ANTHR1L – Biological Anthropology Lab **NAME:_____**

GENES IN POPULATIONS

(Portions of this lab have been adapted from Walker, S., ***Exploring Physical Anthropology: A Lab Manual & Workbook.*** Westview Press, 2007.

INTRODUCTION

Evolution, that is, change in living things over time, occurs through the process of many small genetic changes that take place from generation to generation. That's why the modern definition of evolution is a genetic one: *a change in the frequency of alleles from one generation to the next*. For this definition to be useful, you need to have a working definition of the terms *population*, *allele*, and *allele frequency*. A **population** is a group of freely interbreeding individuals. **Alleles** are different forms of the same gene. **Allele frequency** refers to the percentage of times a particular allele appears in a population. For any given trait (such as blood type or eye color), if you count the number of people with each form of a trait (how many with Type A blood or green eyes), your total count is a *frequency* of individuals with each form of a trait. Frequencies are expressed in percentages.

So what actually causes evolution? Or, rather, what causes allele frequencies to change? Evolution relies on four main "forces" that cause a change in gene frequency in a population over time. These are: **Natural Selection, Gene flow, Genetic drift,** and **Mutation.** The first three processes cause change in the frequency of various genes in the population by "redistributing" the existing alleles, while the fourth, mutation, is the only one to introduce *new* variation into the gene pool.

Natural Selection – Natural selection refers to the process by which those individual organisms that possess a favorable "variation" or "trait" within a particular environmental context are better adapted and thus more likely to survive and reproduce than those individuals that are less well-adapted. Therefore, the genes associated with the well-adapted traits will be passed onto the next generation with greater frequency than the genes for the less well-adapted traits. The effect of this *differential reproductive success* of individuals results in a change in gene frequency within the population. Over time, populations may become reproductively isolated from each other (geographically, behaviorally) such that splintered populations may no longer interbreed with members of the populations from which they were originally derived, that is, they may constitute a new species. Thus, natural selection acts on *individuals*, while evolution occurs at the level of the *population*

Gene Flow – Gene flow is the exchange of genes between populations. As individuals (and the genes they take with them) move to and from different populations, their movement alters the gene frequency in the populations they leave behind, as well as the population they join. This flow or exchange of genes between populations can result in evolutionary change.

Genetic Drift – Genetic drift involves the random "fission" (or splitting) of populations into entirely new or distinct populations with different gene frequencies. Two frequent causes of the fluctuation in gene frequencies are the **founder effect** and **genetic bottleneck**. If, for example, a small group of individuals separates itself from the rest of the population to "found" (i.e. establish) a new population, it will not accurately represent the frequency of the genes in the population at large, called the parent population. The small group that has broken away therefore contributes exclusively to the gene pool of the next generation. This is called the "*founder effect*" and could lead to phenomenon known as a "*genetic bottleneck"* since the "founding" group and its descendants carry only a small proportion of all the alleles (and of the variation) that were present in the original population. The founder effect and genetic bottlenecks can have a major impact on small populations including:

- A higher proportion of recessive genes, which can be "lost" more easily in a large population
- A greater chance of two recessive alleles coming together in zygote formation
- More recessively expressed traits, sometimes including genetic diseases
- Loss of genetic diversity overall, because fewer individuals contribute their genes to the gene pool

Mutation - A gene is a sequence of DNA bases that specifies the order of amino acids in a protein. A gene may take one of several alternative forms, called alleles. If an allele is altered during various genetic processes---mitosis, meiosis, DNA replication, or protein synthesis---a mutation has occurred. For such changes to have evolutionary significance, they must occur in the sex cells, which are passed between generations (Remember: evolution is defined as a change in allele frequencies between generations). Mutations provide entirely new sources of variation on which natural selection can act since alterations in an organism's genotype can translate into a radically different phenotype (physical characteristics/traits). Thus, mutations can bring about major changes to an organism's biology making evolution appear to occur relatively quickly rather than gradually. The *punctuated equilibrium* model of evolution proposes just this; that species tend to remain stable over long periods of time and evolutionary change occurs in sudden bursts (it is important to note, however, that in an evolutionary framework, "sudden" can mean thousands or tens of thousands of years).

In order to determine whether evolution might be at work in a population, we would track and calculate allele frequencies in the population over time. For example, suppose we test a population for the sickle cell genotypes and find the following:

AA (normal hemoglobin)	50 people
AS (sickle cell trait carrier)	100 people
SS (sickle cell anemia)	25 people
Total	175 people

To calculate the allele frequencies, we add up the total number of each allele and then divide by the total number of hemoglobin alleles in the population. Since each person has two alleles for each trait (example, AA for normal hemoglobin; AS for sickle cell trait; SS for sickle cell anemia), the total number of hemoglobin alleles would be twice the number of people in the population at the time of sampling.

Genotype	Number of People	Number of A alleles	Number of S alleles
AA	50	100 (2 x 50 people)	0
AS	100	100 (1 x 100 people)	100 (1 x 100 people)
SS	25	0	50
Total	175	200	150

To determine the frequency of the ***A alleles*** in the population we divide the number of A alleles by the total number of alleles in the population (175 people x 2 alleles = 350 (Total alleles in the population). So, 200 divided by 350 = .57 or 57%. Using the same reasoning, the frequency of the ***S alleles*** is .4285 or 43% (150 divided by 350). NOTE: Your percentages should always add up to 100—this is your self-check.

You will now use this formula to calculate allele frequencies within a population of students inhabiting an ANTHRO LAB classroom.

LAB EXERCISE

To demonstrate the effect of gene flow and genetic drift on allele frequencies in a population. After having been divided up into three sample populations, you will need to gather information for four traits for yourself and your classmates. The genes we will examine, and the phenotypes associated with their genotypes are described below.

1. Hitchhiker's thumb (**H** = straight; **h** = bent)
Hold your hand as though you were hitch-hiking. If the last joint of the thumb will bend back more than 60°, you are hh.

2. Interlacing fingers (**I** = left thumb over right; **i** = right thumb over left)
Clasp or fold your hands together with your fingers comfortably interlaced. Check to see which thumb is on top.

3. Free or attached earlobes (**E** = free earlobes; **e** = attached earlobes)
If there is any sign of an earlobe hanging free, the individual is E__.

4. Tongue rolling (**R** = ability to roll; **r** = inability to roll)
Stick out your tongue and try to roll it into a tube.

Note: If you are dominant for any of the traits above, we will need to determine the second allele of your genotype since we do not have intergenerational data from which to work. We already know that you have at least one dominant allele if you exhibit the dominant form of the trait, so we will flip a coin to randomly assign you either a homozygous dominant (two capital letters) or heterozygous dominant (one capital and one lowercase letter) genotype.

Flip the coin. Heads = a dominant allele and Tails = a recessive allele.

Now, record the data of your group members

VERY IMPORTANT: Make sure as you write the student names in your table, all group members *write them in the same order, one name at a time.* This will make it easier to transfer student information when migration occurs.

PART ONE 1. Record the data for yourself and each student in your group in the table below.

STUDENT NAME	Hitchhiker's Thumb	Interlacing Fingers	Earlobes	Tongue rolling

2. From the above data, determine the frequencies of the dominant and recessive alleles for each trait using the tables below. (Total number of alleles recorded divided by the total number of alleles in the population).

Genotype	# of people	# H alleles	# h alleles
HH			0
Hh			
hh		0	
TOTAL			

3. What is the frequency of the **H alleles** in your group's population?_____

4. What is the frequency of the **h alleles**?_____

Genotype	# of people	# I alleles	# i alleles
II			0
Ii			
ii		0	
TOTAL			

5. What is the frequency of the **I alleles** in your group's population?_____

6. What is the frequency of the **i alleles** in the population?_____

Genotype	# of people	# E alleles	# e alleles
EE			0
Ee			
ee		0	
TOTAL			

7. What is the frequency of the **E alleles**?_____

8. What is the frequency of the **e alleles**?_____

Genotype	# of people	# R alleles	# r alleles
RR			0
Rr			
rr		0	
TOTAL			

9. What is the frequency of the **R alleles**?_____

10. What is the frequency of the **r alleles**?_____

11. Now, go to the table in Part Three and record the allele frequencies under the Population 1 column.

PART TWO

To illustrate the effect of gene flow and genetic drift on allele frequencies, your instructor will select three students at random from each group who will then migrate to and become part of another population. (Wait for directions from instructor).

1. (Re)record the data for yourself and original members of your group in the table below and record the data for the new members (immigrants) who have joined your group.

STUDENT NAME	Hitchhiker's Thumb	Interlacing Fingers	Earlobes	Tongue Rolling

2. From the new set of data, once again calculate the frequencies of the dominant and recessive alleles for each trait using the tables below.

Genotype	# of people	# H alleles	# h alleles
HH			0
Hh			
hh		0	
TOTAL			

3. What is the frequency of the **H alleles** in your group's population?_____

4. What is the freqency of the **h alleles**?_____

Genotype	# of people	# I alleles	# i alleles
II			0
Ii			
ii		0	
TOTAL			

5. What is the frequency of the **I alleles** in your group's population?_____

6. What is the frequency of the **i alleles** in the population?_____

Genotype	# of people	# E alleles	# e alleles
EE			0
Ee			
ee		0	
TOTAL			

7. What is the frequency of the **E alleles**?_____

8. What is the frequency of the **e alleles**?_____

Genotype	# of people	# R alleles	# r alleles
RR			0
Rr			
rr		0	
TOTAL			

9. What is the frequency of the **R alleles**?_____

10. What is the frequency of the **r alleles**?_____

PART THREE

Use the data you collected in Part 2 to compare the allele frequencies of Population 1 to Population 2 by completing the table below.

Allele	Population 1	Population 2
H		
h		
I		
i		
E		
e		
R		
r		

1. Were there any significant changes in allele frequencies between the two populations?_____

2. What alleles showed the most significant change?

3. If the individuals who migrated into your group interbred with the individuals who remained in the group, would this be an example of genetic drift or gene flow? Explain.

4. What if the group members who had migrated out of their original populations had formed their own population instead of joining an existing population? Would this be an example of gene flow or genetic drift?

PART FOUR

BLOOD TYPING (Adapted from France 2001, *Lab Manual and Workbook for Physical Anthropology*, 4th Edition).

The ABO blood group system is one of the best understood multiple allele systems. All of the ABO blood types are determined by two genetically determined proteins called **antigens**, the A antigen and the B antigen. These proteins occur on the surface of the red blood cells. An individual's blood may contain one, both or neither of these antigens, giving rise to the four blood groups: A, B, AB, and O, as shown:

Blood Group (phenotype)	Genotype
Type A	AA or AO
Type B	BB or BO
Type AB	AB
Type O	OO

As one can deduce form the above chart, the A and the B alleles are dominant to the O allele, while the A and B alleles are codominant with one another.

1. Imagine you are a health care worker collecting data in a small village in the Phillipines. You and your research assistants have found the following about blood types from samples given by local villagers:

35 people who are heterozygous dominant for Type A blood; 100 people who are heterozygous dominant for Type B blood; 25 people who have Type AB blood; and 50 people with Type O blood.

Use the above information to complete the table below to calculate the allele frequencies.

Genotype	**# of People**	**# of A alleles**	**# of B alleles**	**# of O alleles**
Totals				

NOTE: Remember, the allele frequency is the total number of that particular allele divided by the total number of ALLELES (not people) in the population.

Frequency of A allele = _____

Frequency of B allele = _____

Frequency of O allele = _____

2. Now, imagine that you return to the same village ten years later to check on the progress of the health programs that were implemented. Once again, your team of researchers must collect data on blood types. This time your data reveal the following:

10 people who are homozygous dominant for Blood type A; 50 people who are homozygous dominant for Blood Type B; 25 people who have blood type AB; and 150 people with Type O blood.

Use the above information to complete the table below to calculate the allele frequencies.

Genotype	**# of People**	**# of A alleles**	**# of B alleles**	**# of O alleles**
Totals				

Frequency of A allele = _____

Frequency of B allele = _____

Frequency of O allele = _____

Obviously, some change has taken place over time in the frequency of genotypes in the total population. Have allele frequencies changed as well? In other words, is evolution, by the genetic definition, taking place?

Explain below why this change is so critical for evolution based on what you have learned about genotypes and phenotypes.