Lab Section: ____________________ **Name:** ______________________________________

Pre-lab Homework Lab 7: Alleles in Populations

1. If you have a population of 30 dandelions and 10 are yellow and 20 are white;

What percent of the populations are yellow? __________ What percent are white? ____________

A "frequency" is just the decimal form of the percent value (ex. 33% is 0.33)

What frequency of the population is yellow? ____________ What frequency is white? ____________

2. Define "allele frequency".

3. How are allele frequencies and genotype frequencies related? How do they differ? (hint: look at the Hardy-Weinberg equations)

4. The Hardy-Weinberg equation is at equilibrium when $\mathbf{p^2 + 2pq + q^2}$ is equal to one. This describes a population that is not evolving. It is only true if certain assumptions are true. List these assumptions below.

5. A. In exercise 3, we will examine the effects of a reducing a population to very small size and the result of this on allele frequencies. What is the size of the initial population we will be modeling? How will we be reducing the population size?

   B. Will the above population stay in Hardy-Weinberg equilibrium? Why or why not? (hint: look at your answer to #3 above)

Name: ____ Date/Lab time: ___________________

Lab 7: Alleles in Populations

LAB SYNOPSIS:

- We will help Yogi Bear by using Hardy-Weinberg to calculate allele and genotype frequencies
- We will examine frequencies in real world populations (in our lab).
- We will look at how **genetic drift** can cause allele frequencies to change over time.

OBJECTIVES: After successfully completing this lab, a student will be able to:

- Describe a population of organisms using genotype and allele frequencies
- Use the Hardy-Weinberg equation to predict future genotype and allele frequencies
- Explain how the assumptions of the Hardy-Weinberg equation identify evolutionary forces that can change genotype and allele frequencies

Overview:

In the early part of the 1900s, Hardy and Weinberg came up with a mathematical equation that predicted the allele distribution in a population. This is useful in tracking alleles for specific genetic traits, for example genetic disorders. The Hardy-Weinberg equation is based on the following assumptions:

- The population is large
- The population is breeding randomly
- No mutation occurs (genetic variability is constant)
- There is no gene flow due to immigration or emigration
- There is no selection: gene combinations do not lead to an advantage over another combination

If these conditions are in place, the frequencies of alleles remain constant from generation to generation. This is known as the Hardy-Weinberg equilibrium.

Allele frequency- the measurement of how frequent an allele occurs within the population (expressed in decimal form)

The allele frequencies are based on the distribution of alleles resulting from monohybrid Punnett Cross (fig.1). Note: of the 8 alleles in the offspring the frequency of **A** would be 4/8 or 0.5 and the frequency of **a** would be 4/8 or 0.5. These are the gene pool of the population.

	A	a
A	AA	Aa
a	Aa	aa

Figure 1. Punnet square

The equation that describes allele frequencies in the Hardy-Weinberg model is:

$$\mathbf{p + q = 1}$$

Where **p** is the frequency of the dominant allele and **q** is the frequency of the recessive allele. Essentially this means that the sum of all the alleles in the population equals 100%, or as expressed in decimal form, equals 1

The genotypic ratio of a Punnett square (1 AA : 2 Aa : 1aa) is used to describe the genotypic frequencies. The Hardy-Weinberg equation mathematically describes the genotypic frequencies of the Punnett square:

$$\mathbf{A^2 + 2Aa + a^2 = 100\% \qquad or \qquad p^2 + 2pq + q^2 = 1}$$

Where $\mathbf{p^2}$ and $\mathbf{q^2}$ are the frequencies of the two homozygous genotypes, and **2pq** is the frequency of the heterozygous genotype. Essentially this means that the sum of all the possible genotypes in the population equals 100%, or as expressed in decimal form, equals 1

These equations are useful in determining expected frequencies of alleles in a population. And tell us when populations are evolving. That is, when allele frequencies do not follow Hardy-Weinberg.

Exercise 1: Calculating Allele Frequencies in the Jellystone Bear Population

The following help illustrate the usefulness of the equations of Hardy-Weinberg in determining a population's allele and genotype frequencies now, and in the future.

Hardy-Weinberg in Jellystone.

Modified from Jack Doepke, 1994. Woodrow Wilson Biology Institute
http://www.woodrow.org/teachers/bi/1994/easy.html

Yogi Bear decided one year that for future conservation plans regarding the usage of the park and its equipment, it would be important for him to gather some data about the bears that inhabit the park, so he petitioned the use of some of the other animals in the park and sent them out to gather some data. A week later the survey was completed and tabulated. It showed that 360 brown bears and 640 black bears resided in the park. The data also showed that the bears mated regardless of color, but that when brown bears mated with brown bears only brown cubs were produced, but when black bears mated with other black bears sometimes brown cubs were produced. Yogi also noticed that there did not appear to be any obvious survival value to having one color or the other.

The first question that Yogi wanted answered was which trait, black or brown, is dominant and which is recessive. Can you help Yogi figure this out with the information provided? Hint: To determine the answer use Punnett squares to set up a cross using all possible combinations of coat colors and look at the expected results. The answer will become obvious. Use "B" to represent the dominant color coat allele and "b" to represent the recessive allele

⇨ Which is the dominant color phenotype? __________ Which is the recessive color phenotype? _________

In making plans for purchasing new band uniforms for the Independence Day parade, Yogi thought it would be nice if the brown bears and black bears had different color ties. He next wanted to know if the percent of bears having brown coat color would be the same for the next generation. So he called upon the wise old owl, Doc Cronkite, for he was the wisest of all the animals in the forest.

'Doc,' he asked, 'using the information that I gathered in my survey, can I determine if the coat colors of the bears here in Jellystone will be the same in future generations to come?'

'Yes, Yogi, thanks to two very smart men who worked on genetics about 100 years ago by the names of Hardy and Weinberg, I think we can extract that information for you from your survey. First we have to go back and look at our old friend the Punnett square from our studies in Mendelian genetics. We are going to modify the way we use the square slightly to suit our purposes. Instead of thinking in terms of a single cross between two mating individuals, we are going to think in terms of the whole population of bears.

Now all the alleles for coat color, all the B and all the b alleles represent 100% of the alleles in the population. So you can represent all the alleles as B + b = 100%. Recall, another way of representing percent is as a frequency so you can write B + b =1. Hardy-Weinberg used **p** to symbolize the dominant allele and **q** to symbolize the recessive allele.

The H-W equation describing allele frequencies is: $\mathbf{p + q = 1}$

The H-W equation describing allele frequencies is: **$p + q = 1$**

So if you know the frequency of the recessive allele in the population, q, you can solve for frequency of dominant allele, p. $p = 1 - q$

'So' Yogi asks, 'how can we find the frequency of p?'

'We can use a Punnett square to help us figure this out.'

	B	b
B	BB	Bb
b	Bb	bb

Or as H-W

	p	q
p	pp	pq
q	pq	qq

Or as

	p	q
p	p^2	pq
q	pq	q^2

'Another way to represent a Punnett square is with math: $pp = p^2$, the two $pq = 2pq$, and $qq = q^2$. Since that is equal to the whole population (100%), we can write this out as the second H-W equation.'

The H-W equation describing genotype frequencies is: **$p^2 + 2pq + q^2 = 1$**

'How does that help? Asks Yogi'
'How many total bears where counted? And how many of them were brown?'
'1000 total bears; <u>360 were brown</u>.'

'In order to be brown, they must have two copies of the brown gene. That is the 360 brown bears must be homozygous recessive (bb). The 36% of bears that are brown is the q^2 from the Punnett square. To calculate its frequency it is:

$q^2 = 360/1000 = 0.36$

'We can now calculate q'. 'it is the square root of q^2

$q = \sqrt{0.36} = 0.6$

'Using the H-W allele frequency equation we can now solve for p'

$p + q = 1$ → $p = 1 - q$ → $p = 1 - 0.6$ so **$p = 0.4$**

*So **$p = 0.4$** and **$q = 0.6$** 'meaning for the population's gene pool, 40% of the alleles are B and 60% are b'

'Using the second H-W equation ($p^2 + 2pq + q^2 = 1$) we can now solve for the frequency and number of bears that would be bb, Bb and BB.'

**For the 1000 bears in Jellystone park, 360 are brown and 640 are black

$bb = q^2$ We already know that 36% of the bears are brown. So <u>360 bears are bb</u>

$Bb = 2pq$ so $2 \times 0.4 \times 0.6 = 0.48$ times 1000 bears or 48% = <u>480 bears</u>

$BB = p^2$ so $0.4^2 = 0.16$ times 1000 bears or 16% = <u>160 bears are BB</u>

]=640 black bears

‘Wow that is great’ says Yogi. ‘But which of the 640 black bears are BB and which are Bb?’

‘I am sorry Yogi, we can’t know by just looking at the bears which is BB or Bb. Either would give us a black bear.’ Remember, we could do a test cross to determine that.’

Tired and hungry from all of this math, Yogi and Doc took a break and walked down by the pool and had some cookies and milk. This was relaxing and fun for Yogi, but all Doc wanted to do was count the number of chocolate chips in the cookies. Yogi guessed that Doc must have inherited a mathematical gene from a parent. Probably his mother. Rested ,they began anew.

Using H-W to predict future frequencies of brown and black bears

'Doc, I'm ready to tackle my next question. How do I tell if the next generation of my bears will be the same frequency of color?'

'For that, Yogi, we have to ask ourselves, what kind of gametes can the bears that we have now produce and in what proportion?'

'How do we do that, Doc?'

'Well, you remember from your study of meiosis, don't you, that each parent carries two genes or alleles for a trait, but they only contribute one allele to their offspring.'

'Sure I remember that, Doc. That is pretty obvious; otherwise our chromosome number wouldn't stay constant from one generation to the next.'

‘That's right, Yogi, but do you remember which controls what gene goes into a gamete?’

‘Does it have something to do with dominance and recessive?’

‘No, Yogi, it is strictly a matter of chance.’

‘Oh! So our job should be easy. We just need to go back to our original percentages (frequencies) and work from there.’

-Can you help Yogi do this?

Recall: the second H-W equation ($p^2 + 2pq + q^2 = 1$)

16% BB + 48% Bb + 36% bb = 100%

Or written as frequencies…..

0.16 BB + 0.48 Bb + 0.36 bb = 1

We need to determine the total frequencies of the B alleles and the b alleles in the bear population.

We have a starting population of 1000 bears. Each bear has two alleles for coat color. That means there are 2000 alleles in the population. How many of these alleles are B and how many are b?

- 160 bears are BB. When they form gametes, what is the number of B? 2 x 160 = __________B
- 480 bears are Bb. When they form gametes, what is the number of B and what is the number of b?

1 x 480 = _________B and 1 x 480 = __________b

- 360 bears are bb. When they form gametes, what is the number of b? 2 x 360 = __________b

Now using these results, calculate the total number of B = __________

Now using these results, calculate the total number of b = __________

Now determine the frequency of B = total number of B/total number of alleles (2000) = __________ = p

Now determine the frequency of b = total number of b/total number of alleles (2000) = __________ = q

----

'Hey those are the same numbers we already got for p and for q.' Confirm this with "*" above.

'That's right. Now we need to set up a Punnett square to see what the frequencies of brown and black bears will be in the next generation.'

	0.4B	0.6b
0.4B	BB	Bb
0.6b	Bb	bb

Or as H-W

	p	q
p	p^2 0.4x0.4 =?	pq 0.4x0.6 =?
q	pq 0.4x0.6 =?	q^2 0.6x0.6 =?

Calculate into the boxes

	p	q
p		
q		

If there are 1000 bears in the next generation, calculate the following: (show work)

The number of brown bears are all the bb

So (q^2) x 1000 bears =____

The number of black bears are all the BB plus all the Bb

So (p^2 + 2pq) x 1000 bears =____

'Oh my gosh', says Yogi, 'that is the exact same numbers we got before.' Confirm this for "**" above.

'That's right. And it would be the same generation after generation after generation. This is a fundamental principle of the Hardy-Weinberg Equilibrium.'

‘Would it ever change?’ asks Yogi.

‘Yes, it would change if any of the following occur:

- Migration of new bears into or out of the park.
- The bear population gets smaller. (resulting in change due to chance)
- Mutations occur in the coat color gene.
- Matings between bears is not random.
- Natural selection for fur color occurs.’

‘Any of the above would cause the population to evolve. That is, they would cause the allele frequency to change over time.’

Exercise 2: Calculating Allele and Genotype Frequencies in a Real Population

Now we will examine a real population-the people in lab-and try to calculate frequencies for this population. To do this you need to score the phenotypes of all the individuals in the class (Table 1 below). We will use traits that are easy to score and we will pretend that there is a simple genetic explanation of their inheritance (although in many cases this is probably not true). To calculate the frequency of the recessive phenotype, we will divide the number with that phenotype by the total number of individuals we have examined.

Table 1: Phenotype frequencies for traits observed in a student population.

Genetic trait	**# with dominant phenotype**	**# with recessive phenotype**	**Total # of people**	**Freq of recessive phenotype** q^2 = # recessive/total
Tongue rolling	Yes =	No =		
Widow’s peak	Yes =	No =		
Free earlobe	Yes =	No =		
Blue eyes	No =	Yes =		
Hitchhiker’s thumb	No =	Yes =		
Hairy knuckles	Yes =	No =		
Taste PTC	Yes =	No =		
Sickle cell anemia	No =	Yes =		
Cystic fibrosis	No =	Yes =		

Recall the only phenotype you <u>know</u> the genotype of, are those showing the recessive phenotype. Those are the q^2. The square root of q^2= q. Recall: if you know q you can determine p.

1. The Hardy-Weinberg equilibrium equation can help us predict the number of people who are homozygous dominant and the number who are heterozygous. If you know (or assume) that the population is in Hardy-Weinberg equilibrium, then you can calculate allele frequencies from genotype frequencies.

Pick the two traits above that you think are most likely to be in equilibrium, and explain why you picked these traits in the space below. (review the assumptions of Hardy-Weinberg)

2. Using the H-W equations, calculate allele frequencies (p and q) for the first trait that you picked. Then determine the following genotypic frequencies. (show your work on a separate piece of paper)

Homozygous recessive (q^2) = ________ how many people in class are homozygous recessive? __________

Heterozygous (2pq) = ________ how many people in class are heterozygous? __________

Homozygous dominant (p^2) = ________ how many people in class are homozygous dominant? __________

3. Using the H-W equations, calculate allele frequencies (p and q) for the second trait that you picked. Then determine the following. (show work on a separate piece of paper)

Homozygous recessive (q^2) = ________ how many people in class are homozygous recessive? __________

Heterozygous (2pq) = ________ how many people in class are heterozygous? __________

Homozygous dominant (p^2) = ________ how many people in class are homozygous dominant? __________

Exercise 3: Gene Frequency and Genetic Drift

According to H-W, allele frequencies stay constant from generation to generation when:

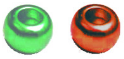
- The population is large
- The population is breeding randomly
- No mutation occurs (genetic variability is constant)
- There is no gene flow due to immigration or emigration
- There is no selection: gene combinations do not lead to an advantage over another combination

What are the consequences to the frequencies of alleles in a population when its size is suddenly reduced, for example by an earthquake, disease or fire?

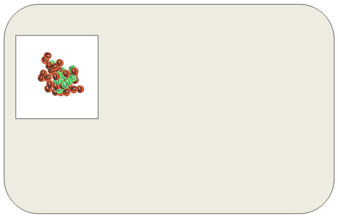
Genetic Drift- Change in allele frequencies over generations due to chance alone. Its effect is most pronounced in very small populations.

In this exercise, you will examine a situation where the allele frequencies of a population changes due to random chance. Changes in allele frequency caused by chance events, is called **genetic drift**. Genetic drift can be an important evolutionary force when a population's size is drastically reduced due to random events such as natural disasters, disease outbreaks, or habitat loss. In small populations, genetic drift can lead to the loss of all but one allele for a given trait; the remaining allele is then **fixed** in the population. Because genetic drift is caused by random chance and not by natural selective forces, beneficial alleles may be lost and harmful alleles may become fixed in the population.

Procedure: Simulating the effect of an earthquake on allele frequency



1. Construct an initial population:
We will track the change in allele frequency within a population of 50 organisms over a chance event. Alleles will be represented by colored beads. Put together a container with 60 “A” (red) beads and 40 “a” (green) beads. Recall: organisms are diploid so 100 beads represent the 100 alleles in these 50 diploid



2. Cut a 4 inch square piece of paper and set inside your lab prep tray. Carefully place all your beads on the paper.

3. Now simulate an earthquake by shaking your tray for 5 seconds.

Recall prior to the earthquake your population was at H-W equilibrium and allele frequencies were:
A (orange beads) = 60 / 100 = 0.6A
a (green beads) = 40 / 100 = 0.4a

4. Results of earthquake

How many beads remain on the piece of paper after shaking? _____________. These represent the remaining gene pool following the earthquake.

Calculate the allele frequencies within the population following the earthquake:

A = number of orange beads / total number of beads = ________________ A

a = number of green beads / total number of beads = ________________ a

1. What happened to allele frequencies in your population? Recall evolution is change in allele frequency over time. Is genetic drift evolution? Why or why not?

2. Adaptive evolution is the process by which organisms become better suited to their environment. Do you think genetic drift is adaptive?

3. If you were to repeat the above procedure, do you think you would obtain the same results?

4. Survey the other groups. Did all of the populations in lab have the same result? Why do you think this is?

Exercise 4: Evolving Populations and Genetic Drift

The above simulation looked at a population size of 50 individuals after just 1 generation. If you wanted to look at a much larger population size, or changes over 50 or 100 (or more!) generations, it would become very difficult to use beans to complete these simulations.

Computer simulations allow you to model genetic drift very quickly. You will use a simple simulation to look at the effect of population size on genetic drift and allele frequency.

Procedure:

1. Use a lab laptop to visit http://darwin.eeb.uconn.edu/simulations/drift.html

Use the drop-down menus to set the following parameters:

- Starting allele frequency: p = 0.5
- Population size: N = 1000
- Generations = 100

This means there are 1000 individuals and 50% of the alleles are p and 50% are q.

2. Push start.

A line will be generated on the graph representing 100 rounds of reproduction. If this population is at H-W equilibrium it should remain constant over time. Note: only one allele **p** is displayed.

3. Is the **p** allele staying constant over time (at H-W equilibrium)? Or is it changing (not at H-W equilibrium)?

4. What will be the effect of decreasing population size?

Without clearing your current results, generate two more lines. Both will use the same parameters as above, except that the population size for your second simulation should be N = 250, and the population size for your third simulation should be N = 25.

5. Compare the three results. What happens to the allele frequency as you decreased population size? Did the amount of fluctuation in allele frequency change? Did the **p** allele become extinct? Fixed? Explain this in the context of genetic drift.

6. Clear your results and reset N = 25.

Push start 8 times to generate 8 graphs on one page. Are results consistent or random? What does that tell you about genetic drift? Does genetic drift give random changes?