

Population Genetics and Evolution

BACKGROUND

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. 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 x p, or p²; for the “a” alleles, q x q, or q². 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.

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.

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.

DRIVING QUESTIONS

- How do we examine the effects of natural selection, heterozygous advantage, and genetic drift on allele frequencies in a simulated mating exercise?
- How do we demonstrate the stability of allele frequencies over five generations in an ideal Hardy-Weinberg population?

KEY TERMS

- | | | |
|-------------------------|----------------------------|----------------------------|
| • Population | • Inbreeding | • Gene |
| • Population size | • Hardy-Weinberg Principle | • Homozygous |
| • Population density | • Recessive alleles | • Heterozygous |
| • Population dispersion | • Natural selection | • Five Evolutionary Forces |
| • Dominant alleles | • Alleles | |

MATERIALS

PTC paper
control paper
4 index cards
coin

PRE-LAB QUESTIONS

1. Explain how natural selection affects populations?
2. State the Hardy-Weinberg Principle? Write the equation and label the variables.
3. Explain what the following statement means: Natural selection affects phenotypes and not genotypes.

PROCEDURAL FLOW CHART – create a flow chart outlining the steps you will take to be able to complete each of the activities below.

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. Record your results in Table 1 in the Analysis section.
4. Fill in the results for each individual in the class and enter the results in Table 2.
5. 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. Each person turns over the top card. This allele combination is the F₁ generation for only one person. Take back your card, place it back in the pile and reshuffle the cards. Each person turns over the top card. This allele combination is the F₁ generation for the other person.
4. Now, using the F₁ generation alleles you just made, create four new cards with those allele combinations. For example, if your F₁ generation was AA, all four cards will have A on them.
5. Find a new partner. Turn your cards upside down and shuffle them. Each person turns over the top card. This allele combination is the F₂ generation for only one person. Take back your card, place it back in the pile and reshuffle the cards. Each person turns over the top card. This allele combination is the F₂ generation for the other person.
6. Now, using the F₂ generation alleles you just made, create four new cards with those allele combinations. For example, if your F₂ generation was aa, all four cards will have a on them.
7. Repeat this procedure for three more generations and with three new partners, for a total of five generations. Record the genotypes for each.
8. Gather all the class data for the fifth generation.

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.

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

TABLE 1

	Taster	Nontaster
PTC		

TABLE 2

	Phenotypes				Allele frequency based on the Hardy-Weinberg equation	
	Tasters		Nontasters		p	q
	#	%	#	%		
Class population						

**Case B: Selection
Removal of aa genotypes**

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 C: Heterozygote
Advantage
Coin Flipping of AA**

Initial Class Frequencies: AA: _____ Aa: _____ aa: _____

My Initial Genotype: _____

F₁ Genotype: _____

F₂ Genotype: _____

F₃ Genotype: _____

F₄ Genotype: _____

F₅ Genotype: _____

Final Class Frequencies:
(After five generations) AA: _____ Aa: _____ aa: _____

 p: _____ q: _____

F₆ Genotype: _____

F₇ Genotype: _____

F₈ Genotype: _____

F₉ Genotype: _____

F₁₀ Genotype: _____

Final Class Frequencies:
(After ten generations) AA: _____ Aa: _____ aa: _____

 p: _____ q: _____

**Case D: Genetic Drift
Smaller Populations**

Initial Class Frequencies: AA: _____ Aa: _____ aa: _____
p: _____ q: _____

My Initial Genotype: _____

F₁ Genotype: _____

F₂ Genotype: _____

F₃ Genotype: _____

F₄ Genotype: _____

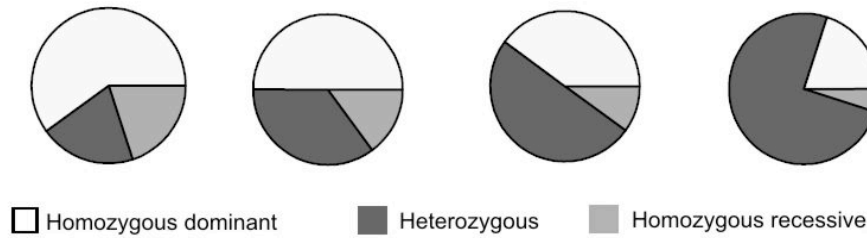
F₅ Genotype: _____

Final Class Frequencies: AA: _____ Aa: _____ aa: _____

QUESTIONS

1. 55% of the North American population can taste PTC while 45% of the population cannot. Using the PTC tasting results for the entire class, how close were the class 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?
3. How close were your class's results to an ideal population.
4. Do you think that the a allele would ever be totally eliminated from the population? Why or why not?
5. Explain the difference in the results of the simulation showing selection to the simulation favoring heterozygotes.
6. Why is the heterozygous condition important in maintaining genetic variation within a population?
7. 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.



LAB EXTENSION/INQUIRY

1. 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.
2. Choose and research one of the following observable traits. Design an experimental procedure where you would research the frequencies of the allele among various groups or within your family.

Tongue Roller
Hitchhiker's Thumb
Dimples

Widow's Peak
Hand clasping
Short Hallux

Free Ear Lobes
Mid-Digital Hair
Short Index Finger