Genetic Equilibrium: Human Diversity Teacher Version

California Science Content Standards:

- 2. Genetics: Mutation and sexual reproduction lead to genetic variation in a population.
- 2c. Students know how random chromosome segregation explains the probability that a particular allele will be in a gamete.
- 2g. Students know how to predict possible combinations of alleles in a zygote from the genetic makeup of the parents.
- 3. Genetics: A multicellular organism develops from a single zygote, and its phenotype depends on its genotype, which is established at fertilization.
- 3a. Students know how to predict the probable outcome of phenotypes in a genetic cross from the genotypes of the parents and mode of inheritance (autosomal or X-linked, dominant or recessive).
- 7. Evolution: The frequency of an allele in a gene pool of a population depends on many factors and may be stable or unstable over time.
- 7d. Students know variation within a species increases the likelihood that at least some members of a species will survive under changed environmental conditions.
- **7e. Students know the conditions for Hardy-Weinberg equilibrium in a population and why these conditions are not likely to appear in nature.
- **7f. Students know how to solve the H-W equation to predict the frequency of genotypes in a population, given the frequency of phenotypes.

Key Concepts:

- A **population** is a group of organisms of the same species that live and breed in the same area.
- **Alleles** are alternate forms of genes. In standard Mendelian genetics, two alleles—one from each parent—control an inherited trait or characteristic (e.g. seed pod color).
- A **gene pool** is the collective term representing all the genes, including all the different alleles, found in a population.
- The written expression of the DNA code, called the **genotype**, is normally represented using two letters, each letter representing one allele (e.g. AA).
- **Dominant** alleles are represented by capital (uppercase) letters.
- **Recessive** alleles are represented by lowercase letters.
- Genotypes with two dominant alleles (e.g. AA) are referred to as **homozygous dominant**.
- Genotypes with two recessive alleles (e.g. aa) are referred to as **homozygous recessive**.
- Genotypes with one dominant and one recessive allele (e.g. Aa) are referred to as **heterozygous**.
- The physical expression of the genotype (e.g. brown hair, blue eyes) is called the **phenotype**.

- In standard Mendelian genetics, the heterozygous condition (e.g. Aa) retains the homozygous dominant phenotype because the dominant allele masks the phenotype of the recessive allele. An example of this in humans would be a heterozygote for brown eye color. The person would carry both a dominant brown allele "A" and a recessive blue allele "a" yet have brown eyes. The dominant brown allele masks the recessive blue allele.
- **Genetic equilibrium** is the state in which allele frequencies remain constant.
- The **Hardy-Weinberg Principle**, named for English mathematician G.H. Hardy and German physician W. Weinberg, is a model used to help clarify evolutionary change by determining what happens if *no* change occurs. When no change occurs and an environment is stable, genetic equilibrium is maintained.
- The **Hardy-Weinberg Principle** states that for genetic equilibrium to be maintained from one generation to the next, five conditions must be met:
 - Random mating all individuals in a population have equal opportunity to pass on their alleles to offspring.
 - O Large populations **genetic drift**, or random changes in allele frequency, has less effect on large populations than small populations.
 - O No mutations if genes randomly mutate, new alleles may be introduced into the population and allele frequencies will change.
 - O No migration individuals cannot move into or out of the population.
 - O No natural selection all genotypes must have equal probabilities of reproduction.
- Two equations are used to model the Hardy-Weinberg Principle.
 - p + q = 1; where p represents the dominant allele, "A," and q represents the recessive allele, "a." This equation, used to calculate **allele frequency**, equals 1 or 100% of the population.
 - $p^2 + 2pq + q^2 = 1$; where p^2 represents the homozygous dominant genotype, 2pq represents the heterozygous genotype, and q^2 represents the homozygous recessive genotype. This equation, used to calculate **genotype frequency**, equals 1 or 100% of the population.

Materials:

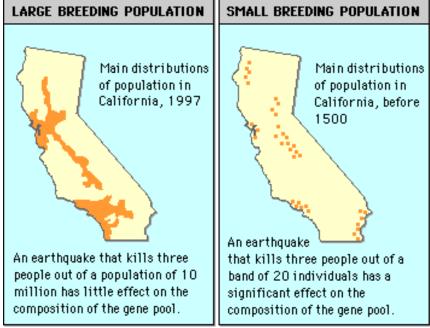
- White plastic or brown paper bag.
- Two types beans that are the same size and different colors.
- 70 dark colored beans (ex. pinto beans).
- 30 white or light colored beans (ex. white great northern beans).
- Calculators
- Computer (optional for part I)

Part I – Introduction

Procedure:

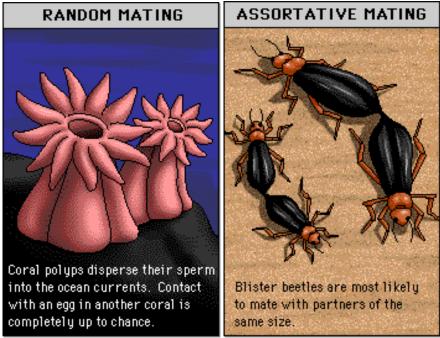
- If computers with Internet access are available for students, have them view the animation on the following website before moving on with parts II-IV below: http://www.phschool.com/science/biology_place/labbench/lab8/intro.html.
- Alternatively, students may view the animation as a group from a teacher's computer if
 available or simply interpret the still pictures included within the teacher/student versions
 of the lab.
 - o According to the Hardy-Weinberg Principle, there are five conditions required in order for a population to remain genetic equilibrium.

1. Large populations:



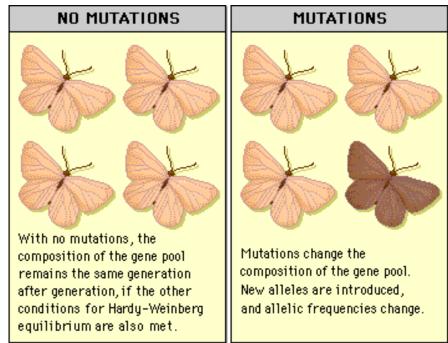
http://wwwphschool.com/science/biology_place/labbench/lab8/breedpop.html

2. Random mating:



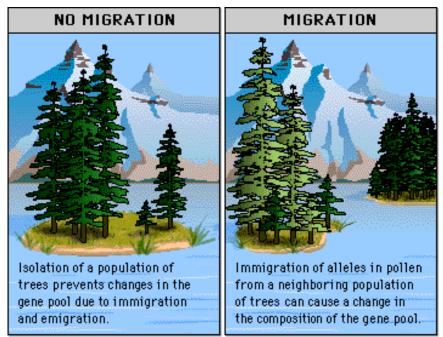
http://www.phschool.com/science/biology_place/labbench/lab8/mating.html

3. No mutations:



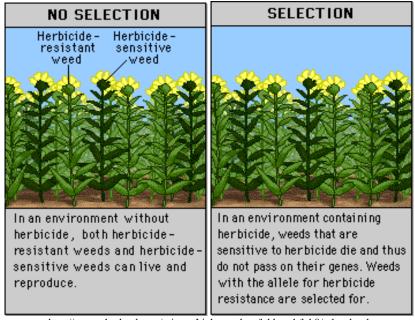
http://www.phschool.com/science/biology_place/labbench/lab8/mutation.htm

4. No immigration or emigration



http://www.phschool.com/science/biology_place/labbench/lab8/migrate.html

5. No natural selection



 $http://www.phschool.com/science/biology_place/labbench/lab8/select.html$

Part II – Testing the Hardy-Weinberg Principle using a Bean Model

Procedure:

Working in pairs, you will be testing the Hardy-Weinberg Principle by simulating a population using beans, beads, or similar materials. The bag of beans represents the gene pool for the population. Each individual bean represents one allele, whereas a pair or beans—either both dark, one dark and one light, or both light—represent the genotype for a single genetic characteristic or trait. Each bag should contain 100 beans of the two colors: 70 dark beans and 30 light beans.

The allele frequency is found by dividing the beans of one color by the total number of beans.

Ex. Allele frequency for dark beans = Number of dark beans / Total number of beans

 $\mathbf{b} = \underline{30 \ light}$ color and $\underline{0.30}$ allele frequency

Q1. How many diploid individuals are represented in this population?

50 diploid individuals are represented in the population.

Q2. What would be the color of the beans for a homozygous dominant individual?

Both beans would be dark in color (genotype "BB" where "B" is the dominant allele).

Q3. What would be the color of the beans for a homozygous recessive individual?

Both beans would be light in color (genotype "bb" where "b" is the recessive allele).

Q4. What would be the color of the beans for a heterozygous individual?

One bean would be dark, the other would be light (genotype "Bb").

- Place all 100 beans in the bag and shake. Without looking into the bag, randomly remove two beans from the bag. These two beans represent the genotype of one diploid individual in the next generation.
- Record the genotype of the individual (BB, Bb, or bb) in the data table below.
- Return the beans to the bag and shake. Replacing the beans to the bag each time maintains the size of the gene pool and the probability of selecting any allele should remain equal to its frequency.
- Select another two beans and record the genotype of this individual in the data table below. **Return the beans to the bag and shake.** Repeat selecting and replacing steps as outlined above until you have recorded the genotypes of 50 individuals who will form the next generation of the population.

Q5. Using the five conditions outlined in the Hardy-Weinberg Principle write a reasonable/logical hypothesis for this experiment.

What is the expected genotype rank (BB, Bb, and bb) from most to least number of times drawn?

Answers vary (Ex: In a large, randomly mating population where mutations, migration, and selection do not occur, allele and genotype frequencies will remain at equilibrium).

Data Table: Genotypes of Next Generation (alleles in population = 100)

Data Table:	Data Table: Genotypes of Next Generation (aneles in population = 100)									
(1)	(2)	(3)	(4)	(5)						
(6)	(7)	(8)	(9)	(10)						
(11)	(12)	(13)	(14)	(15)						
(16)	(17)	(18)	(19)	(20)						
(21)	(22)	(23)	(24)	(25)						
(26)	(27)	(28)	(29)	(30)						
(31)	(32)	(33)	(34)	(35)						
(36)	(37)	(38)	(39)	(40)						
(41)	(42)	(43)	(44)	(45)						
(46)	(47)	(48)	(49)	(50)						

- **Before calculating the results of your experiment**, determine the *expected* frequencies of alleles and genotypes for the new population and record in the table below.
- Use the original allele frequencies for the population provided above. Recall that the frequency of the dominant allele is *p* and the frequency of the recessive allele is *q*.
- Calculate the expected genotype frequencies using the H-W equation $p^2 + 2pq + q^2 = 1$.
- Next, calculate the number of individuals expected for each genotype by multiplying 50 (total diploid number) by the expected genotype frequencies.
- The dominant and recessive allele frequencies for the new population can be found by taking the square root of the homozygous dominant and homozygous recessive genotype frequencies, respectively.

Expected Allele/Genotype Frequencies for the Next Generation (alleles in population = 100)

Parent Po	opulation		N	ew Populatio	ns		
Allele Fr	requency		Frequency (n with specific		Allele Frequency		
"B" (p)	"b" (q)	"BB" (p²)	"Bb" (2pq)	"bb" (q²)	"B" (p)	"b" (q)	
0.70	0.30	0.49 (≈25)	0.42 (21)	0.09 (≈4)	0.70	0.30	

- Next, **using the results of your experiment**, calculate the *observed* frequencies in the new population that was created as you removed and replaced beans from the bag and record in the table below.
- For the parent population, use the same allele frequencies as before since you began with the same gene pool prior to selecting out and replacing individuals.
- Calculate the expected allele frequencies, genotype frequencies, and the number of individuals with specific genotypes for the new population as was done for the expected outcomes. Above

Observed Allele/Genotype Frequencies for the Next Generation (alleles in population = 100)

Parent Po	pulation		N	ew Populatio	ons	
Allele Fr	equency	~ ~	Frequency (n with specific	Allele Frequency		
"B" (p) 0.70	"b" (q) 0.30	"BB" (p²) Student Answer	"Bb" (2pq) Student Answer	"bb" (q²) Student Answer	"B" (p) Student Answer	"b" (q) Student Answer

The observed frequencies from the student experiment should be reasonably close the predicted frequencies (around .1 below or .1 above the predicted values).

Q6. Were your observed results consistent with the expected results based on your statistical analysis? If not, can you suggest an explanation?

Student answers vary. The observed distribution should not be significantly different from the expected distribution. However, do not expect the observed and expected distributions to fit Hardy-Weinberg equilibrium perfectly. Any minor differences can be attributed to chance or sampling error.

Q7. Compare your results to those of other student groups. How variable are the results for each group? Explain.

Student answers will vary, but the results should be similar.

Q8. Is this population evolving? Explain your response.

No. There are no statistically significant changes in genotype frequencies after one generation of random mating. The population is remains at genetic equilibrium.

Part III – Introducing a Conditional Change to Alter Genetic Equilibrium

Procedure:

Once again, you will be working in pairs and testing the Hardy-Weinberg Principle by simulating a population using beans, beads, or similar materials. However, with this new experiment, you will be introducing one conditional change—using a small population rather than a large one—in the hopes of significantly altering genetic equilibrium. As with the previous experiment, the bag of beans represents the gene pool for the population. Each individual bean represents one allele, whereas a pair or beans—either both light, one light and one dark, or both light—represent the genotype for a single genetic characteristic or trait. Each bag should contain 10 beans of the two colors: 7 dark beans and 3 light beans.

B =	<u> 7 dark</u>	color and	0.70	allele frequency
b =	<u> 3 light</u>	color and	0.30	allele frequency

Q9. How many diploid individuals are represented in this population?

Only 5 diploid individuals are represented in the population.

- Place the 10 beans in the bag and shake. Without looking into the bag, randomly remove two beans from the bag. These two beans represent the genotype of one diploid individual in the next generation.
- Record the genotype of the individual (AA, Aa, or aa) in the data table below.
- Return the beans to the bag and shake. Replacing the beans to the bag each time maintains the size of the gene pool and the probability of selecting any allele should remain equal to its frequency.
- Select another two beans and record the genotype of this individual in the data table below. **Return the beans to the bag and shake.** Repeat selecting and replacing steps as outlined above until you have recorded the genotypes of 5 individuals who will form the next generation of the population.

<u>Note</u>: Since the allele frequencies for both parental populations in Parts II and III are the same (p = 0.70 and q = 0.30), it is appropriate to reduce the number of offspring in the next generation in Part III from 50 diploid individuals to 5. It is reasonable to infer that a large

population of 100 alleles can produce 50 potential offspring in the next generation. However, it is unreasonable to infer that a small population of only 10 alleles can support the same number of offspring in the next generation as the large population. Therefore, the number of diploid offspring in Part III has been proportionally reduced compared to the number of diploid offspring in Part II.

Q10. Now that a conditional change is being included in the experiment, your initial hypothesis needs to be revised. Write a new, logical hypothesis that incorporates the change in the testing parameters.

Answers vary (Ex: In a small, randomly mating population where mutations, migration, and selection do not occur, allele and genotype frequencies will be unable to maintain genetic equilibrium, because not all the conditions of the Hardy-Weinberg Principle are occurring simultaneously).

Data Table: Genotypes of Next Generation (alleles in population = 10)

2000 200000 0000		7 4 4 4 4 4 4 4 4 4 4 4 4 4 4 4 4 4 4 4	op #12### 20)	
(1)	(2)	(3)	(4)	(5)

- **Before calculating the results of your experiment**, determine the *expected* frequencies of alleles and genotypes for the new population and record in the table below.
- Use the original allele frequencies for the population provided above. Recall that the frequency of the dominant allele is *p* and the frequency of the recessive allele is *q*.
- Calculate the expected genotype frequencies using the H-W equation $p^2 + 2pq + q^2 = 1$.
- Next, calculate the number of individuals expected for each genotype by multiplying 10 (total diploid number) by the expected genotype frequencies.
- The dominant and recessive allele frequencies for the new population can be found by taking the square root of the homozygous dominant and homozygous recessive genotype frequencies, respectively.

Expected Allele/Genotype Frequencies for the Next Generation (alleles in population = 10)

Parent P	opulation		N	ew Populatio	ns	
Allele Fı	equency	~ ~	Frequency (n with specifi	Allele Frequency		
"B" (p) 0.70	"b" (q) 0.30	(p^2) $(2pq)$ (q^2) 0.49 0.42 0.60		"bb" (q²) 0.09 (≈1.0)	"B" (p) 0.70	"b" (q) 0.30

- Next, **using the results of your experiment**, calculate the *observed* frequencies in the new population that was created as you removed and replaced beans from the bag and record in the table below.
- For the parent population, use the same allele frequencies as before since you began with the same gene pool prior to selecting out and replacing individuals.
- Calculate the expected allele frequencies, genotype frequencies, and the number of individuals with specific genotypes for the new population as was done for the expected outcomes. Above

Observed Allele/Genotype Frequencies for the Next Generation (alleles in population = 10)

Parent Po	pulation		New Populations						
Allele Fr	equency	~ -	Frequency (n with specific	Allele Frequency					
"B" (p)	"b" (q)	"BB" (p²)	"Bb" (2pq)	"bb" (q²)	"B" (p)	"b" (q)			
0.70	0.30	Student Answer	Student Answer	Student Answer	Student Answer	Student Answer			

The observed frequencies from the student experiment could vary dramatically from the predicted frequencies due to the small sample size.

Q11. Were your observed results consistent with the expected results based on your statistical analysis? If not, can you suggest an explanation?

Student answers vary. Students should see more of a significant difference between the observed distribution and the expected distribution than what was seen in the large population experiment. Genetic equilibrium cannot be maintained in a small population according to the Hardy-Weinberg Principle.

Q12. Compare your results to those of other student groups. How variable are the results for each group? Explain.

Student answers will vary, but they could be vary significantly or be identical. The small sample size for the genotypes will make each groups answers extremely variable.

Part IV – Genetic Equilibrium Problems

Procedure:

Use your knowledge of the Hardy-Weinberg Principle to solve the following population dynamics problems.

Teacher's Note: In this part of the lab, we see that the frequency of the recessive allele is greater than the frequency of the dominant allele. The numerical values for the frequencies are similar to those in a classical Hardy-Weinberg case, just flipped - "w" is the recessive allele but has the larger frequency and "W" is the dominant allele but has the smaller frequency. This can be a little confusing, but this scenario is plausible. If the population were to continue for a long time, it would eventually normalize out to what we would expect in a normal Hardy-Weinberg population.

Problem 1: Blue-footed Boobies



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Let "W" represent the allele for webbed feet and "w" represent the allele for non-webbed feet in Blue-footed Boobies. Birds with the "WW" or "Ww" genotype have webbed feet. Birds with the "ww" genotype have non-webbed feet. Below are the genotypes of 200 birds:

ww	ww	ww	$\mathbf{W}\mathbf{w}$	ww	WW	ww	$\mathbf{W}\mathbf{w}$	ww	$\mathbf{W}\mathbf{w}$	ww	ww	WW
ww	ww	ww	ww	ww	Ww	ww	Ww	ww	ww	ww	Ww	ww
Ww	ww	ww	Ww	Ww	ww	ww	ww	Ww	ww	Ww	ww	ww
Ww	ww	ww	Ww	ww	Ww	ww	Ww	ww	Ww	Ww	Ww	Ww
ww	ww	Ww	Ww	ww	Ww	ww	ww	ww	ww	Ww	ww	ww
Ww	ww	Ww	ww	ww	Ww	ww	ww	Ww	ww	ww	ww	ww
Ww	Ww	ww	ww	ww	Ww	ww	ww	Ww	ww	ww	Ww	Ww
ww	Ww	ww	ww	Ww	ww	Ww	ww	Ww	ww	Ww	ww	ww
Ww	ww	Ww	ww	ww	Ww	ww	Ww	ww	ww	ww	ww	ww
Ww	Ww	Ww	Ww	ww	ww	Ww	ww	Ww	ww	Ww	ww	Ww
ww	Ww	ww	Ww	Ww	Ww	Ww	ww	ww	ww	Ww	Ww	ww
Ww	ww	Ww	ww	Ww	ww	Ww	ww	Ww	Ww	ww	Ww	ww
Ww	ww	ww	Ww	ww	Ww	ww	Ww	ww	Ww	Ww	Ww	ww
ww	Ww	Ww	ww	Ww	ww	ww	Ww	ww	Ww	ww	ww	Ww
ww	Ww	Ww	ww	Ww	Ww	ww	ww	ww	ww	ww	ww	ww
ww	$\mathbf{W}\mathbf{w}$	$\mathbf{W}\mathbf{w}$	ww	ww								

Calculate the frequency of the alleles in the population. Express your answer as a decimal. Count of how many of each genome are listed

WW – 18

Ww - 83

ww - 99

Frequency of allele "W" =
$$\frac{119}{400}$$
 = 0.2975

Number of "w" alleles

Frequency of allele "w" = $\frac{119}{400}$ = 0.7025

Q13. The sum of the two frequencies, if calculated correctly will add up to what number?

1.0 or 100% of the population.

In the Hardy-Weinberg equations, the frequency of the dominant allele in a population is represented by the letter p, whereas the frequency of the recessive allele in a population is represented by the letter q.

In the next step, determine the relationship between the frequency of Blue-footed Boobies with non-webbed feet ("ww") and the frequency of the allele for non-webbed feet in the population ("w"). Use the following equation to calculate the frequency of non-webbed Blue-footed Boobies:

Q14. Is this frequency greater or smaller than the frequency of the "w" allele?

Smaller (frequency of Blue-footed Boobies with non-webbed feet = 0.49, whereas the frequency of the "w" allele = 0.7025).

Q15. If q = the frequency of "w" (the recessive allele) in the population, is the frequency of Blue-footed Boobies with non-webbed feet in the population equal to 2q or to q^2 ?

The frequency of Blue-footed Boobies with non-webbed feet in the population is equal to $q^2 [0.7025 \times 0.7025 \text{ or } (0.7025)^2 \approx 0.4935]$.

Q16. 2pq represents the genotype frequency of what individual? Do you expect these individuals to have webbed or non-webbed feet? Why?

2pq represents the genotype frequency of the heterozygote or heterozygous condition ("Ww"). Individuals with this genotype will all have webbed feet because the dominant "W" allele (representing webbed feet) masks the recessive "w" allele (representing non-webbed feet).

Q17. Match each of the following symbols with the phrase that defines it.

p

→ ____B___

A. The frequency of homozygous dominant individuals in the population.

q

→ ____E

B. The frequency of the dominant allele.

p + q = 1

 \rightarrow

<u>G</u>____

C. The Hardy-Weinberg equation for genotype frequency.

 p^2

-

D. The frequency of heterozygotes in the population.

2pq

 \rightarrow

D

E. The frequency of the recessive allele.

 q^2

 \rightarrow

<u>__F___</u>

F. The frequency of homozygous recessive individuals in the population.

 $p^2 + 2pq + q^2 = 1 \rightarrow \underline{\underline{C}}$

G. The Hardy-Weinberg equation for allele frequency.

ADVANCED VERSION ONLY

Problem 2: Wolves in the Forbidden Forest

In the next problem, you should be able to determine the frequencies of genotypes in a population. There are 100 wolves in the Forbidden Forest. Of the 100 wolves, 64 have the dominant phenotype of red fur color and 36 have the recessive phenotype of silver/black fur color. With "R" representing the dominant allele and "r" representing the recessive allele, the resulting genotypes in the population must be: "RR" or "Rr" code for red fur color and "rr" codes for silver/black fur color.

QSA18. What is q^2 (the frequency of the recessive genotype, "rr

The frequency of the recessive genotype, "rr," which is represented by q^2 is stated in the question. There are 36 wolves with the recessive phenotype, silver/black fur, which must be coded for by the genotype, "rr." Therefore, $q^2 = 0.36$.

QSA19. What is p^2 (the frequency of the dominant genotype, "RR")?

The above question showed $q^2 = 0.36$. To find q, the recessive allele frequency, simply take the square root of q^2 . $\sqrt{0.36} = 0.60$. Now, p can be solved for by using the allele frequency equation, p + q = 1.

p + 0.60 = 1, then p = 1 - 0.60. Thus, p = 0.40. Now, square p in order to solve for p^2 . $(0.40)^2 = 0.16$. $p^2 = 0.16$.

QSA20. Calculate 2pq (the frequency of the heterozygous genotype, "Rr."

p is the dominant allele in the population and has a frequency of 0.40. q is the recessive allele in the population and has a frequency of 0.60. Thus, 2pq is 2(0.40)(0.60) = 0.48.

ADVANCED VERSION ONLY

Problem 3: Albino Rats

In a population of 100 albino rats, 10 rats have short tails (genotype "tt"), which is the recessive phenotype. The remainder of the albino rat population shows the dominant phenotype of long tails and is homozygous dominant (genotype "Tt") or heterozygous (genotype "Tt"). Determine the frequencies of the genotypes and the dominant/recessive alleles in the population of albino rats.

$$p^{2} = 0.468$$
 $q^{2} = 0.10$
 $Q = 0.468 + 0.432 + 0.10 = 1.0 (p^{2} + 2pq + q^{2} = 1)$
 $Q = 0.432$
 $Q = 0.684$
 $Q = 0.316$
 $Q = 0.316$

QSA21. Use the above frequencies to help determine how many albino rats in the population will have the homozygous dominant genotype ("TT")? How many albino rats in the population will have the heterozygous genotype ("Tt")?

There will be approximately 47 albino rats with the homozygous dominant genotype ("TT"). 0.468 (p^2) x 100 individuals = approximately 47 albino "TT" rats.

There will be approximately 43 albino rats with the heterozygous genotype ("Tt"). 0.432 (2pq) x 100 individuals = approximately 43 albino "Tt" rats.

G.H. Hardy





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