

LAB #17: Population Genetics and Evolution

Objective:

- In this investigation, students will be able to demonstrate how natural selection can alter allelic frequencies in a population and use the Hardy-Weinberg equation to calculate the genotypes of a population.

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 would 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 $p^2 + 2pq + q^2 = 1$. Hardy and Weinberg also argues 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 sequences 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.

Part I: Estimating Allele Frequencies for a Specific Trait

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). 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).

- 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.
- 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 1.1**.
- 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 1** for the class and also calculate and record values of p and q for the North American population.

Table 1

	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			

- Using the above information, what is the percentage of heterozygous tasters ($2pq$) in your class?
- What percentage of the North American population is heterozygous for the taster trait?

Part II: 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 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* (set for simplicity in this particular case). Record this in **Table 2a**. 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.

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 your **Table 2a**. 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 in their **Table 2a**. 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 becomes 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 in your **Table 2a**. 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. After each generation, your teacher will collect data by asking you to raise your hand to report your genotype. These will be included in **Table 2b**.

Table 2a

Generation	Your Genotype
Initial	Aa
F ₁	
F ₂	
F ₃	
F ₄	
F ₅	

Table 2b

Generation	Class Totals for Each Genotype		
	AA	Aa	aa
F ₁			
F ₂			
F ₃			
F ₄			
F ₅			

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

Final 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 _____ x 2 = _____ A alleles

Number of offspring with genotype Aa _____ x 1 = _____ A alleles

Total = _____ A alleles

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

Number of a alleles present at the fifth generation

Number of offspring with genotype aa _____ x 2 = _____ a alleles

Number of offspring with genotype Aa _____ x 1 = _____ a alleles

Total = _____ a alleles

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

- Do the results you obtained in the simulation agree? (Probably not) Explain why—that is, what major assumption(s) were not strictly followed in this simulation.

CASE II – Selection

In this case study, 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.

The following 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. Enter all data in **Tables 3a and 3b**. 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 that you did for Case I.

Table 3a

Generation	Your Genotype
Initial	Aa
F ₁	
F ₂	
F ₃	
F ₄	
F ₅	

Table 3b

Generation	Class Totals for Each Genotype		
	AA	Aa	aa
F ₁			
F ₂			
F ₃			
F ₄			
F ₅			

Final 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 _____ x 2 = _____ A alleles

Number of offspring with genotype Aa _____ x 1 = _____ A alleles

Total = _____ A alleles

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

Number of a alleles present at the fifth generation

Number of offspring with genotype aa _____ x 2 = _____ a alleles

Number of offspring with genotype Aa _____ x 1 = _____ a alleles

Total = _____ a alleles

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

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

- Besides the small population of the class, what major assumption was not strictly followed in this simulation?

- In a large population following this case study, would it be possible to completely eliminate a deleterious recessive allele? Explain your answer.

CASE III – Heterozygous 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 sickly-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. This fact is easily incorporated into our simulation.

The following procedure is similar to that for Case I

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. Enter all data in **Tables 4a** and **4b**.
3. Starting with the F_5 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.

Table 4a

Generation	Your Genotype
Initial	Aa
F ₁	
F ₂	
F ₃	
F ₄	
F ₅	
F ₆	
F ₇	
F ₈	
F ₉	
F ₁₀	

Table 4b

Generation	Class Totals for Each Genotype		
	AA	Aa	aa
F ₁			
F ₂			
F ₃			
F ₄			
F ₅			
F ₆			
F ₇			
F ₈			
F ₉			
F ₁₀			

Allele Frequency (after five generations): 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 _____ x 2 = _____ A alleles

Number of offspring with genotype Aa _____ x 1 = _____ A alleles

Total = _____ A alleles

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

Number of a alleles present at the fifth generation

Number of offspring with genotype aa _____ x 2 = _____ a alleles

Number of offspring with genotype Aa _____ x 1 = _____ a alleles

Total = _____ a alleles

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

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

Number of A alleles present at the tenth generation

Number of offspring with genotype AA _____ x 2 = _____ A alleles

Number of offspring with genotype Aa _____ x 1 = _____ A alleles

Total = _____ A alleles

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

Number of a alleles present at the tenth generation

Number of offspring with genotype aa _____ x 2 = _____ a alleles

Number of offspring with genotype Aa _____ x 1 = _____ a alleles

Total = _____ a alleles

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

Name: _____

- Do you think the recessive allele will be completely eliminated in either Case II? Why or why not?

- Do you think the recessive allele will be completely eliminated in either Case III? Why or why not?

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

CASE IV – Genetic Drift (OPTIONAL—TBD by teacher)

It is possible to use our simulation to look at the phenomenon of genetic drift in detail.

1. Divide the class into several smaller “populations” so that individuals from one isolated “population” do not interact with individuals from another population
2. Now go through five generations as you did for Case I. Record all data in **Tables 5a and 5b**. Then calculate the new genotypic and allelic frequencies for your population.
3. Compare your data with the other “population” data.

Table 5a

Generation	Your Genotype
Initial	Aa
F ₁	
F ₂	
F ₃	
F ₄	
F ₅	

Table 5b

Generation	Sub-Population Totals for Each Genotype		
	AA	Aa	aa
F ₁			
F ₂			
F ₃			
F ₄			
F ₅			

Final 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 _____ x 2 = _____ A alleles

Number of offspring with genotype Aa _____ x 1 = _____ A alleles

Total = _____ A alleles

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

Number of a alleles present at the fifth generation

Number of offspring with genotype aa _____ x 2 = _____ a alleles

Number of offspring with genotype Aa _____ x 1 = _____ a alleles

Total = _____ a alleles

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

Name: _____

- How do the final genotypic frequencies of each population compare. Why are they different? (They should be—if not, we need to talk about it in class!)

- What do the class results indicate about the importance of population size as an evolutionary force?

Part III: Hardy-Weinberg Problems

1. The allele for the hair pattern called "widow's peak" is dominant over the allele for no "widow's peak". In a population of 1,000 individuals, 510 show the dominant phenotype. How many individuals would you expect of each of the possible three genotypes for this trait?
2. In the United States about 16% of the population is Rh negative. The allele for Rh negative is recessive to the allele for Rh positive. If the student population of a high school in the U.S. is 2,000, how many students would you expect for each of these three possible genotypes?
3. In certain African countries 4% of the newborn babies have sickle cell anemia, which is a recessive trait. Out of a random population of 1,000 newborn babies, how many would you expect for each of the three possible genotypes?
4. In a certain population, the dominant phenotype of a certain trait occurs 91% of the time. What is the frequency of the dominant allele?