

# BIN504 Fall '23 Homework I

<b>Due: December 3 by 23:59</b>
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**Note:** You can submit your answer to the programming problems (2 and 4) as R code only (at least with comments please). But it would be preferable if you created a R Markdown report containing both your code and your discussion of the solution (assumptions, approach etc.). R Markdown was discussed in the lecture; you can get further information about R Markdown at <https://rmarkdown.rstudio.com>. You can also do the non-programming problems (1 and 3) in R Markdown as well, if you wish.

## Problem 1 (25 %)

Tay-Sachs disease is an *autosomal recessive disorder*<sup>1</sup> in children and infants which is fatal. Person A knows that both their parents are carriers of the disease.

- a (5%) What is the probability that person A is a carrier of Tay-Sachs?
- b (10%) Assume that the person is planning to have a child with another person (B), who is known to be a carrier of Tay-Sachs. What is the probability that the child is homozygous for the disease allele?
- c (10%) Let's assume their first child did not have the disease. Update the probability in the first part of the question (that A is a carrier) given this information.

## Problem 2 (25 %)

We will calculate the results for Problem 1, but this time you will be using R and creating a numerical simulation. Please estimate the following probabilities by simulating e.g. 10,000 random outcomes for Person A and their child and use the joint probability table thus created to calculate answers to all parts of the previous question.

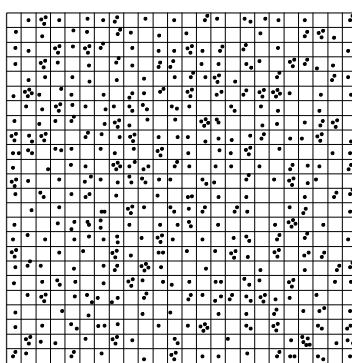
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<sup>1</sup>An autosomal recessive disorder develops only when two copies of a faulty gene are present (i.e. the person is homozygous for the disease allele). Being a carrier for the disease means that a person has only one copy of the disease allele, and can transmit it to offspring, but is not affected by the disease.

### Problem 3 (25 %)

DeMine Inc. is developing a strain of bacteria that glow in the presence of TNT. The idea is to spray spores of this strain on a minefield and detect the location of landmines, so that they can be removed.

In order to test spore viability and effectiveness, they have rented a large square-shaped warehouse  $24m \times 24m$  and have divided the floor into squares that are  $1m$  by  $1m$  each. In each square they spread a nutrient gel, spray the hangar with an aerosol containing the spores, and close up the hangar. After a certain amount of time, they open up the hangar, count the number of bacterial colonies in each square and find the following situation:



After they count, they find that:

- 1 square has 5 colonies,
- 7 squares have 4 colonies,
- 35 squares have 3 colonies,
- 93 squares have 2 colonies,
- 211 squares have 1 colony, and,
- 229 squares have no colonies.

The assumption is that each viable spore in the aerosol has formed a colony. Based on this, answer the following:

- a (10%) If  $\mathbf{X}$  is a random variable representing the number of spore colonies in each square, what do you think is the distribution that governs  $\mathbf{X}$ ?
- b (5%) Using your answer from part (a), find the expected value of  $\mathbf{X}$  (the expected number of spores per  $m^2$ )
- c (10%) The concentration of the aerosol used was  $C$ . Assuming the expected value in part (b) increases linearly with increasing aerosol concentration, what should the minimum concentration be if we want at least a 99.5% chance of there being at least one viable spore in each grid square?

## Problem 4 (25 %)

Suppose the lifetime of a certain kind of fly follows an exponential distribution with unknown  $\lambda$  in days. Some flies have a mutation that shortens this to  $\lambda'$ . A researcher observes  $N$  flies continuously and records the lifetime of each fly as  $y_1, y_2, \dots, y_N$ . You will use the EM algorithm to find an estimate of  $\lambda$  and  $\lambda'$ . The lifetimes recorded are given in the attached file `fly_lifetimes.txt`.

Prepare a program in R to estimate  $\lambda$  and  $\lambda'$  with EM. Your program should also estimate the ratio of mutated flies. Run your program with the data  $y_1, y_2, \dots, y_{120}$  ( $N = 120$ ) given in `fly_lifetimes.txt` and report the results.