Name ______
Period ____
Date ____

LABORATORY 8. POPULATION GENETICS AND EVOLUTION

OVERVIEW

In this laboratory you will learn about the Hardy-Weinberg law of genetic equilibrium and study the relationship between evolution and changes in allele frequency by using your class as a sample population.

OBJECTIVES

At the completion of this laboratory you should be able to

- calculate allele and genotype frequencies using the Hardy-Weinberg theorem
- discuss the effect of natural selection on allelic frequencies
- explain and predict the effect on allelic frequencies of selection against the homozygous recessive
- discuss the relationship between evolution and changes in allele frequencies, as measured by deviations from the Hardy-Weinberg law of genetic equilibrium

INTRODUCTION

In 1908 G. H. Hardy and W. Weinberg independently suggested a scheme whereby evolution could be viewed as changes in the frequency of alleles in a population of organisms. They reasoned that if *A* and *a* are alleles for a particular gene locus and each diploid individual has two such loci, then p can be designated as the frequency of the *A* allele and q as the frequency of the *a* allele. Thus, in a population of 100 individuals (each with two loci) in which 40% of the loci are *A*, p would be 0.40. The rest of the loci (60%) would be *a*, and q would equal 0.60 (i.e., $p + q = 1.0$). These are referred to as **allele frequencies.** If certain conditions are met, the frequency of the possible diploid combinations of these alleles (*AA*, *Aa*, *aa*) should equal $p^2 + 2pq + q^2 = 1.0$. Hardy and Weinberg also argued that if five conditions are met, the population's allele and genotype frequencies will remain constant from generation to generation. These conditions are as follows:

1. The population is very large. The effects of chance on changes in allele frequencies is thereby greatly reduced.
2. Individuals show no mating preference for *A* or *a*, i.e., mating is random.
3. There is no mutation of the alleles.
4. No differential migration occurs (no immigration or emigration).
5. All genotypes have an equal chance of surviving and reproducing, i.e., there is no selection.

Basically, the Hardy-Weinberg equation describes the status quo. If the five conditions are met, then no change will occur in either allele or genotype frequencies in the population. Of what value is such a rule? It provides a yardstick by which changes in allele frequency, and therefore evolution, can be measured. One can look at a population and ask: Is evolution occurring with respect to a particular gene locus?

The purpose of this laboratory is to examine the conditions necessary for maintaining the status quo and see how selection changes allele frequency. This will be done by simulating the evolutionary process using your class as a sample population.

EXERCISE 8A: Estimating Allele Frequencies for a Specific Trait within a Sample Population

Using the class as a sample population, the allele frequency of a gene controlling the ability to taste the chemical PTC (phenylthiocarbamide) could be estimated. A bitter-taste reaction to PTC is evidence of the presence of a *dominant allele* in either the homozygous condition (*AA*) or the heterozygous condition (*Aa*). The inability to taste the chemical at all depends on the presence of homozygous recessive alleles (*aa*). (Instead of PTC tasting, other traits, such as tongue-rolling, may be used.) To estimate the frequency of the PTC-tasting allele in the population, one must find p. To find p, one must first determine q (the frequency of the nontasting PTC allele), because only the genotype of the homozygous recessive individuals is known for sure (i.e., those that show the dominant trait could be *AA* or *Aa*).

Procedure

1. Using the PTC taste-test papers provided, tear off a short strip and press it to your tongue tip. PTC tasters will sense a bitter taste. For the purposes of this exercise these individuals are considered to be tasters.
2. A decimal number representing the frequency of tasters ($p^2 + 2pq$) should be calculated by dividing the number of tasters in the class by the total number of students in the class. A decimal number representing the frequency of nontasters (q^2) can be obtained by dividing the number of nontasters by the total number of students. You should then record these numbers in Table 8.1.
3. Use the Hardy-Weinberg equation to determine the frequencies (p and q) of the two alleles. The frequency q can be calculated by taking the square root of q^2. Once q has been determined, p can be determined because $1 - q = p$. Record these values in Table 8.1 for the class and also calculate and record values of p and q for the North American population.

Table 8.1: Phenotypic Proportions of Tasters and Nontasters and Frequencies of the Determining Alleles

	Phenotypes		Allele Frequency Based on the H-W Equation	
	% Tasters ($p^2 + 2pq$)	% Nontasters (q^2)	p	q
Class Population				
North American Population	0.55	0.45		

Topics for Discussion

1. What is the percentage of heterozygous tasters ($2pq$) in your class?

___

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2. What percentage of the North American population is heterozygous for the taster trait?

___

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Topics for Discussion

1. What is the percentage of heterozygous tasters ($2pq$) in your class?
2. What percentage of the North American population is heterozygous for the taster trait?

EXERCISE 8B: Case Studies

CASE I (A Test of an Ideal Hardy-Weinberg Population)

The entire class will represent a breeding population, so find a large open space for this simulation. In order to ensure random mating, choose another student at random. In this simulation, we will assume that gender and genotype are irrelevant to mate selection.

The class will simulate a population of randomly mating heterozygous individuals with an intital gene frequency of 0.5 for the dominant allele A and the recessive allele **a** and genotype frequencies of 0.25AA, 0.50 **Aa**, and 0.25**aa**. Your initial genotype is **Aa**. Record this on the data page (page 98). Each member of the class will receive four cards. Two cards will have A and two cards will have **a**. The four cards represent the products of meiosis. Each "parent" contributes a haploid set of chromosomes to the next generation.

Procedure

1. Turn the four cards over so the letters are not showing, shuffle them, and take the card on top to contribute to the production of the first offspring. Your partner should do the same. Put the two cards together. The two cards represent the alleles of the first offspring. One of you should record the genotype of this offspring in the Case I section on page 98. Each student pair must produce two offspring, so all four cards must be reshuffled and the process repeated to produce a second offspring.

2. The other partner should then record the genotype of the second offspring on page 98. The very short reproductive career of this generation is over. You and your partner now become the next generation by assuming the genotypes of the *two offspring*. That is, student 1 assumes the genotype of the first offspring and student 2 assumes the genotype of the second offspring.

3. Each student should obtain, if necessary, new cards representing the alleles in his or her respective gametes after the process of meiosis. For example, student 1 becomes genotype Aa and obtains cards *A*, *A*, *a*, *a*; student 2 becomes *aa* and obtains cards *a*, *a*, *a*, *a*. Each participant should **randomly** seek out another person with whom to mate in order to produce the offspring of the next generation. Remember, the sex of your mate does not matter, nor does the genotype. You should follow the same mating procedures as for the first generation, being sure to record your new genotype after each generation in the table. Class data should be collected after each generation for five generations. At the end of each generation, remember to record the genotype that you have assumed. Your teacher will collect class data after each generation by asking you to raise your hand to report your genotype.

Allele Frequency: The allele frequencies, p and q, should be calculated for the population after five generations of simulated random mating.

Number of *A* alleles present at the fifth generation

Number of offspring with genotype *AA* __________ × 2 = ________________ *A* alleles

Number of offspring with genotype *Aa* __________ × 1 = ________________ *A* alleles

Total = ________________ *A* alleles

$$p = \frac{\text{TOTAL number of } A \text{ alleles}}{\text{TOTAL number of alleles in the population}} = \_\_\_\_\_\_\_\_$$

In this case, the total number of alleles in the population is equal to the number of students in the class × 2.

Number of *a* alleles present at the fifth generation

Number of offspring with genotype *aa* ____________ × 2 = ________________ *a* alleles

Number of offspring with genotype *Aa* ____________ × 1 = ________________ *a* alleles

Total = ________________ *a* alleles

$$q = \frac{\text{TOTAL number of } a \text{ alleles}}{\text{TOTAL number of alleles in the population}} = \_\_\_\_\_\_\_\_$$

1. What does the Hardy-Weinberg equation predict for the new p and q?

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2. Do the results you obtained in this simulation agree? ____________ If not, why not?

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3. What major assumption(s) were not strictly followed in this simulation?

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CASE II (Selection)

In this Case, you will modify the simulation to make it more realistic. In the natural environment, not all genotypes have the same rate of survival; that is, the environment might favor some genotypes while selecting against others. An example is the human condition, sickle-cell anemia. It is a disease caused by a mutation on one allele, in which individuals who are homozygous recessive often do not survive to reach reproductive maturity. For this simulation, you will assume that the homozygous recessive individuals never survive (100 percent selection against), and that heterozygous and homozygous dominant individuals survive 100 percent of the time.

The procedure is similar to that for Case I. Start again with your initial genotype, and produce your "offspring" as in Case I. This time, however, there is one important difference. Every time your "offspring" is *aa*, it does not reproduce. Since we want to maintain a constant population size, the same two parents must try again until they produce two surviving offspring. You may need to get new "allele" cards from the pool.

Proceed through five generations, selecting against the homozygous recessive offspring 100 percent of the time. Then add up the genotype frequencies that exist in the population and calculate the new p and q frequencies in the same way as it was done in Case I.

1. How do the new frequencies of p and q compare to the initial frequencies in Case I?

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2. How has the allelic frequency of the population changed?

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3. Predict what would happen to the frequencies of p and q if you simulated another five generations.

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4. In a large population, would it be possible to completely eliminate a deleterious recessive allele? Explain.

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CASE III (Heterozygote Advantage)

From Case II, it is easy to see that the lethal recessive allele rapidly decreases in the population. However, data from many human populations show an unexpectedly high frequency of the sickle-cell allele in some populations. In other words, our simulation does not accurately reflect the real situation; this is because individuals who are heterozygous are slightly more resistant to a deadly form of malaria than homozygous dominant individuals. In other words, there is a slight selection against homozygous dominant individuals as compared to heterozygotes. This fact is easily incorporated into our simulation. In this round, keep everything the same as in Case II, except that if your offspring is *AA*, flip a coin. If heads, the individual does not survive, and if tails the individual does survive.

Simulate five generations, starting again with the initial genotype from Case I. The genotype *aa* never survives, and homozygous dominant individuals only survive if the coin toss comes up tails. Total the class genotypes and calculate the p and q frequencies. Starting with the F_5 genotype, go through five more generations, and again total the genotypes and calculate the frequencies of p and q. If time permits, the results from another five generations would be extremely informative.

1. Explain how the changes in p and q frequencies in Case II compare with Case I and Case III.

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2. Do you think the recessive allele will be completely eliminated in either Case II or Case III?

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3. What is the importance of heterozygotes (the heterozygote advantage) in maintaining genetic variation in populations?

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Data Page

CASE I
Hardy-Weinberg Equilibrium

Initial Class Frequencies:

AA ____ Aa____ aa ____

My Initial Genotype: ____

F_1 Genotype ____

F_2 Genotype ____

F_3 Genotype ____

F_4 Genotype ____

F_5 Genotype ____

Final Class Frequencies:

AA ____ Aa____ aa ____

p____ q ____

CASE II
Selection

Initial Class Frequencies:

AA____ Aa____ aa _____

My Initial Genotype: ____

F_1 Genotype ____

F_2 Genotype ____

F_3 Genotype ____

F_4 Genotype ____

F_5 Genotype ____

Final Class Frequencies:

AA____ Aa____ aa____

p____ q ____

CASE III
Heterozygote Advantage

Initial Class Frequencies:

AA____ Aa____ aa _____

My Initial Genotype: ____

F_1 Genotype ____ F_6 Genotype ____

F_2 Genotype ____ F_7 Genotype ____

F_3 Genotype ____ F_8 Genotype ____

F_4 Genotype ____ F_9 Genotype ____

F_5 Genotype ____ F_{10} Genotype ____

Final Class Frequencies: (after five generations)

AA____ Aa____ aa _____

p____ q ____

Final Class Frequencies: (after ten generations)

AA____ Aa____ aa _____

p____ q ____

CASE IV
Genetic Drift

Initial Class Frequencies:

AA____ Aa____ aa _____

p____ q ____

My Initial Genotype: ____

F_1 Genotype ____

F_2 Genotype ____

F_3 Genotype ____

F_4 Genotype ____

F_5 Genotype ____

Final Class Frequencies:

AA____ Aa____ aa _____

p____ q ____