

LAB EIGHT POPULATION GENETICS AND EVOLUTION

OVERVIEW

In this lab you will:

1. learn about the Hardy-Weinberg law of genetic equilibrium, and
2. study the relationship between evolution and changes in allele frequency by using your class to represent a sample population.

OBJECTIVES

Before doing this lab you should understand:

- how natural selection can alter allelic frequencies in a population;
- the Hardy-Weinberg equation and its use in determining the frequency of alleles in a population; and
- the effects on allelic frequencies of selection against the homozygous recessive or other genotypes.

After doing this lab you should be able to:

- calculate the frequencies of alleles and genotypes in the gene pool of a population using the Hardy-Weinberg formula, and
- discuss natural selection and other causes of microevolution as deviations from the conditions required to maintain Hardy-Weinberg 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. In this scheme, 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 alleles are A , p would be 0.40. The rest of the alleles (60%) would be a , and q would equal 0.60 (i.e., $p + q = 1.0$). These are referred to as **allele frequencies**. The frequency of the possible diploid combinations of these alleles (AA , Aa , aa) is expressed as $p^2 + 2pq + q^2 = 1.0$. 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 breeding population is large.** (The effect of chance on changes in allele frequencies is thereby greatly reduced.)
2. **Mating is random.** (Individuals show no mating preference for a particular phenotype.)
3. **There is no mutation of the alleles.** (No alteration in the DNA sequence of alleles.)
4. **No differential migration occurs.** (No immigration or emigration.)
5. **There is no selection.** (All genotypes have an equal chance of surviving and reproducing.)

The Hardy-Weinberg equation describes an existing situation. 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? Since evolution is difficult (if not impossible) to observe in most natural populations, we will model the evolutionary process using the class as a simulated population. The purpose of this simulation is to provide an opportunity to test some of the basic tenets of population genetics and evolutionary biology.

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 attached earlobes, 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).

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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)		<i>p</i>	<i>q</i>
Class Population	#	%	#	%		
North American Population	0.55		0.45			

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.

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The class will simulate a population of randomly mating heterozygous individuals with an initial gene frequency of 0.5 for the dominant allele A and the recessive allele a and genotype frequencies of 0.25 AA , 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 written on them 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 that the letters do not show, 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 the Data Page. 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 the Data Page. 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 you did for the first generation, being sure to record your new genotype after each generation. 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.

* For simplicity, in this model all individuals in the initial population have a genotype of Aa .

4. **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 _____ $\times 2 =$ _____ A alleles

Number of offspring with genotype Aa _____ $\times 1 =$ _____ A alleles

Total = _____ A alleles

$$p = \frac{\text{TOTAL number of A alleles}}{\text{TOTAL number of alleles in the population (number of students } \times 2)} = \underline{\hspace{2cm}}$$

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

Number of a alleles present at the fifth generation

Number of offspring with genotype aa _____ $\times 2 =$ _____ a alleles

Number of offspring with genotype Aa _____ $\times 1 =$ _____ a alleles

Total = _____ a alleles

$$q = \frac{\text{TOTAL number of a alleles}}{\text{TOTAL number of alleles in the population (number of students } \times 2)} = \underline{\hspace{2cm}}$$

Questions

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

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

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 of sickle-cell anemia. This is a disease caused by a mutation on one allele, and 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% selection against), and that heterozygous and homozygous dominant individuals survive 100% of the time.

Procedure

The procedure is similar to that for Case I.

1. Start again with your initial genotype and produce your "offspring" as you did for 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, allowing each individual to complete the activity.
2. Proceed through five generations, selecting against the homozygous recessive offspring 100% 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 you did for Case I.

Questions

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

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

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

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 what happens to the lethal recessive allele in the population. However, data from many human populations show an unexpectedly high frequency of the sickle-cell allele in some populations. Thus, 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.

Procedure

1. In this round keep everything the same as it was in Case II, except that if your offspring is AA, flip a coin. If the coin lands heads up, the individual does not survive; if tails, the individual does survive.
2. 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. Since we want to maintain a constant population size, the same two parents must try again until they produce two surviving offspring. Get new "allele" cards from the pool as needed. Total the class genotypes and calculate the *p* and *q* frequencies.
3. Starting with the F₅ genotype, go through five more generations, and again total the genotypes and calculate the frequencies of *p* and *q*.
4. If time permits, the results from another five generations would be extremely informative.

Questions

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

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

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

Data Page

**CASE I
Hardy-Weinberg Equilibrium**

Initial Class Frequencies:

AA ____ Aa ____ aa ____

My Initial Genotype: ____

F₁ Genotype ____

F₂ Genotype ____

F₃ Genotype ____

F₄ Genotype ____

F₅ Genotype ____

Final Class Frequencies:

AA ____ Aa ____ aa ____

p ____ q ____

**CASE II
Selection**

Initial Class Frequencies:

AA ____ Aa ____ aa ____

My Initial Genotype: ____

F₁ Genotype ____

F₂ Genotype ____

F₃ Genotype ____

F₄ Genotype ____

F₅ Genotype ____

Final Class Frequencies:

AA ____ Aa ____ aa ____

p ____ q ____

**CASE III
Heterozygote Advantage**

Initial Class Frequencies:

AA ____ Aa ____ aa ____

My Initial Genotype: ____

F₁ Genotype ____ F₆ Genotype ____

F₂ Genotype ____ F₇ Genotype ____

F₃ Genotype ____ F₈ Genotype ____

F₄ Genotype ____ F₉ Genotype ____

F₅ Genotype ____ F₁₀ 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: ____

F₁ Genotype ____

F₂ Genotype ____

F₃ Genotype ____

F₄ Genotype ____

F₅ Genotype ____

Final Class Frequencies:

AA ____ Aa ____ aa ____

p ____ q ____