

# Population Genetics and Evolution

As early as the 500s B.C., several Greek philosophers theorized about the union of male and female traits to form offspring. In the 17th century, Leeuwenhoek concluded that semen and eggs carried hereditary factors conveyed to the offspring. Throughout the next century, scientists developed theories on the processes of development; LaMarck was one of the first to discuss the possibility of acquiring changed traits from parents. For example, he thought that if giraffes had to stretch to eat the tops of trees, their offspring would be born with longer necks.

During the 19th century, Darwin published his theory of evolution, stating that members of a population vary considerably in their genetic makeup. Those that are the “fittest” for their environment are better able to survive and reproduce, and therefore pass these suitable traits on to the next generation. This “natural selection” creates a population that is different from the previous generations. Since Darwin’s theories were published, several others have expounded on his work, leading to the ideas of adaptation and mutation. Recent research has determined that chromosomes, present in each sex’s reproductive material, carry the genes that determine individual characteristics.

A population—all the individuals of a species that live in the same place at the same time—are affected by their own characteristics. Population genetics is the effect that heredity has on a population. What happens to that group of people over the course of a number of generations is expressed mathematically.

There are three key elements of any population: size, density, and dispersion. Population size is important to the groups’ ability to reproduce without a lot of inbreeding. Inbreeding can be the downfall of a population if recessive traits, many of which are harmful, become a common occurrence. Population density can affect the ability of individuals to reproduce, based on whether they ever encounter another to mate with. Dispersion, or how populations are arranged, can also affect populations.

Populations evolve by responding to their surroundings through natural selection. This change actually occurs in the frequency of gene alleles in the population. William Castle, an American scientist; Geoffrey Hardy, a British mathematician; and Wilhelm Weinberg, a German physician, independently determined that the frequencies of genes in a population remain constant unless certain forces act on the population. Dominant alleles will not replace recessive alleles, and the ratio of heterozygous and homozygous individuals does not change over the course of several generations. This theory has come to be known as the Hardy–Weinberg principle; it is the basis of the study of population genetics.

The Hardy–Weinberg principle is normally stated as a mathematical equation:

$$p^2 + 2pq + q^2 = 1$$

The frequencies of the dominant and recessive alleles are represented by  $p$  and  $q$ , respectively. For example, if a diploid individual has two alleles, “A” and “a”, at a particular locus, only three possible genotypes can be the result: AA, Aa, and aa. The probability of receiving the “A” alleles from both parents is  $p \times p$ , or  $p^2$ ; for the “a” alleles,  $q \times q$ , or  $q^2$ . Those who received the Aa combination are described by  $2pq$ , since it is possible for the “A” or the “a” to come from either parent, thereby doubling the chance.

To apply the principle, at least one of the allele frequencies must be known. For example, if the frequency of the recessive allele for cystic fibrosis is one in 2,080 Caucasian North Americans, or 0.00048, this is equal to  $q^2$ . After calculating the square root,  $q = 0.022$ . Then the frequency of the dominant allele is calculated: since  $p + q = 1$ ,  $p = 1 - q$  or  $p = 1 - 0.022$ ,  $p = 0.978$ .

Having identified the values of  $p$  and  $q$ , the frequency of heterozygotes in the population can be determined:  $2pq = 2(0.978)(0.022) = 0.043$ . Therefore, 43 out of every 1,000 Caucasian North Americans are heterozygous for cystic fibrosis.

If the relationship between  $p$  and  $q$  are constant through randomly mating generations, the population is said to be in Hardy–Weinberg equilibrium; no evolution occurs. However, five evolutionary forces act on a population to affect it: mutation, migration, non-random mating, genetic drift, and natural selection. If any of these conditions are present, the proportions of heterozygotes and homozygotes can differ. Therefore, the Hardy–Weinberg principle is a useful tool for measuring the degree of genetic change or evolution occurring in a population.

Mutation is a fairly uncommon occurrence, which will not significantly change allele frequencies itself, but is the main source of change and therefore evolution. Migration of individuals in or out of a population is the cause of gene flow, which introduces new alleles to a group and removes others. Non-random mating can be the result of either inbreeding or an organism's ability to choose a mate based on certain characteristics such as size, coloration, or lifestyle. In either case, the proportion of homozygotes can increase, upsetting the equilibrium. Genetic drift refers to the possibility that by chance, certain alleles could be eliminated from a population. For example, two heterozygous parents,  $Aa$ , could possibly have two children, both  $AA$ , thus eliminating the  $aa$  allele from the next generation. Normally this is balanced by similar events that might eliminate the  $A$  allele from another family, but in small populations it is possible to lose the same allele at a large number of loci, causing evolution to take place. Natural selection is the predominant force in creating evolutionary change. An allele can increase or decrease in frequency based on its effect on reproduction and survival. This can change from one generation to the next, and from one area to another, since a trait that is harmful to its carrier in one circumstance could be beneficial in another.

## OBJECTIVES

In this experiment, you will

- Investigate a genetically inherited trait and apply the Hardy-Weinberg Principle to a population.
- Calculate allele frequencies and genotypes for a population using the Hardy-Weinberg formula.
- Compare allele frequencies within the classroom to North American averages.
- Demonstrate the stability of allele frequencies over five generations in an ideal Hardy-Weinberg population.
- Examine the effects of natural selection, heterozygous advantage, and genetic drift on allele frequencies in a simulated mating exercise.

## MATERIALS

PTC paper  
control paper  
4 index cards  
coin

## **PROCEDURE**

### **Part I Calculating Allele Frequencies Using the Hardy–Weinberg Principle**

Testing different individuals' ability to taste PTC (phenylthiocarbamide) is a good way to demonstrate the Hardy–Weinberg principle. Homozygous-dominant (AA) and heterozygous (Aa) individuals can taste this bitter chemical, although homozygous-recessive (aa) individuals cannot. Use your class as a representative population to calculate the frequencies of the two alleles with the Hardy–Weinberg equation

1. Obtain a piece of PTC test paper. **Note:** Use each strip of PTC and control test paper only once. Do not share test papers with other students in your class.
2. Place it on your tongue and note whether you can detect a bitter taste. **Note:** Used test papers may be disposed of with general waste.
3. Obtain a piece of control paper and place it on your tongue. Comparing the control paper with the PTC paper will help determine whether you detected a taste on the PTC paper.
4. Record your results in Table 1 in the Analysis section.
5. Fill in the results for each individual in the class and enter the results in Table 2.
6. Tally the results for the entire class and calculate the frequencies for each allele, using the Hardy-Weinberg equation, in the Analysis section. Be sure to show your work.

### **Part II Testing the Hardy–Weinberg Principle**

#### **Case A: Testing an Ideal Population**

1. Obtain four index cards. Label two “A” and two “a”. These will be your haploid chromosomes.
2. Randomly pair off with another student for “breeding”. Choose any other student; your gender, and your partner’s, doesn’t matter in the simulation.
3. Turn your cards upside down and shuffle them. Turn over the top card in your pile. Pair this card with your partner’s card; this will be the genotype of your first offspring. Record this on your data sheet.
4. Turn over the next card in your pile. Pair this card with your partner’s card; this will be the genotype of your second offspring. Record this on your data sheet.
5. Now begin the second generation, taking the genotype of your offspring. For example, if you produced offspring with the genotype AA, begin the next generation with four A cards. Record the results in the Analysis section of the lab.
6. Repeat the above procedure for the second generation. Record the genotypes in the Analysis section.
7. Repeat the procedure for three more generations, for a total of five generations. Record the genotypes for each.

8. Combine the genotype of your fifth generation results with the rest of the students' fifth generation results and enter the totals in the Analysis section.
9. Using the table in the Analysis section, calculate the allele frequencies after five generations of random mating.

**Case B: Selection**

The previous exercise was conducted with ideal parameters. For a more realistic situation, selection must be used. There is 100% selection against homozygous-recessive offspring. If offspring are recessive (if they receive two mutated alleles), they will never live long enough to reach a reproductive age; offspring that are either heterozygous or homozygous dominant will survive long enough to reproduce.

10. Follow the same procedure as the previous exercise, with one difference: If offspring is produced with the genotype aa, this offspring will not survive; eliminate the alleles from the population. To maintain population size, you must produce two surviving offspring. If two alleles are eliminated, draw two new alleles from the extra cards.
11. Repeat the procedure for a total of five generations, selecting against homozygous-recessive offspring in each generation. Record the genotypes after every generation in the Analysis section.
12. Combine your fifth generation results with the rest of the students' fifth generation results and record in the Analysis section.
13. Using the table in the Analysis section, calculate the allele frequencies after five generations of random mating.

**Case C: Heterozygote Advantage**

The previous exercise showed how selection against homozygous-recessive individuals clearly alters the allelic frequencies in a population. Another form of selection that operates within a gene pool is diseases, such as a deadly form of malaria, that affect homozygous-dominant individuals more severely than heterozygous individuals. The heterozygote is therefore favored in a population.

14. Follow the same procedure, eliminating homozygous-recessive individuals as before. In addition, if a homozygous-dominant individual is produced, flip a coin. If the result is heads, the offspring dies; if it is tails, the offspring survives.
15. Repeat the procedure for a total of five generations. Record the genotypes after every generation in the Analysis section.
16. Combine your fifth generation results with the rest of the students' fifth generation results and record in the Analysis section.
17. Continue the procedure for five more generations, for a total of ten generations, this time starting with the genotypes from the end of the fifth generation. Record the genotypes in the Analysis section.
18. Using the table in the Analysis section, calculate the allele frequencies after ten generations of random mating.

Part II Testing the Hardy-Weinberg Principle

Case A

Testing an Ideal Population

Initial Class Frequencies: AA: 1 Aa: 2 aa: 1  
 My Initial Genotype: aa AA  
 F<sub>1</sub> Genotype: Aa  
 F<sub>2</sub> Genotype: Aa  
 F<sub>3</sub> Genotype: \_\_\_\_\_  
 F<sub>4</sub> Genotype: \_\_\_\_\_  
 F<sub>5</sub> Genotype: \_\_\_\_\_  
 Final Class Frequencies: AA: 4 13 Aa: 7 39 aa: 1 16  
 p: .711 .68 q: .288 .32  
.5055 .4095 .0784 = .9934

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

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

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

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

**Case D: Genetic Drift**

Genetic drift is a phenomenon where an allele is lost solely from chance instead of through selection. The most important factor in genetic drift is population size; smaller populations have a much greater potential for genetic drift.

19. Your instructor will divide the class into several smaller populations. Within your smaller population, follow the mating procedure, as in the first exercise, for a total of five generations. Record the genotypes after every generation in the Analysis section.
20. Combine your group's fifth generation results with those of the other small populations and calculate the new allele frequencies.

**ANALYSIS**

**Part I Calculating Allele Frequencies Using the Hardy-Weinberg Principle**

Table 1		
	Taster	Nontaster
PTC	23 17	10 9
Control		

Table 2						
	Phenotypes				Allele frequency based on the Hardy-Weinberg equation	
	Tasters ( $p^2+2pq$ )		Nontasters ( $q^2$ )		p	q
Class population	#	%	#	%		
		23 17	70 65	10 9	30 35	.45 .41

$$.20 + .50 + .30 = 1.00$$

**Case B  
Selection**

Initial Class Frequencies: AA: \_\_\_\_\_ Aa: \_\_\_\_\_ aa: \_\_\_\_\_  
My Initial Genotype: \_\_\_\_\_  
F<sub>1</sub> Genotype: \_\_\_\_\_  
F<sub>2</sub> Genotype: \_\_\_\_\_  
F<sub>3</sub> Genotype: \_\_\_\_\_  
F<sub>4</sub> Genotype: \_\_\_\_\_  
F<sub>5</sub> Genotype: \_\_\_\_\_  
Final Class Frequencies: AA: \_\_\_\_\_ Aa: \_\_\_\_\_ aa: \_\_\_\_\_  
p: \_\_\_\_\_ q: \_\_\_\_\_

**Case C  
Heterozygote Advantage**

Initial Class Frequencies: AA: \_\_\_\_\_ Aa: \_\_\_\_\_ aa: \_\_\_\_\_  
My Initial Genotype: \_\_\_\_\_  
F<sub>1</sub> Genotype: \_\_\_\_\_  
F<sub>2</sub> Genotype: \_\_\_\_\_  
F<sub>3</sub> Genotype: \_\_\_\_\_  
F<sub>4</sub> Genotype: \_\_\_\_\_  
F<sub>5</sub> Genotype: \_\_\_\_\_  
Final Class Frequencies:  
(After five generations) AA: \_\_\_\_\_ Aa: \_\_\_\_\_ aa: \_\_\_\_\_  
p: \_\_\_\_\_ q: \_\_\_\_\_  
F<sub>6</sub> Genotype: \_\_\_\_\_  
F<sub>7</sub> Genotype: \_\_\_\_\_  
F<sub>8</sub> Genotype: \_\_\_\_\_  
F<sub>9</sub> Genotype: \_\_\_\_\_  
F<sub>10</sub> Genotype: \_\_\_\_\_  
Final Class Frequencies:  
(After ten generations) AA: \_\_\_\_\_ Aa: \_\_\_\_\_ aa: \_\_\_\_\_  
p: \_\_\_\_\_ q: \_\_\_\_\_

**Case D  
Genetic Drift**

Initial Class Frequencies: AA: \_\_\_\_\_ Aa: \_\_\_\_\_ aa: \_\_\_\_\_  
p: \_\_\_\_\_ q: \_\_\_\_\_

My Initial Genotype: \_\_\_\_\_

F<sub>1</sub> Genotype: \_\_\_\_\_

F<sub>2</sub> Genotype: \_\_\_\_\_

F<sub>3</sub> Genotype: \_\_\_\_\_

F<sub>4</sub> Genotype: \_\_\_\_\_

F<sub>5</sub> Genotype: \_\_\_\_\_

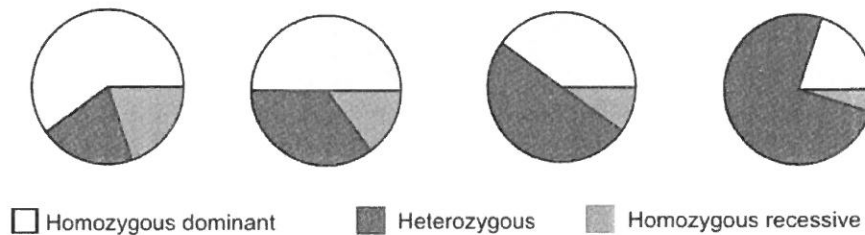
Final Class Frequencies: AA: \_\_\_\_\_ Aa: \_\_\_\_\_ aa: \_\_\_\_\_

**QUESTIONS**

1. Using the PTC tasting results for the entire class, how close were the frequencies of each phenotype (taster vs. non-taster) to those found in the North American population? If there was variation, what could have accounted for this?
2. In the first exercise in Part B, 'Testing an Ideal Hardy-Weinberg Population', what would be the expected values of P and Q after the five generations?
3. How close were your class's results to an ideal population.
4. What do you think would happen if you carried this simulation out for 10 more generations?
5. How do the frequencies of p and q compare after the factor of selection was added to the simulation?
6. What do you think would happen if you carried this simulation out for another 10 generations?
7. Do you think that the a allele would ever be totally eliminated from the population? Why or why not?
8. Suppose there was a medical advance that allowed most individuals with the double recessive condition to survive and reach reproductive age. How would this affect the allele frequencies?



9. Explain the difference in the results of the simulation showing selection to the simulation favoring heterozygotes.
10. Why is the heterozygous condition important in maintaining genetic variation within a population?
11. In the simulation demonstrating genetic drift, what do the resulting allele frequencies suggest about population size as an evolutionary force?
12. You are a population geneticist and you have recently visited a remote Pacific island where you have discovered a race of giant purple-skinned, seven-toed, three-horned dragons. In examining the new race you have discovered that a small number of the dragons have only six toes as opposed to seven and it seems to be a genetically inherited trait. After a population survey, you have found that in a population of 1,378 dragons, 174 of them have only six toes. Assuming the six-toed organisms are carrying a recessive trait, calculate the gene frequencies and the percentage of homozygous dominant, heterozygous, and recessive individuals on the island.
13. Below are four pie charts representing the percentage of homozygous dominant, heterozygous, and recessive individuals within a population over a period of 400 years. Examine the charts and explain below what you believe is happening within the population.



14. In this lab you examined the effects of selection, the heterozygous advantage, and genetic drift on a population. Several other factors can affect population genetics, including mutation, inbreeding, geographic isolation, and migration. Research one of these factors and explain how it may affect a population over time.
15. Divide the class in half and discuss the following statement: "All mutations are detrimental to a population."

