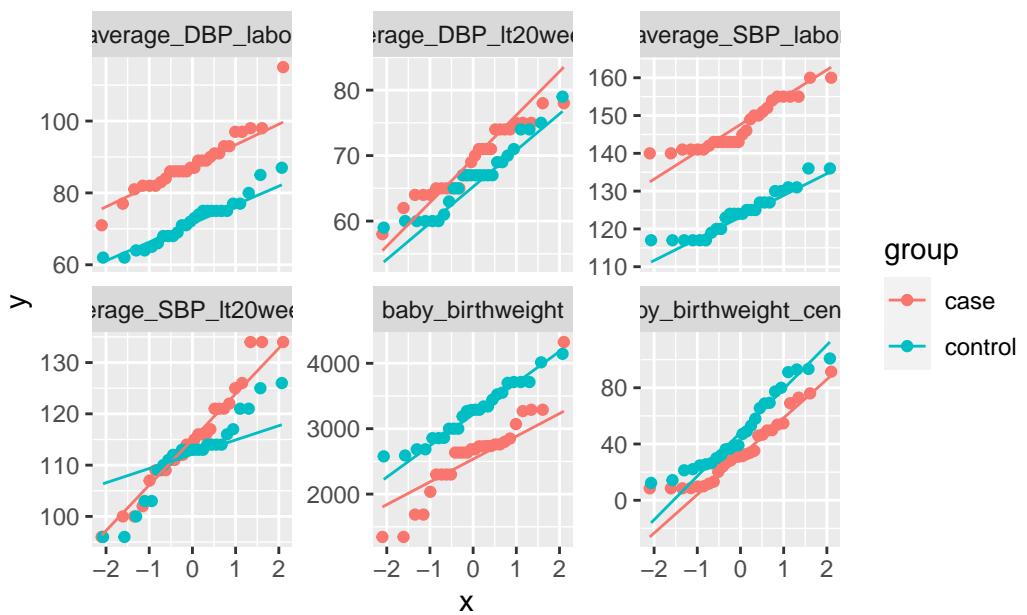
Page 2

24.9.3 QQ plots

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



Page 1



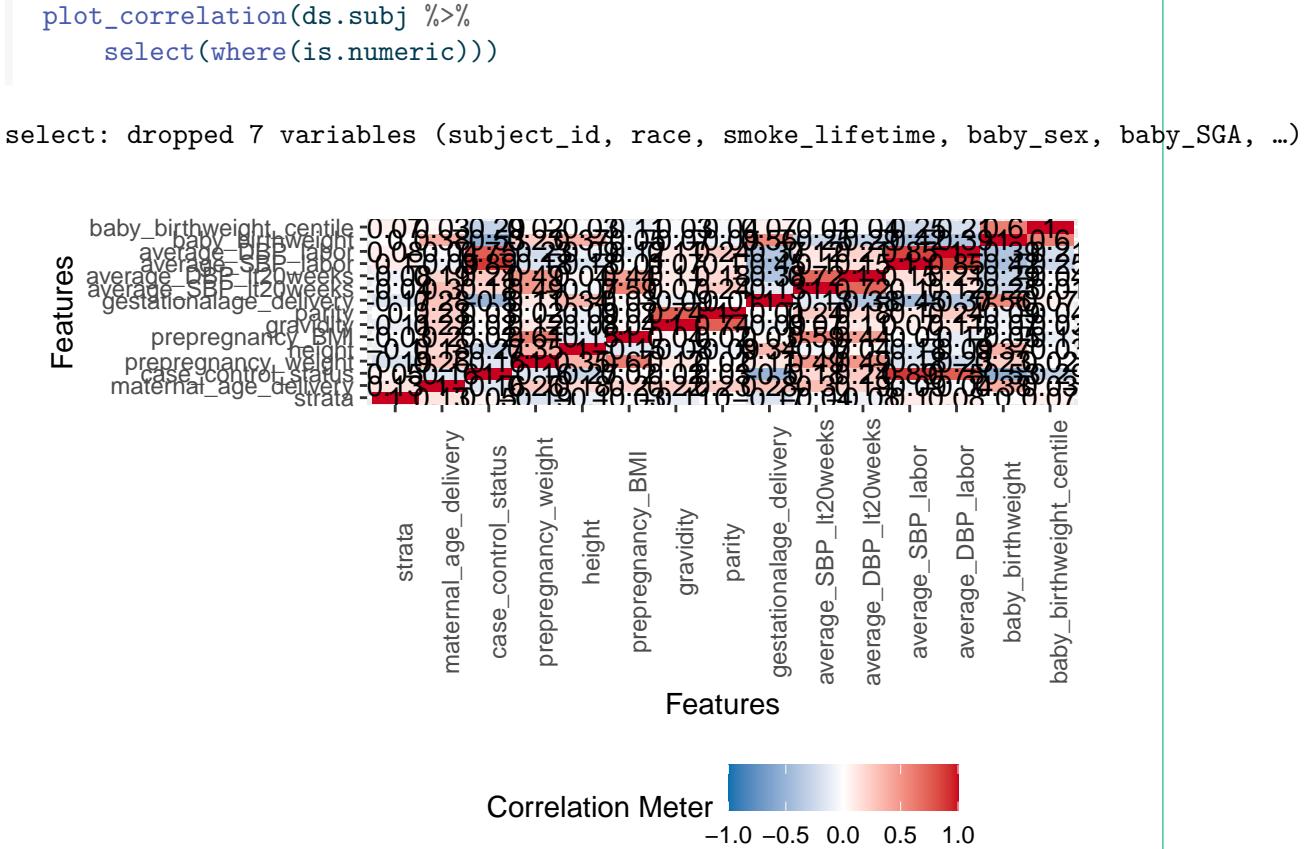
Page 2

24.9.4 Correlation

For plotting correlation matrices, `DataExplorer` provides the `plot_correlation` command.

Task: Try `plot_correlation` out, on the subset of numeric columns.

💡 Expand to see solution



24.9.5 ggpairs

Use `ggpairs` from the `GGally` R package.

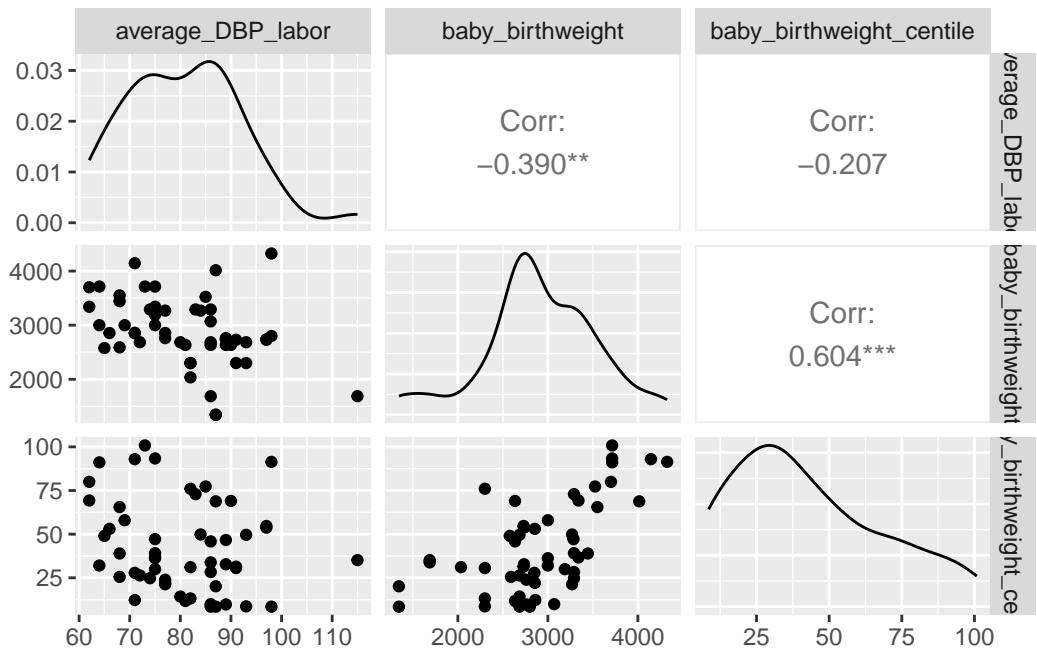
```
# To illustrate, let's just use three of the numeric traits:
ds1 <- ds.subj[, c(15, 17, 19)]
names(ds1)
```

```
[1] "average_DBP_labor"           "baby_birthweight"  
[3] "baby_birthweight_centile"
```

Task: Try it out - apply `ggpairs` to `ds1`.

💡 Expand to see solution

```
ggpairs(ds1)
```



Task: Redraw the `ggpairs` plot, using the `mapping` argument to color by `PE_status`.

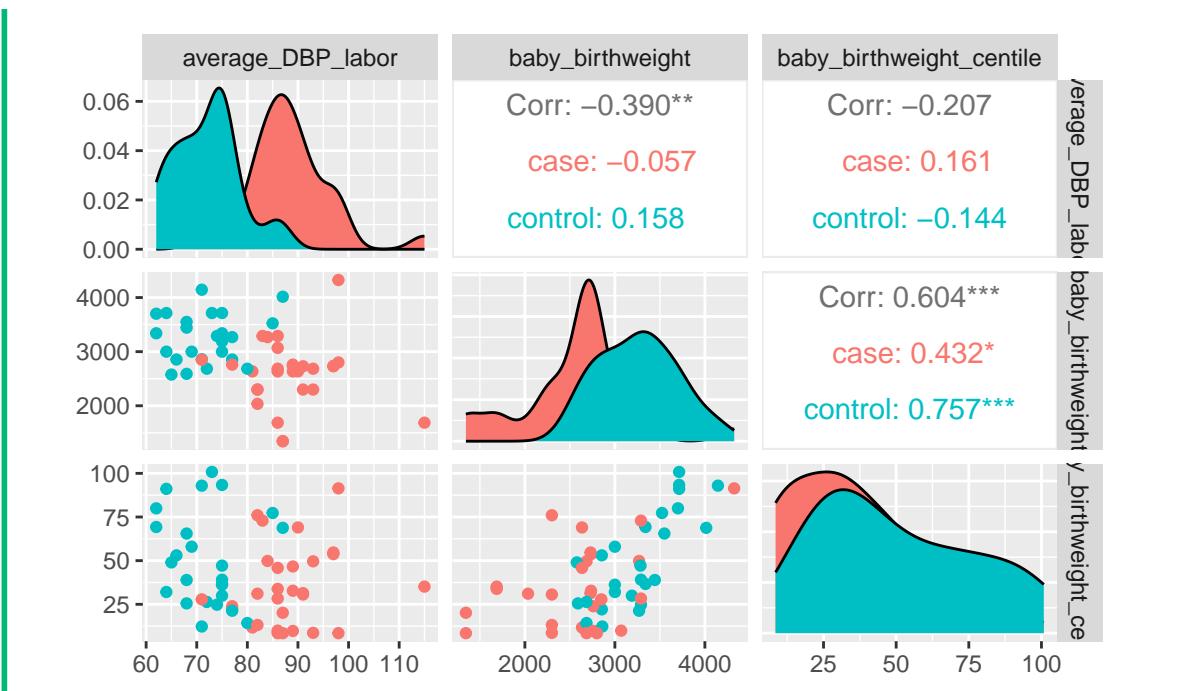
To figure out how do this, look at the examples in the `?ggpairs` function documentation.

💡 Hint

This cannot be done using the `ds1` object because that does not contain any `PE_status` information.

💡 Expand to see solution

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

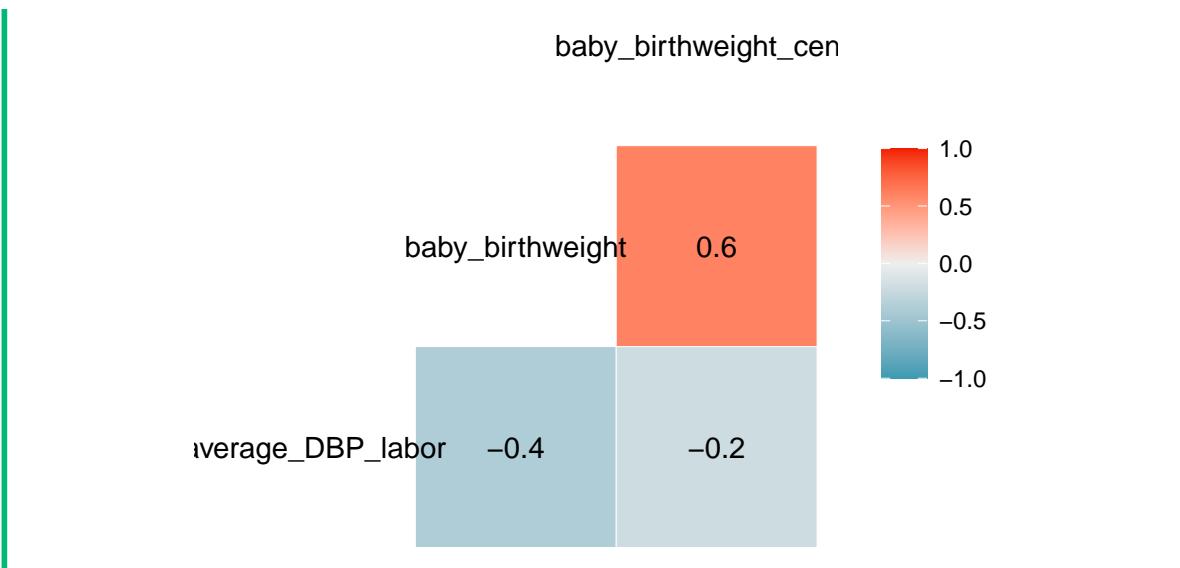


24.9.6 ggcorm

Note

The `ggcorr` function from the `GGally` R package can also be used to make a correlation matrix plot.

```
ggcorr(ds1, label = TRUE)
```



24.10 DataExplorer

We can quickly create a report using the `create_report` function from the `DataExplorer` R package

```
create_report(ds.subj)
```

See

<https://boxuancui.github.io/DataExplorer/>

24.11 dataMaid

The `dataMaid` R package can also be used to create an exploratory data analysis report.

```
library(dataMaid)
makeDataReport(ds.subj, output="html")
```

See

<https://www.jstatsoft.org/article/view/v090i06>

24.12 SmartEDA

The **SmartEDA** R package also has a command to create an exploratory data analysis report - this command is **ExpReport**.

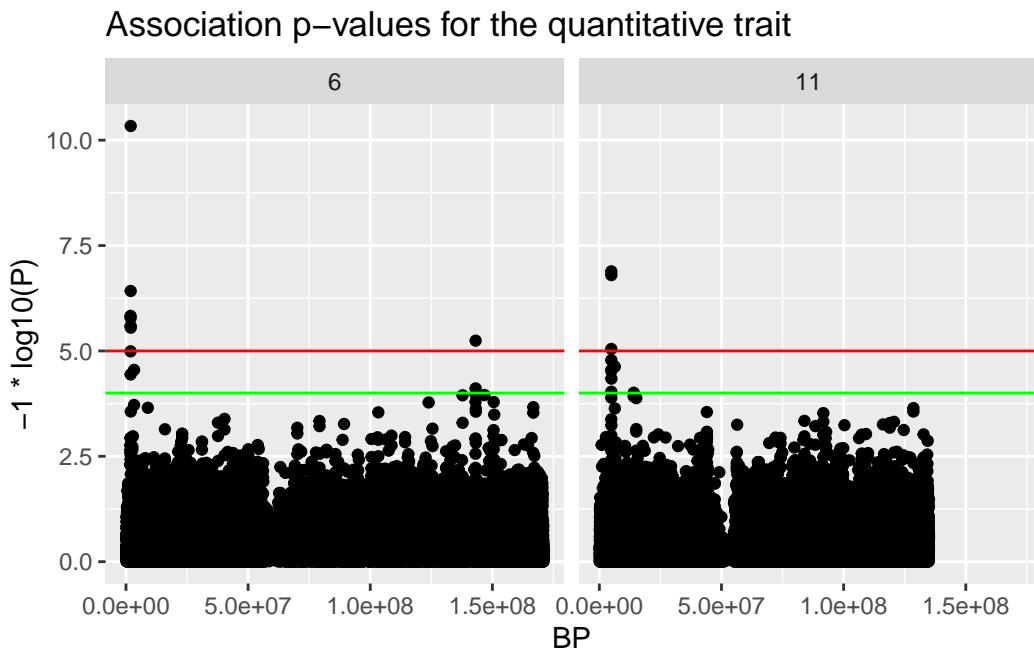
```
library(SmartEDA)
ExpReport(ds.subj, op_file="SmartEDAResult.html")
ExpReport(ds.subj, Target="PE_status", Rc="control", op_file="SmartEDAResultII.html")
```

For more information, see <https://cran.r-project.org/web/packages/SmartEDA/vignettes/SmartEDA.html>

25 GRanges Exercise

In an example genome-wide association scan, we had several hits:

```
library(tidyverse)
library(knitr)
library(pander)
ASSOC.file <- "data/trait.qassoc.gz"
b <- read_table(ASSOC.file)
qplot(BP,-1.0*log10(P),main="Association p-values for the quantitative trait",data=b) +
  geom_hline(yintercept=5,col='red') +
  geom_hline(yintercept=4,col='green')
```



Here are our top hits:

```

top.hits <- b[order(b$P),c("CHR","BP","SNP","P")]
row.names(top.hits) <- NULL
pander(head(top.hits),caption="Top hits from our genome-wide association scan")

```

Table 25.1: Top hits from our genome-wide association scan

CHR	BP	SNP	P
6	1942538	rs3800143	4.608e-11
11	4913057	rs10836914	1.295e-07
11	4913314	rs12577475	1.587e-07
6	1926650	rs11242725	3.785e-07
6	1880964	rs3800116	1.475e-06
6	1915129	rs3778552	1.646e-06

Now we'd like to annotate each of these SNPs in terms of nearby or overlapping genes.

While we could do this using online databases like UCSC Genome Brower (<http://genome.ucsc.edu/cgi-bin/hgGateway>), we could also use BioConductor R packages to write our own functions to retrieve this information into a nice tabular format, for each SNP, listing any gene that they might be in, including the SNP's position and the gene boundaries.

25.1 Introductory Background

An Introduction to Bioconductor's Packages for Working with Range Data

<https://github.com/vsbuffalo/genomicranges-intro/blob/master/notes.md>

25.2 Active Learning

Working with genomics ranges

<https://carpentries-incubator.github.io/bioc-project/07-genomic-ranges.html>

25.3 Suggested readings

In “Bioinformatics Data Skills”, see Chapter 9 “Working with Range Data”

Bioinformatics Data Skills

Editor: Vince Buffalo

Publisher: O'Reilly
Web access: [link](#)

Hello Ranges: An Introduction to Analyzing Genomic Ranges in R.
[link](#)

25.4 Install the needed Bioconductor libraries (one time only)

First, we need to figure out which genomic build was used in our data set. I usually do this by looking up the base pair positions of a couple of SNPs by hand in the 'SNP' data base. So if we go there and search for `rs3800143`, we end up on this web page:

https://www.ncbi.nlm.nih.gov/snp/rs3800143#variant_details

which shows a Build GRCh37.p13 position of 1942538 on chromosome 6. So it looks like Build GRCh37.p13 was used, which is also known as Build hg19.

To double check, we can check that the given position for rs10836914 matches the Build GRCh37.p13 position.

So to determine the gene boundaries in Build hg19, we need to download and install the `TxDb.Hsapiens.UCSC.hg19.knownGene` library from BioConductor.

So we search for it on BioConductor and that it can be installed by issuing these commands at the R prompt:

```
if (!requireNamespace("BiocManager", quietly = TRUE))
  install.packages("BiocManager")

BiocManager::install("TxDb.Hsapiens.UCSC.hg19.knownGene")
```

You will also need to install another library, `org.Hs.eg.db`:

```
if (!requireNamespace("BiocManager", quietly = TRUE))
  install.packages("BiocManager")

BiocManager::install("org.Hs.eg.db")
```

25.5 Construct the gene list

Using the annotation embedded in `TxDb.Hsapiens.UCSC.hg19.knownGene`, construct a `GRangesList` object that contains a list of all the genes.

Hint: Use `transcriptsBy`.

```
library(TxDb.Hsapiens.UCSC.hg19.knownGene)
txdb <- TxDb.Hsapiens.UCSC.hg19.knownGene
class(txdb)

[1] "TxDb"
attr(,"package")
[1] "GenomicFeatures"
```

The `transcripts(txdb)` is a `GenomicRanges` object:

```
transcripts(txdb)

GRanges object with 82960 ranges and 2 metadata columns:
  seqnames      ranges strand |      tx_id      tx_name
      <Rle>      <IRanges>  <Rle> | <integer> <character>
 [1]     chr1  11874-14409    + |      1 uc001aaa.3
 [2]     chr1  11874-14409    + |      2 uc010nxq.1
 [3]     chr1  11874-14409    + |      3 uc010nxr.1
 [4]     chr1  69091-70008    + |      4 uc001aal.1
 [5]     chr1 321084-321115    + |      5 uc001aaq.2
 ...
 [82956]   ...       ...    ... |      ...
 [82957] chrUn_g1000237    1-2686   - |  82956 uc011mgu.1
 [82958] chrUn_g1000241 20433-36875   - |  82957 uc011mgv.2
 [82959] chrUn_g1000243 11501-11530   + |  82958 uc011mgw.1
 [82960] chrUn_g1000247 13608-13637   + |  82959 uc022brq.1
 -----
 seqinfo: 93 sequences (1 circular) from hg19 genome
```

We now group the transcripts by gene, and so create a `GRangesList` object:

```
tx.by.gene <- transcriptsBy(txdb, "gene")
tx.by.gene
```

```

GRangesList object of length 23459:
$`1`
GRanges object with 2 ranges and 2 metadata columns:
  seqnames      ranges strand |   tx_id    tx_name
  <Rle>        <IRanges>  <Rle> | <integer> <character>
 [1] chr19 58858172-58864865     - |     70455 uc002qsd.4
 [2] chr19 58859832-58874214     - |     70456 uc002qsf.2
-----
seqinfo: 93 sequences (1 circular) from hg19 genome

$`10`
GRanges object with 1 range and 2 metadata columns:
  seqnames      ranges strand |   tx_id    tx_name
  <Rle>        <IRanges>  <Rle> | <integer> <character>
 [1] chr8 18248755-18258723     + |     31944 uc003wyw.1
-----
seqinfo: 93 sequences (1 circular) from hg19 genome

$`100`
GRanges object with 1 range and 2 metadata columns:
  seqnames      ranges strand |   tx_id    tx_name
  <Rle>        <IRanges>  <Rle> | <integer> <character>
 [1] chr20 43248163-43280376     - |     72132 uc002xmj.3
-----
seqinfo: 93 sequences (1 circular) from hg19 genome

...
<23456 more elements>
```

The names of the list elements are Entrez gene IDs.

25.6 Construct a GRange containing our top SNP

Construct a GRange containing the top SNP. Note that the chromosome name needs to be in the same style as is seen in `tx.by.gene`. That is, the chromosome name needs to be “chr6” instead of ‘6’.

```
top.snp <- with(top.hits[1,], GRanges(seqnames=paste0("chr",CHR),
                                         IRanges(start=BP, width=1),
                                         rsid=SNP, P=P))
```

```

top.snp

GRanges object with 1 range and 2 metadata columns:
  seqnames      ranges strand |      rsid      P
  <Rle> <IRanges> <Rle> | <character> <numeric>
[1]   chr6    1942538     * |    rs3800143 4.608e-11
-----
seqinfo: 1 sequence from an unspecified genome; no seqlengths

```

25.7 Search for match

Now use `findOverlaps` and `subsetByOverlaps` to find any genes that overlap our top SNP.

```

top.snp.overlaps <- findOverlaps(tx.by.gene,top.snp)
top.snp.overlaps

Hits object with 1 hit and 0 metadata columns:
  queryHits subjectHits
  <integer> <integer>
[1]       8841           1
-----
queryLength: 23459 / subjectLength: 1

hits <- subsetByOverlaps(tx.by.gene,top.snp)
hits

GRangesList object of length 1:
$`2762`
GRanges object with 2 ranges and 2 metadata columns:
  seqnames      ranges strand |      tx_id      tx_name
  <Rle> <IRanges> <Rle> | <integer> <character>
[1]   chr6 1624035-2176225     - |    25755 uc021ykn.1
[2]   chr6 1624035-2245868     - |    25756 uc003mtq.3
-----
seqinfo: 93 sequences (1 circular) from hg19 genome

```

So our top SNP rs3800143 overlaps two transcripts in a single gene with the same start position of 1624035.

However, the transcript name is not very human-readable, as it uses an Entrez Gene ID 2762.

25.8 Convert Entrez Gene IDs to Gene Names

Convert the integer Entrez Gene ID 2762 to a human-readable Gene Name.

This can be done using the `org.Hs.eg.db` R library.

To convert Entrez Gene IDs to Gene Names, we can use another R database:

```
library(org.Hs.eg.db)
columns(org.Hs.eg.db)

[1] "ACCNUM"      "ALIAS"        "ENSEMBL"       "ENSEMLPROT"   "ENSEMLTRANS"
[6] "ENTREZID"     "ENZYME"       "EVIDENCE"      "EVIDENCEALL"  "GENENAME"
[11] "GENETYPE"     "GO"           "GOALL"         "IPI"          "MAP"
[16] "OMIM"         "ONTOLOGY"     "ONTOLOGYALL"  "PATH"         "PFAM"
[21] "PMID"         "PROSITE"      "REFSEQ"        "SYMBOL"       "UCSCKG"
[26] "UNIPROT"

names(hits)

[1] "2762"

gene.name <- select(org.Hs.eg.db, keys=names(hits), columns=c("ENTREZID", "SYMBOL", "GENENAME"))

'select()' returned 1:1 mapping between keys and columns

gene.name

  ENTREZID SYMBOL          GENENAME
1    2762   GMDS GDP-mannose 4,6-dehydratase
```

So our top SNP rs3800143 overlaps the gene with the Entrez Gene ID 2762, which is also known as GMDS (GDP-mannose 4,6-dehydratase).

25.9 Question 1

Read in the data:

```

ASSOC.file <- "data/trait.qassoc.gz"
b <- read_table(ASSOC.file)

Warning: Missing column names filled in: 'X10' [10]

-- Column specification -----
cols(
  CHR = col_double(),
  SNP = col_character(),
  BP = col_double(),
  NMISS = col_double(),
  BETA = col_double(),
  SE = col_double(),
  R2 = col_double(),
  T = col_double(),
  P = col_double(),
  X10 = col_logical()
)
head(b)

# A tibble: 6 x 10
  CHR SNP      BP NMISS     BETA     SE      R2     T     P X10
  <dbl> <chr>   <dbl> <dbl>    <dbl>    <dbl>    <dbl>    <dbl> <dbl> <lgl>
1     6 rs9392298 203878  1573  0.110  0.0984  0.000801  1.12  0.262 NA
2     6 rs9405486 204072  1569  0.118  0.0982  0.000913  1.20  0.232 NA
3     6 rs7762550 204909  1574  0.0384  0.0685  0.000199  0.560 0.576 NA
4     6 rs1418706 205878  1575 -0.00783 0.0717  0.00000757 -0.109 0.913 NA
5     6 rs6920539 206528  1574  0.119  0.0955  0.000984  1.24  0.214 NA
6     6 rs9502959 206599  1575 -0.0317  0.0538  0.000220  -0.589 0.556 NA

```

Task: Using GRanges and associated Bioconductor tools, figure out if our top SNP, as ranked by the P-value (column P) from data frame b lies within an intron or exon.

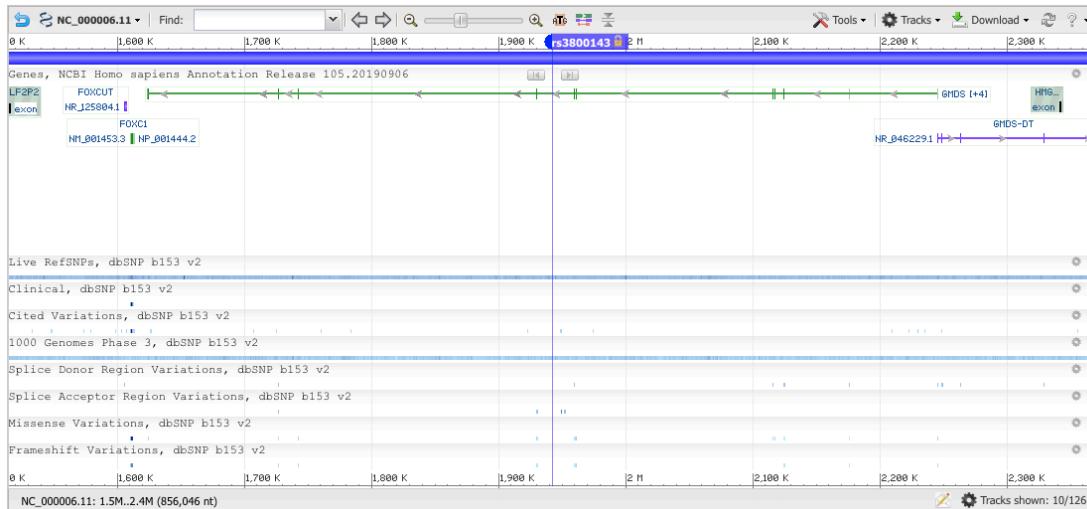
For this, the functions `exonsBy` and `intronsByTranscript` might be useful.

 Expand to see solution

25.10 Answer 1

The top SNP rs3800143 lies in an intron of GMDS (GDP-mannose 4,6-dehydratase), Gene ID: 2762.

Build hg19 (Human Feb. 2009 (GRCh37/hg19)) gene boundaries from UCSC Genes: GMDS (uc003mtq.3) - chr6:1624035-2245868 - Homo sapiens GDP-mannose 4,6-dehydratase (GMDS), transcript variant 1, mRNA.



Does our top SNP overlap any exons?

```
top.snp <- with(top.hits[1,], GRanges(seqnames=paste0("chr",CHR),
                                         IRanges(start=BP, width=1),
                                         rsid=SNP, P=P))
top.snp
```

```
GRanges object with 1 range and 2 metadata columns:
  seqnames      ranges strand |      rsid      P
  <Rle> <IRanges> <Rle> | <character> <numeric>
 [1]   chr6    1942538     * |    rs3800143 4.608e-11
 -----
 seqinfo: 1 sequence from an unspecified genome; no seqlengths
```

```
exons.by.gene <- exonsBy(txdb, "gene")
exons.by.gene
```

```

GRangesList object of length 23459:
$`1`  

GRanges object with 15 ranges and 2 metadata columns:  

  seqnames      ranges strand | exon_id exon_name  

    <Rle>      <IRanges>  <Rle> | <integer> <character>  

[1] chr19 58858172-58858395 - | 250809 <NA>  

[2] chr19 58858719-58859006 - | 250810 <NA>  

[3] chr19 58859832-58860494 - | 250811 <NA>  

[4] chr19 58860934-58862017 - | 250812 <NA>  

[5] chr19 58861736-58862017 - | 250813 <NA>  

...   ...   ...   ... .   ...   ...  

[11] chr19 58868951-58869015 - | 250821 <NA>  

[12] chr19 58869318-58869652 - | 250822 <NA>  

[13] chr19 58869855-58869951 - | 250823 <NA>  

[14] chr19 58870563-58870689 - | 250824 <NA>  

[15] chr19 58874043-58874214 - | 250825 <NA>  

-----  

seqinfo: 93 sequences (1 circular) from hg19 genome  

$`10`  

GRanges object with 2 ranges and 2 metadata columns:  

  seqnames      ranges strand | exon_id exon_name  

    <Rle>      <IRanges>  <Rle> | <integer> <character>  

[1] chr8 18248755-18248855 + | 113603 <NA>  

[2] chr8 18257508-18258723 + | 113604 <NA>  

-----  

seqinfo: 93 sequences (1 circular) from hg19 genome  

...
<23457 more elements>  

  top.snp.overlaps <- findOverlaps(exons.by.gene,top.snp)
  top.snp.overlaps

Hits object with 0 hits and 0 metadata columns:
  queryHits subjectHits
    <integer>  <integer>
-----  

  queryLength: 23459 / subjectLength: 1

```

```
hits <- subsetByOverlaps(exons.by.gene,top.snp)
hits
```

```
GRangesList object of length 0:
<0 elements>
```

From the output above, we find that our top SNP overlaps 0 sets of exons grouped by transcripts.

Does our top SNP overlap any introns?

```
introns.by.transcript <- intronsByTranscript(txdb)
```

```
top.snp.overlaps <- findOverlaps(introns.by.transcript,top.snp)
top.snp.overlaps
```

```
Hits object with 2 hits and 0 metadata columns:
```

```
  queryHits subjectHits
  <integer>   <integer>
[1]      25755          1
[2]      25756          1
-----
```

```
queryLength: 82960 / subjectLength: 1
```

```
hits <- subsetByOverlaps(introns.by.transcript,top.snp)
hits
```

```
GRangesList object of length 2:
```

```
$`25755`
```

```
GRanges object with 10 ranges and 0 metadata columns:
```

	seqnames	ranges	strand
	<Rle>	<IRanges>	<Rle>
[1]	chr6	1624467-1624706	-
[2]	chr6	1624776-1726649	-
[3]	chr6	1726747-1742701	-
[4]	chr6	1742821-1930336	-
[5]	chr6	1930465-1960100	-
[6]	chr6	1960206-1961007	-
[7]	chr6	1961201-2116004	-
[8]	chr6	2116115-2117702	-

```

[9] chr6 2117791-2124920      -
[10] chr6 2124966-2176165      -
-----
seqinfo: 93 sequences (1 circular) from hg19 genome

$`25756`  

GRanges object with 10 ranges and 0 metadata columns:  

  seqnames      ranges strand
    <Rle>      <IRanges> <Rle>
[1] chr6 1624467-1624706      -
[2] chr6 1624776-1726649      -
[3] chr6 1726747-1742701      -
[4] chr6 1742821-1930336      -
[5] chr6 1930465-1960100      -
[6] chr6 1960206-1961007      -
[7] chr6 1961201-2116004      -
[8] chr6 2116115-2117702      -
[9] chr6 2117791-2124920      -
[10] chr6 2124966-2245554     -
-----
seqinfo: 93 sequences (1 circular) from hg19 genome

```

From the output above, we find that our top SNP overlaps 2 sets of introns grouped by transcripts.

25.11 Question 2

While we have figure out above which gene our top SNP is in, what we worked out above involves very specific code. Now let's try to generalize what we did above:

Task: Using GRanges and associated Bioconductor tools, write a function that takes as input a ranked list of the SNPs, and returns a nice table that lists the top N of these SNPs and any gene that they might be in, including the SNP position and the gene boundaries.

```

# snp.list = the ranked list of the top SNPs
# N = the number of top SNPs to annotate
snp.table <- function(snp.list, N=15) {

}

```

Apply your `snp.table` function to the top 15 SNPs in our example data set.

 Hint

Use the `subsetByOverlaps` followed by the `select(org.Hs.eg.db, ...)` approach described above in the **Search for match** and **Convert Entrez Gene IDs to Gene Names** sections above.

 Expand to see solution

25.12 Answer 2

It is important to understand what the output of the `findOverlaps` function means.

```
top.snp.gene <- findOverlaps(gene.bounds, top.snp)
top.snp.gene
```

returns:

```
Hits object with 10 hits and 0 metadata columns:
  queryHits subjectHits
  <integer>   <integer>
[1]      3269         13
[2]      8841          5
[3]      8841         11
[4]      8841          7
[5]      8841          6
[6]      8841          4
[7]      8841          1
[8]      8841          8
[9]     17049         14
[10]     18258         9
-----
queryLength: 23459 / subjectLength: 15
```

Here, the ‘subjectHits’ is an index into `top.snp` and the `queryHits` is an index into `gene.bounds`.

So the first line indicates that the 13th SNP in `top.snp` overlaps the gene that is in the 3,269 slot of the `gene.bounds` object.

```

# snp.list = the ranked list of the top SNPs
# N = the number of top SNPs to annotate
snp.table <- function(snp.list, N=15) {
  require(org.Hs.eg.db)
  require(TxDb.Hsapiens.UCSC.hg19.knownGene)
  txdb <- TxDb.Hsapiens.UCSC.hg19.knownGene
  tx.by.gene <- transcriptsBy(txdb, "gene")
  # Find the gene boundaries
  gene.bounds <- reduce(tx.by.gene)
  # Set up a GRange with the first N top SNPs
  top.snp <- with(snp.list[1:N, ], GRanges(seqnames = paste0("chr", CHR),
                                             IRanges(start = BP, width = 1,
                                             SNP = SNP, P = P))
  # Find overlaps and hits
  top.snp.gene <- findOverlaps(gene.bounds, top.snp)
  hits <- subsetByOverlaps(gene.bounds, top.snp)
  # The SNP hits
  snp.info <- data.frame(SNP.ID = subjectHits(top.snp.gene),
                         SNP.chr = seqnames(top.snp[subjectHits(top.snp.gene)]),
                         ranges(top.snp[subjectHits(top.snp.gene)]),
                         mcols(top.snp[subjectHits(top.snp.gene)]))
  # The Gene hits
  gene.info <- data.frame(gene.ID = queryHits(top.snp.gene),
                         seqnames(gene.bounds[queryHits(top.snp.gene)]),
                         ranges(gene.bounds[queryHits(top.snp.gene)]))
  # Reduce gene.info to distinct entries
  gene.info <- gene.info %>% dplyr::select(-group, -group.1) %>% distinct()
  # Construct a key linking SNPs to Genes
  key <- data.frame(gene.ID = queryHits(top.snp.gene),
                     SNP.ID = subjectHits(top.snp.gene))
  snp.gene <- key %>% left_join(gene.info, by = "gene.ID") %>%
    left_join(snp.info, by = "SNP.ID")
  gene.name <- select(org.Hs.eg.db, keys = names(hits),
                      columns = c("ENTREZID", "SYMBOL", "GENENAME"), keytype = "ENTREZID")
  snp.gene <- snp.gene %>% dplyr::rename(ENTREZID = group_name) %>%
    left_join(gene.name, by = "ENTREZID")
  snp.gene <- snp.gene %>% arrange(P)
  snp.gene <- left_join(snp.list[1:N, ], snp.gene, by = c("SNP"))
  snp.gene <- snp.gene %>%
    dplyr::select(CHR, BP, SNP, P.x, SYMBOL, GENENAME, start.x, end.x, width.x)
  snp.gene <- snp.gene %>%
    dplyr::rename(P = P.x, start = start.x, end = end.x, width = width.x)
  return(snp.gene)
}

```

```
(t <- snp.table(top.hits, 15)) %>% kable(digits=15)
```

```
Warning in .local(x, row.names, optional, ...): 'optional' argument was ignored  
'select()' returned 1:1 mapping between keys and columns
```

CHR	BP	SNP	P	SYMBOL	GENENAME	start	end	width
6	1942538	rs38001434.608e-11	GMDS	GDP-mannose		1624035	2245868	621834
11	4913057	rs10836914.295e-07	NA	NA		NA	NA	NA
11	4913314	rs12577475.587e-07	NA	NA		NA	NA	NA
6	1926650	rs11242723.785e-07	GMDS	GDP-mannose		1624035	2245868	621834
6	1880964	rs38001161.475e-06	GMDS	GDP-mannose		1624035	2245868	621834
6	1915129	rs37785521.646e-06	GMDS	GDP-mannose		1624035	2245868	621834
6	1908210	rs28757112.550e-06	GMDS	GDP-mannose		1624035	2245868	621834
6	1955398	rs93786642.809e-06	GMDS	GDP-mannose		1624035	2245868	621834
6	143144885926372	5.704e-06	HIVEP2	HIVEP zinc finger 2		1430726044326633893735		
11	4912192	rs10836918.977e-06	NA	NA		NA	NA	NA
6	1899671	rs38001221.022e-05	GMDS	GDP-mannose		1624035	2245868	621834
11	4910224	rs25705911.656e-05	NA	NA		NA	NA	NA
11	6340706	rs10519922.356e-05	CAVIN3	caveolae associated protein 3		6340176	6341740	1565
6	3317016	rs49598042.821e-05	SLC22A23	solute carrier family 22 member 23		3269207	3456793	187587
11	4915072	rs18164482.834e-05	NA	NA		NA	NA	NA

```
dim(t)
```

```
[1] 15 9
```

Here's an alternate way to construct the SNP table which is an incomplete example because it doesn't include the gene boundaries. However, it is simpler than the function above because it avoids the construction of a key and the left_joins used above by looping through each SNP, one by one:

```
snp.table2 <- function(snp.list, N=15) {
  require(TxDb.Hsapiens.UCSC.hg19.knownGene)
  require(org.Hs.eg.db)

  txdb <- TxDb.Hsapiens.UCSC.hg19.knownGene
  tx.by.gene <- transcriptsBy(txdb, "gene")
  gene.name <- data.frame()

  for (i in 1:N) {
    gene.name[i, 1] <- snp.list[i,]$CHR
    gene.name[i, 2] <- snp.list[i,]$BP
    gene.name[i, 3] <- snp.list[i,]$SNP
    gene.name[i, 4] <- snp.list[i,]$P
    top.snp <- with(snp.list[i,], GRanges(seqnames=paste0("chr", CHR),
                                           IRanges(start=BP, width=1,
                                                   rsid=SNP, P=P)))
    hits <- subsetByOverlaps(tx.by.gene, top.snp)
    genename <- select(org.Hs.eg.db, keys=names(hits), columns=c("SYMBOL", "GENENAME"),
                        gene.name[i, 5] <- ifelse(nrow(genename) == 0, NA, genename[[2]])
                        gene.name[i, 6] <- ifelse(nrow(genename) == 0, NA, genename[[3]]))
  }
  colnames(gene.name) <- c("CHR", "BP", "SNP", "P", "Symbol", "Gene name")
  return(gene.name)
}

(t <- snp.table2(top.hits, 15)) %>% kable(digits=15)

'select()' returned 1:1 mapping between keys and columns
```

```
'select()' returned 1:1 mapping between keys and columns
```

CHR	BP	SNP	P	Symbol	Gene name
6	1942538	rs3800143	4.608e-11	GMDS	GDP-mannose 4,6-dehydratase
11	4913057	rs10836914	1.295e-07	NA	NA
11	4913314	rs12577475	1.587e-07	NA	NA
6	1926650	rs11242725	3.785e-07	GMDS	GDP-mannose 4,6-dehydratase
6	1880964	rs3800116	1.475e-06	GMDS	GDP-mannose 4,6-dehydratase
6	1915129	rs3778552	1.646e-06	GMDS	GDP-mannose 4,6-dehydratase
6	1908210	rs2875711	2.550e-06	GMDS	GDP-mannose 4,6-dehydratase
6	1955398	rs9378664	2.809e-06	GMDS	GDP-mannose 4,6-dehydratase
6	143144885	rs926372	5.704e-06	HIVEP2	HIVEP zinc finger 2
11	4912192	rs10836912	8.977e-06	NA	NA
6	1899671	rs3800122	1.022e-05	GMDS	GDP-mannose 4,6-dehydratase
11	4910224	rs2570591	1.656e-05	NA	NA
11	6340706	rs1051992	2.356e-05	CAVIN3	caveolae associated protein 3
6	3317016	rs4959804	2.821e-05	SLC22A23	solute carrier family 22 member 23
11	4915072	rs1816448	2.834e-05	NA	NA

```
dim(t)
```

```
[1] 15 6
```

25.13 Question 3

Task: Using GRanges and associated Bioconductor tools, write a function that takes as input a ranked list of the SNPs, and returns a nice table that lists the top N of these SNPs and the **closest flanking genes** on both sides, including the SNP position and the gene boundaries.

```
# snp.list = the ranked list of the top SNPs
# N = the number of top SNPs to annotate
snp.flank <- function(snp.list, N=15) {
```

}

Apply your `snp.flank` function to the top 15 SNPs in our example data set.

Note that the purpose here is to think more about how to creatively use GRanges than to search for nice annotation packages.

 Hint

The `GenomicRanges` package has a useful function `nearest` that can be used to find the nearest gene.

Note that if we want to augment information about the nearest gene with the genes that precede and follow each top SNP, we could use functions like this:

```
genes <- genes(txdb, columns = "gene_id")
precede(top.snp, genes)
follow(top.snp, genes)
```

 Expand to see solution

25.14 Answer 3

Note that we need to set up the `txdb` object within the function itself to avoid creating a dependence on the global `txdb` object. Although, it might be more efficient to create the `txdb` object once and pass it in via a function parameter instead of recreating it each time the function is called.

Here we use `require` statements to indicate dependence on certain libraries having been loaded, but if this were part of an R package, we'd take care of library dependencies at the package level instead of inside of specific functions. Usually when writing functions, we assume that all of the required packages have been loaded, so we tend not to use the `library` or the `require` command within functions.

```

# snp.list = the ranked list of the top SNPs
# N = the number of top SNPs to annotate
snp.flank <- function(snp.list, N=15) {
  require(org.Hs.eg.db)
  require(TxDb.Hsapiens.UCSC.hg19.knownGene)
  txdb <- TxDb.Hsapiens.UCSC.hg19.knownGene
  tx.by.gene <- transcriptsBy(txdb, "gene")
  # Find the gene boundaries
  gene.bounds <- reduce(tx.by.gene)
  # Set up a GRange with the first N top SNPs
  top.snp <- with(snp.list[1:N, ], GRanges(seqnames = paste0("chr", CHR),
                                             IRanges(start = BP, width = 1),
                                             SNP = SNP, P = P))
  # Find overlaps and hits
  top.snp.gene <- findOverlaps(gene.bounds, top.snp)
  hits <- subsetByOverlaps(gene.bounds, top.snp)
  # The SNP hits
 .snp.info <- data.frame(SNP.ID = subjectHits(top.snp.gene),
                           SNP.chr = seqnames(top.snp[subjectHits(top.snp.gene)]),
                           ranges(top.snp[subjectHits(top.snp.gene)]),
                           mcols(top.snp[subjectHits(top.snp.gene)]))
  # The Gene hits
  gene.info <- data.frame(gene.ID = queryHits(top.snp.gene),
                           seqnames(gene.bounds[queryHits(top.snp.gene)]),
                           ranges(gene.bounds[queryHits(top.snp.gene)]))
  # Reduce gene.info to distinct entries
  gene.info <- gene.info %>% dplyr::select(-group, -group.1) %>% distinct()
  # Construct a key linking SNPs to Genes
  key <- data.frame(gene.ID = queryHits(top.snp.gene),
                     SNP.ID = subjectHits(top.snp.gene))
 .snp.gene <- key %>% left_join(gene.info, by = "gene.ID") %>%
    left_join(.snp.info, by = "SNP.ID")
  gene.name <- select(org.Hs.eg.db, keys = names(hits),
                      columns = c("ENTREZID", "SYMBOL", "GENENAME"), keytype = "ENTREZID")
  .snp.gene <- .snp.gene %>% dplyr::rename(ENTREZID = group_name) %>%
    left_join(gene.name, by = "ENTREZID")
  .snp.gene <- .snp.gene %>% arrange(P)
  .snp.gene <- left_join(snp.list[1:N, ], .snp.gene, by = c("SNP"))
  .snp.gene <- .snp.gene %>%
    dplyr::select(CHR, BP, SNP, P.x, SYMBOL, GENENAME, start.x, end.x, width.x)
  .snp.gene <- .snp.gene %>%
    dplyr::rename(P = P.x, start = start.x, end = end.x, width = width.x)
  # Now find the nearest genes
  genes <- genes(txdb, columns = "gene_id")
  # Find the Nearest gene
  nearest_gene_index <- nearest(213top.snp, genes)
  EntrezID <- unlist(genes[nearest_gene_index]$gene_id)
  lookup <- select(org.Hs.eg.db, keys = EntrezID, keytype = "ENTREZID",
                    columns = c("SYMBOL", "GENENAME"))
  symbol <- lookup$SYMBOL
  genename <- lookup$GENENAME
  nearest <- bind_cols(SNP = mcols(top.snp)$SNP, EntrezID = EntrezID,
                        symbol = symbol, genename = genename)
}

```

```
(t <- snp.flank(top.hits, 15)) %>% kable(digits=15)

Warning in .local(x, row.names, optional, ...): 'optional' argument was ignored
'select()' returned 1:1 mapping between keys and columns

403 genes were dropped because they have exons located on both strands
of the same reference sequence or on more than one reference sequence,
so cannot be represented by a single genomic range.
Use 'single.strand.genes.only=FALSE' to get all the genes in a
GRangesList object, or use suppressMessages() to suppress this message.

'select()' returned many:1 mapping between keys and columns
'select()' returned many:1 mapping between keys and columns
'select()' returned many:1 mapping between keys and columns
```

CHBP	SNIP	SYMBOL	GENE NAME	wid	Nearest	GENE NAME	wid	GENE NAME	wid	GENE NAME	wid	GENE NAME	wid
6	19425380	0184MDS	SP-162	20458183	MDS	SP-162	20458183	MDS	22459874	Xkhead	610684129		
	11	mannose		mannose		DT diver-				box			
		4,6-		4,6-		gent				C1			
		dehydratase		dehydratase		trans-				script			
11	491305782654	NA	NANAOR51	factory	49029000R51	factory	49289000R51	factory	49029000R51	factory	49029000R51		
	07		re-		re-		re-		re-				
			cep-		cep-		cep-		cep-				
			tor		tor		tor		tor				
			fam-		fam-		fam-		fam-				
			ily 51		ily 51		ily 51		ily 51				
			sub-		sub-		sub-		sub-				
			fam-		fam-		fam-		fam-				
			ily T		ily A		ily A		ily T				
			mem-		mem-		mem-		mem-				
			ber 1		ber 7		ber 7		ber 1				

11	49133125.5875NA	NANANAOR51factory	4902000IR51factory	4928000IR51factory	4902000113
	07	re- cep- tor fam- ily 51 sub- fam- ily T mem- ber 1	re- cep- tor fam- ily 51 sub- fam- ily A mem- ber 7	re- cep- tor fam- ily 51 sub- fam- ily T mem- ber 1	re-
6	192665024264MGS P- 1624245818MGS P- 1624245818MGS D S 224298T0XGkhead610684129				
	07	mannose 4,6- dehydratase	mannose 4,6- dehydratase	DT diver- gent trans- script	box C1
6	1880968104764MGS P- 1624245818MGS P- 1624245818MGS D S 224298T0XGkhead610684129				
	06	mannose 4,6- dehydratase	mannose 4,6- dehydratase	DT diver- gent trans- script	box C1
6	19151377.8564MGS P- 1624245818MGS P- 1624245818MGS D S 224298T0XGkhead610684129				
	06	mannose 4,6- dehydratase	mannose 4,6- dehydratase	DT diver- gent trans- script	box C1
6	19082187.5704MGS P- 1624245818MGS P- 1624245818MGS D S 224298T0XGkhead610684129				
	06	mannose 4,6- dehydratase	mannose 4,6- dehydratase	DT diver- gent trans- script	box C1
6	19553937.8664MGS P- 1624245818MGS P- 1624245818MGS D S 224298T0XGkhead610684129				
	06	mannose 4,6- dehydratase	mannose 4,6- dehydratase	DT diver- gent trans- script	box C1

6	1431928371	143074300633	SHV EP	143073100615	Ba10112147519061	277143287358719
06	zinc	zinc		LOC153910	inter-	
	fin-	fin-			genic	
	ger	ger 2			non-	
	2				protein	
					cod-	
					ing	
					RNA	
					1277	
11	491219283671	ANA	NANANAOR51	Editor	4904900R51	Editor
06		re-		re-	re-	
		cep-		cep-	cep-	
		tor		tor	tor	
		fam-		fam-	fam-	
		ily 51		ily 51	ily 51	
		sub-		sub-	sub-	
		fam-		fam-	fam-	
		ily T		ily A	ily T	
		mem-		mem-	mem-	
		ber 1		ber 7	ber 1	
6	189967800224	MDS	-1624245863MDS	-1624245863MDS	224298740X6	khead610684129
05	mannose	mannose	DT diver-		box	
	4,6-	4,6-	gent		C1	
	dehydratase	dehydratase	trans-			
			script			
11	491022570581	ANA	NANANAOR51	Editor	4904900R51	Editor
05		re-		re-	re-	
		cep-		cep-	cep-	
		tor		tor	tor	
		fam-		fam-	fam-	
		ily 51		ily 51	ily 51	
		sub-		sub-	sub-	
		fam-		fam-	fam-	
		ily T		ily A	ily T	
		mem-		mem-	mem-	
		ber 1		ber 7	ber 1	

11	634070613902AVIN	634083403741560CAVIN	634083403741560CAVIN	lae634083403741560MBP	Dillingo641411622KBR	Recys6281903357
05	as-	asso-	phos-	B re-		
	so-	ci-	pho-	cep-		
	ci-	ated	di-	tor		
	ated	pro-	esterase			
	pro-	tein	1			
	tein	3				
	3					
6	3317940598261G22A23269240787387G22A23	3269240779BB2Bn	3223297981	leas03259162300		
05	car-	car-	beta	as-		
	rier	rier	2B	sem-		
	fam-	fam-	class	bly		
	ily	ily 22	IIb	chap-		
	22	mem-		erone		
	mem-	ber		4		
	ber	23				
	23					
11	49150172164NA	NANAOR51Editor	49020000R51Editor	49280000R51Editor	49020004113	
05		re-	re-	re-		
		cep-	cep-	cep-		
		tor	tor	tor		
		fam-	fam-	fam-		
		ily 51	ily 51	ily 51		
		sub-	sub-	sub-		
		fam-	fam-	fam-		
		ily T	ily A	ily T		
		mem-	mem-	mem-		
		ber 1	ber 7	ber 1		

`dim(t)`

`[1] 15 21`

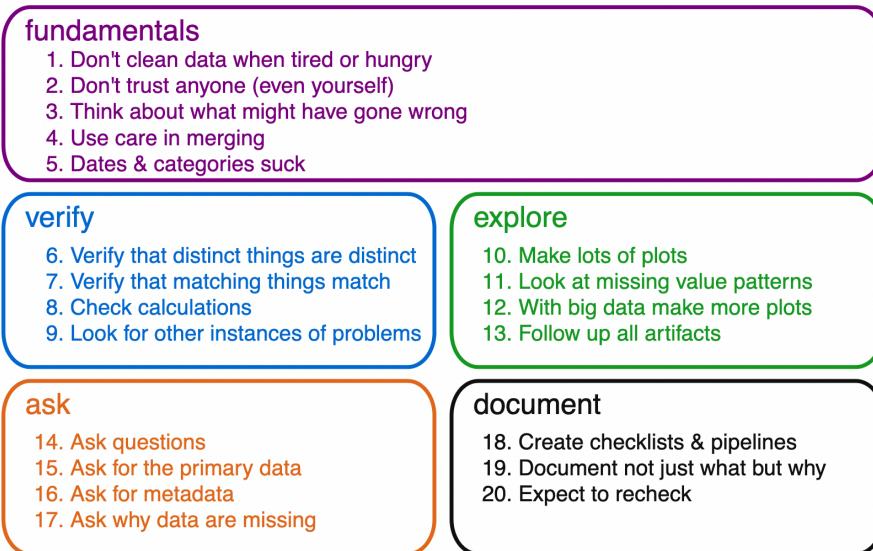
26 Data Quality Checking

26.1 Data cleaning principles

Data cleaning principles from Karl Broman's slide set:

https://kbroman.org/Talk_DataCleaning/data_cleaning.pdf

Data cleaning principles



26.2 dbGaP quality control

As described in:

Tryka KA, Hao L, Sturcke A, Jin Y, Wang ZY, Ziyabari L, Lee M, Popova N, Sharopova N, Kimura M, Feolo M. NCBI's Database of Genotypes and Phenotypes: dbGaP. Nucleic Acids Research. 2014 Jan 1;42(D1):D975–D979. PMID: 24297256 PMCID: PMC3965052 DOI: <https://doi.org/10.1093/nar/gkt1211>

“The Database of Genotypes and Phenotypes (dbGap, <http://www.ncbi.nlm.nih.gov/gap>) is a National Institutes of Health-sponsored repository charged to archive, curate and distribute information produced by studies investigating the interaction of genotype and phenotype.”

Under NIH data sharing guidelines, all properly consented large-scale genetic or 'omics studies must deposit their data in dbGaP. To do so, one must closely follow the formatting requirements as described in the dbGaP Submission Guide:

<https://www.ncbi.nlm.nih.gov/gap/docs/submittingguide/>

This involves setting up a data dictionary that follows dbGaP specifications:

Column Headers	Description
VARNAME*	Variable name. The VARNAME must not contain backward slashes (\). Do not use "dbGaP" in the variable name. "dbGaP" is reserved for dbGaP generated items.
VARDESC*	Variable description. The description should be understandable and enable users to replicate the variable. For example, "blood pressure" is useful, but "brachial blood pressure while sitting" provides more context. Alternatively, study documents with detail are also acceptable.
DOCFILE	Study document name associated with the variable. To list multiple documents, add a semicolon (;) between documents. Please list only study document filenames that are submitted to dbGaP.
TYPE	Data value type: integer (1,2,3,4,...), encoded value (integers or strings are coded for non-numerical meaning, ex. 1=Control; 2=Case, see VALUES), decimal (0.5,2.5,...), string (African American, Asian, Caucasian, Hispanic, Non-Hispanic). For mixed values (any combination of string, integers, decimals and/or encoded values) in a single data column, list all types present.
UNITS*	Units of measurement of variable
MIN	The logical minimum value of the variable. If a separate code such as -1 is used for a missing field, this should not be considered as the MIN value.
MAX	The logical maximum value for the variable. If a separate code such as 9999 is used for a missing field, this should not be considered as the MAX value.
RESOLUTION	Measurement resolution – the number of decimal places to which a measured value is presented in the data. For example, in 54.321 the resolution is 3.
COMMENT1, COMMENT2	Additional information not included in the VARDESC that will further define the variable. If additional comments are needed beyond COMMENT2, insert new columns (COMMENT3, COMMENT4, etc.) before the column "ORDER."
VARIABLE_SOURCE	Source of controlled vocabularies. Ex. PhenX, MeSH, SNOMED, NCI. If there is no match, leave blank. (Must be submitted as a group with SOURCE_VARIABLE_ID and VARIABLE_MAPPING).
SOURCE_VARIABLE_ID	A unique identifier from the VARIABLE_SOURCE or a unique text concept/term from various controlled vocabularies. (Must be submitted as a group with VARIABLE_SOURCE and VARIABLE_MAPPING).
VARIABLE_MAPPING	For example, a variable from the source could be Identical, Related, or Comparable. (Must be submitted as a group with VARIABLE_SOURCE and SOURCE_VARIABLE_ID).
UNIQUEKEY	Unique key is a combination of variables that is designed to uniquely identify a row in a longitudinal dataset or rows that have repeating SUBJECT_IDS or SAMPLE_IDS. Mark "X" for variables that constitute the unique keys, and leave other values blank. Ex. SUBJECT_ID and VISIT_NUMBER. UNIQUEKEYs can only be used in the subject phenotypes file and some cases of the sample attributes file. The SC, SSM, and pedigree files should never have UNIQUEKEYs marked, since there should be a unique identifier appearing once in each file.
COLLINTERVAL	Collection interval is the time frame in which the data for the variable or dataset was collected.
ORDER	The order in which VALUES appear on the variable summary report page. If VALUES of a single variable/column of data are integers or decimals, leave blank. If VALUES are encoded values, string, or mixed, define the order. VALUES can be ordered by Frequency (highest to lowest frequency of VALUES) or by List (user specifies order through placement in VALUES columns). For mixed values within a single variable/column of data, see examples: "age" and "weight" in example file 5b_SubjectPhenotypes_DD.xlsx .
VALUES*	List of all unique values and/or descriptions of all encoded values, one value per cell. Encoded values are defined as a value and its meaning. For example, if a data file contains a variable named "EDUCATION" and its data values are "1, 2, 3, and 99," these coded values will need to be defined in the data dictionary. The format of an encoded value is VALUE=MEANING . Therefore, in the data dictionary, there should be 4 separate data cells filled out with the following: 1=Completed High School, 2=Completed College, 3=Completed Graduate School, 99=Unknown. The "VALUES" header must be the last column header (farthest right in the table). It should appear only in the column above the first encoded value that is listed. The remaining column header cells should be left blank. The script will identify the first code meanings and continue right until there are no more code meanings. For example, if the variable "SEX" has 3 encoded values: 1=Male, 2=Female and 3=Unknown, the column header "VALUES" will appear only above the cell that contains 1=Male. 1=Male, 2=Female and 3=Unknown will be listed in three separate cells next to each other. The header column cells above "2=Female" and "3=Unknown" should be left blank.

26.3 Minimum and Maximum Values Check

26.3.1 MIN, MAX check

In the data dictionary, for some variables, MIN and MAX values may be specified. For example, for age, it has a natural minimum of zero.

MIN The logical minimum value of the variable. If a separate code such as -1 is used for
MAX The logical maximum value for the variable. If a separate code such as 9999 is used :

Task: Design and implement a check that the specified MIN and MAX values observed in the data are consistent with the values as specified in the data dictionary.

```
suppressMessages(library(tidyverse))
library(dbGaPCheckup)
## Load DD.dict.I and DS.data.I
data(ExampleI)
```

Using DD.dict.I and DS.data.I, check the PERCEIVED_CONFLICT variable to see if all the values fall within the stated MIN and MAX values.

```
DD.dict.I %>%
  filter(VARNAME=="PERCEIVED_CONFLICT") %>%
  select(VARNAME,MIN,MAX)
```

```
# A tibble: 1 x 3
  VARNAME      MIN   MAX
  <chr>     <dbl> <dbl>
1 PERCEIVED_CONFLICT     1     15
```

26.3.2 Pseudo-code

💡 Hint

First try to write out an algorithm for this Minimum and Maximum Values Check in pseuo-code, outlining each step.

💡 Expand to see solution

Possible steps:

1. Read the vector of PERCEIVED_CONFLICT from DS.data.I
2. Read the MIN and MAX values for PERCEIVED_CONFLICT from DD.dict.I
3. Count and list any PERCEIVED_CONFLICT values that lie outside of the range [MIN, ..., MAX].

26.3.3 Implement MIN, MAX check in R

Implement your algorithm in code.

💡 Expand to see solution

```
# Read the vector of `PERCEIVED_CONFLICT` from `DS.data.I`  
trait <- DS.data.I$PERCEIVED_CONFLICT  
# Read the `MIN` and `MAX` values for `PERCEIVED_CONFLICT` from `DD.dict.I`  
min.val <- DD.dict.I %>%  
  filter(VARNAME=="PERCEIVED_CONFLICT") %>%  
  pull(MIN)  
max.val <- DD.dict.I %>%  
  filter(VARNAME=="PERCEIVED_CONFLICT") %>%  
  pull(MAX)  
# List any `PERCEIVED_CONFLICT` values that lie outside of the range [`MIN`, ..., `MAX`]  
trait[trait < min.val | trait > max.val] %>%  
  unique() %>%  
  sort()  
  
[1] 16 17 19 20 21 24 25 26 28 29 30  
  
# Count any `PERCEIVED_CONFLICT` values that lie outside of the range [`MIN`, ..., `MAX`]  
trait[trait < min.val | trait > max.val] %>%  
  length()  
  
[1] 44
```

These results are consistent with those returned by the `minmax_check` function from the `dbGaPCheckup` R package:

```
details <- minmax_check(DD.dict.I, DS.data.I, non.NA.missing.codes=c(-9999, -4444))$Info  
  
$Message  
[1] "ERROR: some variables have values outside of the MIN to MAX range."  
  
$Information  
# A tibble: 1 x 5  
  Trait           Check ListedMin ListedMax OutOfRangeValues  
  <chr>          <lgl>    <dbl>     <dbl> <list>  
1 PERCEIVED_CONFLICT FALSE        1         15 <int [11]>
```

```
details[[1]]$OutOfRangeValues[[1]] %>%  
  sort()
```

```
[1] 16 17 19 20 21 24 25 26 28 29 30
```

26.3.4 Make your check more robust

After implementing your algorithm in R code, think about it a bit further - is it robust to the situation where only one of the MIN and MAX values is specified and the other is missing? Is it robust to the situation where both MIN and MAX are missing?

 Expand to see solution

```
# List any `PERCEIVED_CONFLICT` values that lie outside of the range [`MIN`, ..., `MAX`]  
trait[trait < min.val | trait > max.val] %>%  
  unique() %>%  
  sort()
```

```
[1] 16 17 19 20 21 24 25 26 28 29 30
```

The code proposed here is not robust to MIN or MAX being NA because, for example, if MIN is NA and MAX is 15, in some situations the logical indexing into the `trait` vector used of `trait < min.val | trait > max.val` would return NA instead of TRUE or FALSE as intended.

```
# MIN=NA, MAX=15  
16 < NA | 16 > 15
```

```
[1] TRUE
```

```
# MIN=1, MAX=NA  
16 < 1 | 16 > NA
```

```
[1] NA
```

```
# MIN=NA, MAX=NA  
16 < NA | 16 > NA
```

```
[1] NA
```

This are possible steps toward writing a more robust check:

```
vals.low <- NA
vals.high <- NA
if (!is.na(min.val)) {
  vals.low <- trait[trait < min.val]
}
if (!is.na(max.val)) {
  vals.high <- trait[trait > max.val]
}

vals.OutOfRange <- c(vals.low,vals.high)
vals.OutOfRange %>%
  na.omit() %>%
  unique() %>%
  sort()
```

```
[1] 16 17 19 20 21 24 25 26 28 29 30
```

But what would the above code return if both MIN and MAX were NA?

26.3.5 Check the PREGNANT variable

Now apply your MIN and MAX checking algorithm to the PREGNANT variable.

```
DD.dict.I %>%
  filter(VARNAME=="PREGNANT") %>%
  select(VARNAME,MIN,MAX)

# A tibble: 1 x 3
  VARNAME    MIN    MAX
  <chr>     <dbl> <dbl>
1 PREGNANT      0      1
```

 Expand to see solution

```
# Read the vector of `PREGNANT` from `DS.data.I`  
trait <- DS.data.I$PREGNANT  
# Read the `MIN` and `MAX` values for `PREGNANT` from `DD.dict.I`  
min.val <- DD.dict.I %>%  
  filter(VARNAME=="PREGNANT") %>%  
  pull(MIN)  
max.val <- DD.dict.I %>%  
  filter(VARNAME=="PREGNANT") %>%  
  pull(MAX)  
# List any `PREGNANT` values that lie outside of the range [`MIN`, ..., `MAX`].  
trait[trait < min.val | trait > max.val] %>%  
  unique() %>%  
  sort()  
  
[1] -9999 -4444  
  
# Count any `PREGNANT` values that lie outside of the range [`MIN`, ..., `MAX`].  
trait[trait < min.val | trait > max.val] %>%  
  length()  
  
[1] 53
```

These out-of-range values of `-9999` and `-4444` look kind of strange and are unexpected given the first two entries of the `VALUES` column of the data dictionary for this variable:

```
DD.dict.I[which(DD.dict.I=="PREGNANT"),c(1,17,18)]  
  
# A tibble: 1 x 3  
  VARNAME  VALUES ...18  
  <chr>    <chr>  <chr>  
1 PREGNANT 0=no   1=yes
```

Based on this, we'd expect to see only 0 and 1 values in the `PREGNANT` variable. What's going on?

26.3.6 Handle missing values

If we further check the data dictionary for the PREGNANT variable, we see that the out-of-range values we observed in our check above are actually missing value codes and so should not be flagged as being out of range.

```
DD.dict.I[which(DD.dict.I=="PREGNANT"),c(1,19,20)]  
  
# A tibble: 1 x 3  
  VARNAME    ...19          ...20  
  <chr>      <chr>        <chr>  
1 PREGNANT -9999=missing value -4444=not applicable, participant assigned male ~
```

Extend your algorithm to handle missing value codes. To do this first outline your approach in pseudo-code.

 Expand to see solution

Possible steps:

1. Read the vector of PREGNANT from DS.data.I
2. Read the MIN and MAX values for PREGNANT from DD.dict.I
3. Have the user provide a list of missing value codes
4. Recode any PREGNANT value that matches one of the missing value codes to the standard NA R missing value code.
5. Count and list any non-missing PREGNANT values that lie outside of the range [MIN, ..., MAX].

This is essentially the approach used in the `minmax_check` function of the `dbGaPCheckup` R package.

```
# Without missing value codes specified  
details <- minmax_check(DD.dict.I, DS.data.I)$Information  
  
$Message  
[1] "ERROR: some variables have values outside of the MIN to MAX range."  
  
$Information  
# A tibble: 2 x 5  
  Trait           Check ListedMin ListedMax OutOfRangeValues  
  <chr>          <lgl>    <dbl>     <dbl>   <list>  
1 PREGNANT       FALSE      0         1 <int [2]>  
2 PERCEIVED_CONFLICT FALSE      1         15 <int [11]>
```

```

details[[1]]$OutOfRangeValues

[[1]]
[1] -4444 -9999

[[2]]
[1] 25 24 16 28 17 21 30 19 26 20 29

# With missing value codes specified
# PREGNANT is no longer flagged as having out of range values.
details <- minmax_check(DD.dict.I, DS.data.I, non.NA.missing.codes=c(-9999, -4444))$Info

$Message
[1] "ERROR: some variables have values outside of the MIN to MAX range."

$Information
# A tibble: 1 x 5
  Trait      Check ListedMin ListedMax OutOfRangeValues
  <chr>     <lgl>    <dbl>     <dbl>   <list>
1 PERCEIVED_CONFLICT FALSE        1        15 <int [11]>

```

If we examine the `minmax_check` code by typing `minmax_check` without parentheses at the R prompt, we see that this is how the missing value recoding step is done:

```

for (value in na.omit(non.NA.missing.codes)) {
  dataset_na <- dataset_na %>% mutate(across(everything(),
    ~na_if(.x, value)))
}

```

27 Questions about R

27.1 Adding a new column to a data frame

Which of these is an **incorrect** way to add a new column to a data frame in R?**

- (A) `df <- cbind(df, new_column3 = c(1, 2, 3, 4))`
- (B) `new_column(df) <- c(1, 2, 3, 4)`
- (C) `df$new_column <- c(1, 2, 3, 4)`
- (D) `df[, 'new_column'] <- c(1, 2, 3, 4)`

28 R gotchas

28.1 Mis-counting

This example is modeled on this Mastodon post:

<https://fediscience.org/@thadryanjs/111188342897535820>

```
(df <- data.frame(a=c(1,1,1,2), b=c(1,1,NA,2)))  
  
   a   b  
1 1   1  
2 1   1  
3 1 NA  
4 2   2  
  
# How many times is 1 in column a  
nrow(df[df$a == 1,])  
  
[1] 3  
  
# How many times is 1 in column b  
nrow(df[df$b == 1,])  
  
[1] 3
```

As there are only two 1's in column **b**, this answer of 3 is incorrect.

What's going on here?

What's a correct way to count the number 1's in each of these two columns?

 Expand to see solution

This doesn't work because of the NA causes this to return three rows:

```
df[df$b == 1,]
```

	a	b
1	1	1
2	1	1
NA	NA	NA

Using a `data.table` instead of a `data.frame` would work:

```
library(data.table)
(dt <- data.table(a=c(1,1,1,2), b=c(1,1,NA,2)))
```

	a	b
1:	1	1
2:	1	1
3:	1	NA
4:	2	2

```
# How many times is 1 in column a
nrow(dt[dt$a == 1,])
```

```
[1] 3
```

```
# How many times is 1 in column b
nrow(dt[dt$b == 1,])
```

```
[1] 2
```

Counting it more directly is another possibility:

```
sum(df$b==1, na.rm = TRUE)
```

```
[1] 2
```

Tidyverse commands also gives the correct answer:

```
suppressMessages(library(tidyverse))
df %>% filter(b == 1) %>% nrow()
```

```
[1] 2
```

28.2 Are there any r's in the vector LETTERS?

I used ‘which’ to determine there were zero copies of the target in the vector of interest, but then testing whether the answer returned by ‘which’ is zero is tricky.

See discussion on Mastodon here:

<https://fediscience.org/@StatGenDan/111052432535136731>

```
# LETTERS contains the uppercase letters
LETTERS

[1] "A" "B" "C" "D" "E" "F" "G" "H" "I" "J" "K" "L" "M" "N" "O" "P" "Q" "R" "S"
[20] "T" "U" "V" "W" "X" "Y" "Z"

# This returns a vector of length zero:
which(LETTERS == "r")

integer(0)

# Testing if it is equal to the integer zero does not work
0 == which(LETTERS == "r")

logical(0)

# Testing if it is equal to the integer one does not work
1 == which(LETTERS == "r")

logical(0)
```

So what is more correct way to test if there are any r is present in the LETTERS vector?

Thomas Lumley commented:

“In general, there are functions in R that return a length-1 answer (any, length, sum, min,...) and there are functions that return a variable-length answer (==, which, +, -,...). You have a length-1 question: are there any ’r’s? You need a function with a fixed length-1 return value.”

<https://fediscience.org/@tslumley/111053882380113100>

 Expand to see solution

```
# Number of r's in LETTERS  
length(which(LETTERS == "r"))  
  
[1] 0  
  
# Number of r's in LETTERS  
sum(LETTERS=='r')  
  
[1] 0  
  
# Is r present in LETTERS?  
any(LETTERS=='r')  
  
[1] FALSE
```

28.3 Strange R behavior

June Choe shared this on Mastodon

<https://fosstodon.org/@yjunechoe/111026163637396686>

A student in my intro #rstats class taught me something new today (by way of a cryptic “bug”).

Suppose you’re asked why this {purrr} code that should return the mean of each list element is not working as expected.

```
map(list(x=1:3, y=4:6), mean)  
#> $x  
#> [1] 1  
#>  
#> $y  
#> [1] 4
```

What do you think is the simplest explanation for this behavior (in terms of the mistake that the student could’ve made)? It’s not so obvious - there are multiple R “quirks” cascading!

 Expand to see solution

```
library(purrr)
set.seed(123)
mean <- mean(sample(2, 10, replace=TRUE))
mean

[1] 1.4

# These means are correct
mean(1:3)

[1] 2

mean(4:6)

[1] 5

# These means are correct:
lapply(list(x=1:3, y=4:6), mean)

$x
[1] 2

$y
[1] 5

# But these means are incorrect:
map(list(x=1:3, y=4:6), mean)

$x
[1] 1

$y
[1] 4
```

Why are the means computed using the `map` function from the `purrr` package incorrect? It is not applying the `mean` function, but rather it is applying the `mean` variable, which has a value of 1.4.

As the `map` documentation states, while the `map` command is typically used to apply a function in its `.f` argument, the `.f` argument can also accept an integer - when it does so, it is interpreted as follows:

A string, integer, or list, e.g. "idx", 1, or list("idx", 1) which are shorthand for pluck(x, "idx"), pluck(x, 1), and pluck(x, "idx", 1) respectively.

So when we `map` using the `mean` variable, it is used as an index to pluck elements out of the list - during the double to integer conversion, it is rounded down to 1, so it plucks the first element of each list.

```
map(list(x=1:3, y=4:6), 1.4)

$x
[1] 1

$y
[1] 4

map(list(x=1:3, y=4:6), 1)

$x
[1] 1

$y
[1] 4
```

Moral: Be careful to avoid using existing R function names, like `mean`, as the names of your variables.

Relevant discussion can be found in

<https://adv-r.hadley.nz/functions.html#functions-versus-variables>

where it is stated:

“For the record, using the same name for different things is confusing and best avoided!”

29 Basic Shell Commands

29.1 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.

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29.2 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. <i>Note:</i> this command does NOT work on Windows machines (Mac and Linux are okay)
cd	changes the current directory to the directory <code>dirname</code>
<code>./dirname</code>	
<code>ls -F</code>	tells you what files and directories are in the current directory
<code>pwd</code>	tells you what directory you are in (<code>pwd</code> stands for <i>print working directory</i>)
<code>history</code>	lists previous commands you have entered. <code>history less</code> lets you page through the list.
<code>man cmd</code>	displays the <i>manual</i> page for a command.

29.3 Creating Things:

29.3.1 How to create new files and directories..

Command Definition

`mkdir` makes a new directory called `dirname` below the current directory. *Note:* Windows `./dirname`s 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` exists, `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)

29.3.2 How to delete files and directories...

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

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

29.3.3 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/filename` 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
.

29.4 Pipes and Filters

29.4.1 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.

29.4.1.1 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.

29.4.2 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).

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

29.4.2.1 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 |.

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

29.4.2.1.1 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.

29.5 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.

29.5.1 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.

29.5.2 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

29.5.2.1 Commonly used conditional operators

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

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

29.6 Finding Things

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

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

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

29.6.1.1 Commonly used grep options

grep options	
-E	tells grep you will be using a regular expression. Enclose the regular expression in quotes. <i>Note:</i> the power of <code>grep</code> 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

29.6.2 How to find files with certain properties...

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

Example	com-	mand	Description
		<code>find .</code>	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
		<code>-type d</code>	

29.6.2.1 Commonly used find options

find options

-type d lists directories; f lists files
[df]

-maxdepth find automatically searches subdirectories. If you don't want that, specify the
n number of levels below the working directory you would like to search

-mindepth starts **find**'s search n levels below the working directory
n

30 Summary

In summary, this book is a work in progress.

References

A Technical Details

A.1 Quarto

This book was build using [Quarto](#).

A.1.1 Callout blocks

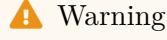
To hide a solution that then can be clicked to view, we use a `.callout-tip collapse="true"` callout block.

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.



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.

A.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.

A.2 Previewing the book

Type `quarto preview` in the Terminal window.

A.3 Deploying the book to GitHub Pages

Type `quarto publish` in the Terminal window.

A.4 Deploying the book to Netlify

Type `quarto publish netlify` in the Terminal window.

A.5 Multiple choice questions

To create multiple choice questions, use functions from the `webexercises` R package.

The multiple choice question below is created by the inline R code

```
r longmcq(opts_ci)
```

What is true about a 95% confidence interval of the mean?

```
opts_ci <- c(
  answer = "If you repeated the process many times, 95% of intervals calculated in this way
  \"There is a 95% probability that the true mean lies within this range\",
  \"Approximately 95% of the data fall within this range\""
)
```

- (A) If you repeated the process many times, 95% of intervals calculated in this way contain the true mean

- (B) There is a 95% probability that the true mean lies within this range
- (C) Approximately 95% of the data fall within this range

A.6 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 `webr-r` chunks.

```
# Edit/add/try out R code here
```

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
```

A.7 embedpdf Quarto extension

This book uses the `embedpdf` Quarto extension from <https://github.com/jmgirard/embedpdf>, which was installed via this command:

```
quarto add jmgirard/embedpdf
```

To embed a PDF, use code like this:

```
{ {< pdf dummy.pdf width=100% height=800 >} }
```

However, the PDF embedding done this way did not work in Chrome.

Example:

So instead we used an iframe, which works on Chrome, Firefox, and Safari:

```
<iframe width="100%" height="800" src="pdfs/GitHubIntro.pdf">
```

Note that for iframe embedding of Panopto video from the University of Pittsburgh, one needs to use a credentialless iframe.

B 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)
library(ggplot2)
ggplot(mtcars, aes(x=am,y=mpg)) +
  geom_point() +
  geom_smooth(method="lm")
```

B.1 Link: [WebR](#) and [quarto-webr](#).

C JSLinux terminal

C.1 Interactive Linux terminal

Here is an interactive Linux terminal (x86 Alpine Linux 3.12.0) created by the [JSLinux](#) project.

You can upload files to it by clicking the arrow below the terminal window.

This virtual Unix machine has a throttled very slow connection to the internet, so to install files and programs, it is better to download them to your computer outside of the browser, and then use the upload button to upload them onto this virtual machine.

For example, the 32-bit version of PLINK2 can be installed in this manner:

1. Download the Linux 32-bit PLINK2 zip file from <https://www.cog-genomics.org/plink/2.0/>
2. Unzip the downloaded file.
3. Use the upload arrow here below the terminal window to upload the plink2 binary.
4. Make the plink2 binary executable via this Unix command: `chmod +x plink2`
5. Run PLINK2 via this Unix command: `./plink2`

D webLinux terminal

D.1 Interactive Linux terminal

Here is an interactive Linux terminal created by the [webLinux](https://github.com/remisharrock/sysbuild) project. The source code for this project can be found at <https://github.com/remisharrock/sysbuild>.

For simplicity, switch the left panel to ‘File Browser’ mode.

Click on the “<” icon in the upper right hand corner of the terminal window for a simpler interface.

Click on the expansion arrows icon in the upper right of the terminal window to enter full screen mode. When you return from it, you can re-open the terminal window by clicking in the right margin of the window.

To copy text into a text file in the Terminal window, open it in the File Browser window, and then edit it in the Code window. Then click the ‘Save it’ button to save your changes to the text file in the webLinux file system.

[Link to full screen version](#)

D.2 webLinux License

The webLinux object <https://remisharrock.github.io/sysbuild/> used here was released under the following license terms:

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Version 0.0.1

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D.2.1 Acknowledgement

Lawrence Angrave, University of Illinois and UIUC students with their [original project](#).

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