Population Genetics Modeling

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# 1 Homepage

This is the course homepage and digital textbook for Population Genetics Modeling (AS.020.369).

#### 1.0.0.1 Instructor

Andrew Bortvin, abortvi2[at]jhu.edu

#### 1.0.0.2 Schedule & Logistics

| Session | Content |
| --- | --- |
| **Session 1:** | Course Introduction |

# 2 Course Syllabus

**COMPUTATIONAL APPROACHES TO POPULATION GENETICS**  
Instructor: Andrew Bortvin  
Times: Tuesday, Thursday 3:00PM - 4:30PM  
Office Hours: To be determined by when2meet at the start of the semester, or by appointment  
Course website: <https://andrew-bortvin.github.io/popGenModeling/>

### 2.0.1 Course Description

The size, composition, and genetics of populations fluctuate over time. These fluctuations are the product of dynamics between individuals, the interactions between populations, and the context of a population within a broader ecological landscape. The quantitative tools developed to study population genetics allow biologists to discover the simple fundamental principles that govern these complex systems. This course will introduce the basic theory of population genetics while teaching students the fundamental skills of programming in the R programming language, which will allow them to directly implement and visualize theoretical concepts. Students will model and simulate theoretical populations and analyze population-scale genomic data. This course will examine evolution on a variety of scales, ranging from the competition between cells within a single organism, to population dynamics in conservation biology that span decades, to the evolution of contemporary human populations over hundreds of thousands of years.

### 2.0.2 Learning Goals

By the end of this course, students will be able to:

• outline, using biology theory and terms, how populations grow and interact with each other  
• describe the external factors that can cause changes in genetic diversity and use this information to predict how specific demographic scenarios would impact a population  
• manipulate, analyze, and visualize data using the R programming language  
• describe and interpret common formats used to store genomic data, and implement standard analytic protocols used to analyze these data types

### 2.0.3 Grading

30% Participation  
40% Weekly Assignments  
30% Final Project

**Weekly Assignments**

Class sessions will consist of a lecture in which theory is introduced, instructor-led live-coding sessions that implement the models studied in class, and time for students to work independently on assignments that further develop the models designed in class. There will be one assignment per week, assigned each Thursday and due the following Thursday. After submission, students will receive feedback from the instructor on each assignment and will have until the end of the semester to submit any revisions necessary.

Each assignment will consist of a set of required exercises that can be completed by students of any coding background. These will be followed by optional, more computationally-focused exercises, which will allow students to leverage their computational knowledge in creative application to genomic questions.

Work will be graded on reasonable completion–that is to say, code that demonstrates an understanding of an algorithm and its general implementation will receive full marks, regardless of whether output is exactly correct. Students will also be assessed on the clarity and interpretability of the data visualizations that their code outputs and the accuracy of their responses to short questions prompting biological interpretation of their results.

**Googling and AI**

Googling is always an acceptable way to find answers or help, and I encourage you to utilize it extensively. If you adopt a solution following a Google search, make sure you understand what you incorporate, rather than just copy/paste without comprehension of the logic or code. Google is also a good way to learn more about any error messages you encounter in your code.

You may be familiar with ChatGPT and other large language models. After trying each problem/assignment/task on your own, if you’re still running into issues, feel free to use ChatGPT as you would any other online resource (Google, stack overflow, etc.). Learning how to succinctly describe exactly what you want to accomplish is a skillset in itself, so this can be good practice. If you find code that seems to work (e.g., from Google) but you’re not sure how exactly it works, you can also type it into ChatGPT and ask it to explain what’s happening. As always, please do not submit any code if you are not familiar entirely with how it works; flag it and ask an instructor for assistance. Be aware that ChatGPT might confidently offer an answer that is not correct; so always check the output on your own.

### 2.0.4 Schedule

Week 1: Introduction to Python Python Fundamentals: Variables, arithmetic, built-in functions, loops and conditionals, plotting Creating and maintaining a Git repository

Unit 1: Population Biology

Week 2: One Population Models Exponential growth Growth with limited resources: the logistic growth model Age-structured population dynamics

Week 3: Multi-population models Competition, cooperation Predator-Prey Interactions and Lotka-Volterra Dynamics

Week 4: Advanced multi-population models Host-Parasite Interactions and the Nicholson-Bailey model Disease modeling: the SEIR model and community spread

Unit 2: Population Genetics – Theory and Simulation

Week 5: Introduction to Population Genetics The Wright-Fisher model and evolutionarily neutral scenarios

Week 6: Mutation and Natural Selection The Wright-Fisher model with mutation and selection

Week 7: Population Structure and Genetics Population Size Changes Non-random mating

Week 8: Spatial Modeling Migration and local adaptation

Unit 3: Studying Genetic Variation – Data Analysis

Week 9: Quantifying Variation Measures of genome-wide genetic diversity The Site Frequency Spectrum

Week 10: Measuring Selection Tests for selection Local adaptation and adaptive introgression

Week 11: Population Structure Quantification of population structure Measuring admixture and admixture mapping

Week 12: Phylogenetics and Historical Genetics Inferring the timing of selection – coalescent modeling

Week 13: Independent Projects Small-group work sessions In-class presentations

***DATE:*** ALL REVISIONS DUE

# 3 Introduction to R

R is a programming language developed primarily for use by statisticians, and it is a common tool in the field for data exploration, analysis, visualization, etc. We will be using it throughout the course to manipulate, view, and interpret data.

In this module, we will learn the basics of R by manipulating some real-world data. In particular, we’ll look at data from the Hudson Bay looking at the size of the hare and lynx populations.

We’ll talk a lot more about this dataset when we study predator-prey models in a few weeks, but for now, let’s make some exploratory plots.

Data adapted from here: <http://people.whitman.edu/~hundledr/courses/M250F03/M250.html>

Please note that input data is available on the course Posit cloud page - please feel free to work alongside this page!

## 3.1 Variables in R

## 3.2 Data import

The bread and butter of R is the dataframe, a tabular data structure which contains data of multiple types.

R has a few built-in functions that allow for easy and efficient import of data. As we go on, we’ll learn a lot of nuances about how these work, but for now we’ll start with a tidy, well-behaved dataset that is easy to work with.

We would like for R to import our data and save it into memory. We also want to give it a name, so that we can refer to it later.

In R, we use the assignment operator <- to associate a name with some data. The general syntax for this is:

variableName <- value

Where **left - name right -> value**

**if you know python or c or something, “+” is equivalent**

**lets test it out**

populationData <- read.table("hare\_lynx.csv", sep = ",", header = TRUE)

Let’s break down what’s happening here.

On the left of the <-, we have the text populationData

head(populationData)

## Year Species Count  
## 1 1845 Hare 19.58  
## 2 1846 Hare 19.60  
## 3 1847 Hare 19.61  
## 4 1848 Hare 11.99  
## 5 1849 Hare 28.04  
## 6 1850 Hare 58.00

## 3.3 The Data Frame

R has the ability to manipulate multiple different types of data. The primary data types we will use in this course are:

**Numeric** are whole numbers. For example:

## 3.4 Lecture Notes

In this module, we will learn the basics of R by manipulating some real-world data. In particular, we’ll work with data from the Hudson Bay looking at the size of historical hare and lynx populations.

We’ll talk a lot more about this dataset and the biology it represents when we study predator-prey models, but for now let’s use it make some exploratory plots.

Data adapted from here: <http://people.whitman.edu/~hundledr/courses/M250F03/M250.html>

### 3.4.1 Data import

Our data is available as a comma-delimited text file, or csv. Here’s the top of it as it appears in a text editor:



The first thing we have to do to work with it is to import it into R. R has a few built-in functions that allow for easy and efficient import of data. As we go on, we’ll learn a lot of nuances about how these work, but for now we’ll start with a tidy, well-behaved dataset that is easy to work with. The read.table function will be our workhorse for reading in data.

Let’s try it out. To run this function, we need to provide one **argument**: the location of the data we want to load.

read.table("hare\_lynx.csv")

***ADD IMAGE HERE***

A couple things went wrong here:

1. The data is imported as a single column - we want R to know that there are multiple columns delimited by commas
2. The header (Year,Species,Count) was imported as the first row of our data - we would instead like R to recognize that this is a header rather than a row of data

How do we fix this? Along with the essential argument of the file location, we can provide read.table with many **optional arguments**. Because there are many possibilities for what our second argument may be, we need to explicitly reference the name of our optional arguments.

Let’s inform R that our data is split by commas. We can do this using the sep (short for “separator”) argument.

read.table("hare\_lynx.csv", sep = ",")

***ADD IMAGE***

Now we correctly have three columns of data! We now have to tell R that the first row is a header. There is an optional argument header which can be set to TRUE or FALSE (case sensitive - more on this later).

read.table("hare\_lynx.csv", sep = ",", header = TRUE)

***ADD IMAGE***

Looks good! Now, a couple nuances:

1. The order **named arguments** (here, sep and header doesn’t matter. The following lines of code are equivalent:

read.table("hare\_lynx.csv", sep = ",", header = TRUE)  
read.table("hare\_lynx.csv", header = TRUE, sep = ",")

1. For better legibility, we can add line breaks after commas, like so:

read.table("hare\_lynx.csv",   
 sep = ",",   
 header = TRUE)

This does not impact how data is evaluated.

Now that we read in the data correctly, the last thing to do is to save it as a **variable**. This means that rather than just reading in the data and printing it out, we truly save it in the computer’s memory and assign it a name.

The general syntax for saving a variable in R is:

variableName <- value

For example:

hareLynxData <- read.table("hare\_lynx.csv",   
 sep = ",",   
 header = TRUE)

Now, when we refer to hareLynxData, we are referring to the data we have just imported.

For example we can use the head function, which displays the first 10 rows of our data:

head(hareLynxData)

## Year Species Count  
## 1 1845 Hare 19.58  
## 2 1846 Hare 19.60  
## 3 1847 Hare 19.61  
## 4 1848 Hare 11.99  
## 5 1849 Hare 28.04  
## 6 1850 Hare 58.00

### 3.4.2 DataFrames

### 3.4.3 Data Manipulation

### 3.4.4 Subsetting

### 3.4.5 Logical subsetting

library(ggplot2)

# 4 R Guidebook

This section of the website will contain a concise texbook form introduction to the R programming language, tailored to topics covered in class.

## 4.1 Data Types

R allows us to work with many different types of data. The crucial ones for us are:

**Numeric** data are all numbers. For <example:\> • 78  
• 3.2  
• -9.8

**Characters** are any text enclosed by single or double quotation marks. For <example:\> • My Grandpa's deck has no pathetic cards  
• "85.3"

Note that 85.3 - no quotes - is numeric data; "85.3" is character data.

**Logical** data has three possible values: TRUE, FALSE, and NA for missing data. These are case sensitive and lack quotes ("TRUE" would be character data).

**Factors** are characters with associated **levels**, i.e. an order. When we work with numeric data, it is sorted in ascending/descending order. When we work with character data, there is no comparable way to order our data so we can use factors to manually create an order for our data. If this is a bit abstract right now, it’ll make more sense in practice once we work with real data.

## 4.2 Variables

Variables allow us to store data in memory, associated with a name.

We create a variable using the assignment operator <- or =:

variable\_name <- value

For our purposes, these are interchangeable. For example:

• composer <- "buxtehude"  
• year = 1637

The variable name can be almost anything. Here are some general rules to consider when naming a variable:

• Variable names start with a letter and consist of letters, numbers, ., and \_  
• Some words are considered “reserved” - i.e. they are already used by R to mean something and therefore cannot be a variable name. These include TRUE, if, NULL, etc. For a full list, check [here](https://www.datamentor.io/r-programming/reserved-words).

# Authors

| Credits | Names |
| --- | --- |
| **Pedagogy** |  |
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| Content Author | [Andrew Bortvin](https://andrew-bortvin.github.io/) |
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| Template | [Jeff Leek](https://jtleek.com/) & [The Johns Hopkins Data Science Lab](https://jhudatascience.org/index.html) |
| Design Inspiration | [Stephanie Yan](https://stephaniemyan.github.io/) & [Ali Madooei](https://engineering.jhu.edu/faculty/ali-madooei/) & [JHU Data Structures](https://cs226sp22.github.io/) |
| **Funding** |  |
| JHU Dean’s Teaching Fellowship |  |

## ─ Session info ───────────────────────────────────────────────────────────────  
## setting value  
## version R version 4.3.2 (2023-10-31)  
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## system x86\_64, linux-gnu  
## ui X11  
## language (EN)  
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## ctype en\_US.UTF-8  
## tz Etc/UTC  
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## cli 3.6.2 2023-12-11 [1] RSPM (R 4.3.0)  
## colorspace 2.1-0 2023-01-23 [1] RSPM (R 4.3.0)  
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## digest 0.6.34 2024-01-11 [1] RSPM (R 4.3.0)  
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## sessioninfo 1.2.2 2021-12-06 [1] RSPM (R 4.3.0)  
## shiny 1.8.0 2023-11-17 [1] RSPM (R 4.3.0)  
## stringi 1.8.3 2023-12-11 [1] RSPM (R 4.3.0)  
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