STUDY GUIDE *- Ch. 23.1 - Genetic Variations Make Evolution Possible* **NAME:** _____
- Ch. 23.2 - The Hardy-Weinberg Equation can be Used to Test Whether a Population is Evolving

- **PHYSICALLY PRINT OUT this PDF and HANDWRITE (with a black or blue pen) your answers directly on this PDF.** *Typed or digitally-written work is **not** accepted. Do **not** answer questions on separate paper.*
- **Importantly, study guides are NOT GROUP PROJECTS!!!** *You, and you alone, are to answer the questions as you **read** your assigned textbook. You are **not** to share answers with other students. You are **not** to copy any answers from any other source, including the internet.*
- **Get in the habit of writing LEGIBLY, neatly, and in a medium-sized font.** *AP essay readers and I will skip grading anything that cannot be easily read so start perfecting your handwriting, and don't write so large you can't add all the relevant details and key elaborations in the space provided.*
- **SCAN physical documents in color and with good resolution. Then, upload your final work as PDFs to Archie.** *Avoid uploading dark, shaded, washed-out, sideways, or upside-down scans of homework. Keep completed physical study guides organized in your biology binder to use as future study and review tools.*
- **READ FOR UNDERSTANDING and not merely to complete an assignment.** ***First**, read a section quickly to get an overview of the topic covered. Then, read it a **second** time slowly, paraphrasing each paragraph **out loud** and analyzing every figure. Finally, read it a **third** time as you answer the study guide questions if assigned and start building your memory. Try to write answers out in your own words, when possible, and try to purposefully and accurately use all new terminology introduced.*

Remember: INDIVIDUALS DO NOT EVOLVE! POPULATIONS EVOLVE.

Evolution occurs when the genetic composition of an entire population changes over time.

Individuals' phenotypes (traits of characters), **not** their genotypes, interact with the environment, affecting each individual's chance of survival and, therefore, each individual's reproductive success. **We say that natural selection acts on an individual level therefore, each individual being more or less capable of surviving and reproducing in that particular environment, given their particular phenotype.**

Any **mechanisms** that changes the number and/or types of alleles of genes found **collectively** in all individuals in a **population** changes the POPULATION'S GENETIC COMPOSITION. Those mechanisms cause EVOLUTION, **a change in the genetic (genotypic or allelic) make up of a population.**

When tackling the study guide for this chapter, read each concept first to get the big picture, then go back to work on the details in the questions. Don't lose sight of gaining a larger conceptual understanding by getting lost in the details!

1. Define the term **microevolution**.

2. a. In addition to new **mutations** and **sexual selection (nonrandom mating)**, what are **three main mechanisms that can cause changes in allele frequency**? Define all three terms.

   1. _____ = (definition)

   2. _____ = (definition)

   3. _____ = (definition)

   b. All mechanisms for evolution change the frequency of alleles in a population, but which is the **only mechanism that is adaptive**, improving the match between organisms and their environment and causing the spread of adaptations (features that help organisms survive and reproduce in particular environments) in subsequent generations?

3. a. Using the techniques of molecular biology, what are the **two ways of measuring genetic variation in a population**? Define each term.

   1. _____ = (definition)

   2. _____ = (definition)

   b. What does it mean if a population has an **average heterozygosity** of 20%, but a **nucleotide variability** of 5%?

   c. Explain **two reason why nucleotide variation may not actually result in phenotype variation** in the organism?

   1.

   2.

4. *Think* - Review 23.4, 17.6, and 17.11, and think over all you now know about gene expression.

   a. Explain how a base-pair substitution that alters a coding region of the Adh locus could have no effect on amino acid sequence. *Use the proper terms you learned in ch.17 in your answer.*

   b. Then explain how an insertion in an exon ***could*** have no effect on the protein produced (even though other times it can have an effect).

(Check your answers to 4.a. & b. by going to the Ch.23 ***Figure Questions*** *for* ***Figure 23.4*** *in Appendix A of your textbook)*

5. Explain which **type of phenotypic variation influences a population's evolution**?

6. a. What is the **ultimate source of new alleles**?

   b. What is the **ultimate source of new genes**?

   c. How does **sexual reproduction contribute to genetic variation** in organisms in a sexually reproducing population?

   d. What are three **examples of causes of mutations**?

   1.

   2.

   3.

7. a. A **mutation is any change in the DNA nucleotide sequence of an organism's DNA**. These mutations provide the raw material from which new traits may arise, these traits than being selected for or against by natural selection. What occurs in a **point mutation** again?

   b. Why do **most point mutations in coding regions of genes have a small or large negative effect on organisms rather and only rarely a positive one**?

   c. If mutations change phenotypes into versions that decrease survivability or reproductive success, **why does natural selection not quickly result in the removal of such harmful alleles from a population**? (This especially true for organisms that have a ploidy of 2 or more such as diploids).

8. a. What is **neutral variation**?

   b. **If you change the structure/shape of a protein, often you change its function, ability to function, or functional effectiveness.** Neutral variation can result from changes in DNA outside of coding and (noncoding) regulatory sequences of genes. Those areas of DNA may not influence the phenotype of the organism as they may not influence when, how much, or what type of RNA or protein is made by a cell/organism. Contrastingly, changes in regulatory sequences or coding regions definitely have the ability to influence a cell/organisms's phenotype for better or worse. How come you can occasionally have a mutation occur in a coding region of a gene, which leads to a change in a protein's primary structure (amino acid sequence), and still consider that mutation neutral? Explain.

9. a. Occasionally, a mutation may turn out to be beneficial to a cell or to an organism by altering the phenotype in a advantageous way. When is a **point mutation considered beneficial in biology?**

   b. **When do mutations become a part of the next generation's gene pool**? (When can mutations be passed onto offspring and become part of the genotypes of all the cells that make up the offspring, being able to be passed down to the offspring's offspring, and through the generations, as well)?

10. a. *Think* - Why do you think large scale mutations in DNA that involve many genes are often quite harmful?

b. What is a **translocatio**n mutation again?

b. Though usually harmful, how could it in rare instances be **beneficial**?

11. a. What three events could lead to **gene duplication**?

1.

2.

3.

b. Importantly, how might **gene duplication play a role in the future evolution of a population**?

12. **Why do prokaryotes seem to evolve so much quicker than many eukaryote**s even though mutation rates are sometimes slightly higher in eukaryotes than prokaryotes and both make DNA repair proteins?

13. Much of the genetic variation that makes evolution possible comes through sexual reproduction. Review your ch.13. Name and describe what happens during each of the **three mechanisms by which sexual reproduction shuffles existing alleles**.

1. _____ =

2. _____ =

3. _____ =

14. *Think:* Because Darwin did not know about the work of Gregor Mendel, he could not explain how organisms pass heritable traits to their offspring. Today we know that DNA (genes) are passed from parents to offsprings, those genes encoding RNA and proteins that then produce phenotypes. Why is **genetic variation within a population a prerequisite for evolution**? *(Check your answers by going to the Ch.23.1* ***Concept Check Question #1*** *in Appendix A of your textbook)*

15. Let's see if you understood all you just read about. Of all the mutations that occur in a population, why do only a small fraction become widespread?*(Check your answers by going to the Ch.23.1* ***Concept Check Question #2*** *in Appendix A of your textbook)*

16. *Think:* If a sexually reproducing population suddenly started reproducing asexually only, how would its genetic variation be affected over time? **Explain.** *(Check your answers by going to the Ch.23.1* ***Concept Check Question #3*** *in Appendix A of your textbook)*

17. Just because genetic variation exists in a population *(different alleles exist for certain genes)* does **not** mean the population is currently evolving. Certain **mechanisms** must be active causing a change in the frequency of alleles for one or more genes in order for us to say that the population is evolving. Define the term **population** formally.

18. a. What is a population's **gene pool**?

    b. **The greater the number of fixed alleles, the lower the species' diversity.** When do we say that an **allele is fixed** within a population's gene pool?

    c. What are the possible genotypes of all individuals in a population when it comes to two genes, one of which is fixed with allele "A" and the other gene is not fixed, having two allele possibilities, "B" or "b"? *(Assume the individuals of this species are diploid).*

    _____ **&** _____ **&** _____

    d. Suppose a population of organisms with 500 loci (locus = location of a gene) is fixed at half of these loci and has two alleles at each of the other loci. How many different **types** of alleles for genes are found in its entire gene pool? **Explain.**

    e. If a population has 2000 diploid organisms. How many total **number** of alleles for one locus are in the gene pool?

P.S. The answer to 18.d. is ***750*** *different* ***types*** *of alleles among all the 500 genes (250 different allele types - one for each of the 250 fixed genes - plus 500 different allele types for the 250 heterozygote genes - 2 possible alleles per gene) and the answer to 18.e. is* ***4000*** *total* ***#*** *of alleles (every organism carries two copies of that gene so that is 4000 copies in 2000 individuals)*

19. a. **Always calculate a population's ALLELE frequency DIRECTLY from the gene pool** *when possible,* **using the following formula:**

Frequency of an Allele in a Population = $\frac{\text{\# of Copies of the Specific Allele in the Population}}{\text{Total \# of all Type of Alleles in the Population}}$

b. What variable is used to symbolize the **FREQUENCY of one allele** (not the the allele itself!) in a population where two alleles exist for a given gene?

c. What variable is used to symbolize the **FREQUENCY of a second allele** (not the the other allele itself!) in a population where two alleles exist for a given gene?

FYI: When the two alleles of a gene are **co-dominant** and both the effects of both alleles can be seen discretely in a heterozygote's phenotype or the alleles follow an **incomplete inheritance pattern,** the phenotype of the heterozygote looking like a mixture of or intermediate between of the phenotype of the two homozygous genotypes, the **FREQUENCY of either allele can be designated as the p or q.**

FYI: When one allele behaves dominantly over another allele, however, it is convention to use **p** to symbolize the **FREQUENCY of the DOMINANT allele** and **q** to symbolize the **FREQUENCY of the RECESSIVE allele.**

d. *Let's practice:* A population has 400 individuals of genotype BB, 200 of genotype Bb, and 50 of genotype bb. Calculate the frequency of the b allele. Show your work.

20. a. "**The Hardy-Weinberg Principle" is used to describe a population that is NOT evolving**! What does this principle state has to happen to the **FREQUENCIES** of alleles and of genotypes in a population for the population to **NOT** evolve? In other words, in reference to the frequencies of the alleles and genotypes in a population, **what does it mean when a population is said to be in Hardy-Weinberg Equilibrium**?

b. If a population has only two alleles, then the **frequency of the two alleles must add up to 1 if that population is not evolving** (if no mechanism is occurring that would change the frequency of p or q). So **for a population in Hardy-Weinberg Equilibrium:**

$$p + q = 1$$

21. a. Read the rest of **Ch.23 Section 2**, including **Figure 23.7** and **Figure 23.8** to familiarize yourself with the concept and the mathematics related to a population being in Hardy-Weinberg Equilibrium.

If a population with two alleles for a gene is in Hardy-Weinberg Equilibrium for that one gene, and you know the frequency of the dominant and recessive alleles of this gene, which are not changing over time in this population (because the population is not evolving at this time), what must be the **FREQUENCY of the three types of possible genotypes**.

- **FREQUENCY of the Homozygous Dominant (or Homozygous for Allele #1) Genotype** = _____
- **FREQUENCY of the Heterozygous Genotype** = _____
- **FREQUENCY of the Homozygous Recessive (or Homozygous for Allele #2) Genotype** = _____

b. If a population has three possible genotypes, and the frequency of the p and q alleles is not changing (because the population is **not** evolving), then the **frequency of the three genotypes must add up to 1** (if no evolutionary mechanism is occurring that would change the frequency of the two alleles or three genotypes expected). So **for a population in Hardy-Weinberg Equilibrium:**

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

22. Memorize Table 23.1. What are the **five conditions that must be met for a gene in a population to be in Hardy-Weinberg Equilibrium**. Explain each one. *(It is very important for you to know and understand these five!)*

1. **Condition** = NO _____
   Description of the Evolutionary Mechanism that Breaks this Condition =

2. **Condition** = _____ **(NO Sexual Selection)**
   Description of the Evolutionary Mechanism that Breaks this Condition =

3. **Condition** = NO _____
   Description of the Evolutionary Mechanism that Breaks this Condition =

4. **Condition** = _____ **(NO Genetic Drift)**
   Description of the Evolutionary Mechanism that Breaks this Condition =

5. **Condition** = NO _____
   Description of the Evolutionary Mechanism that Breaks this Condition =

It is very unlikely that all five of these conditions will occur and that the allelic or genotypic frequencies will stay the same for every single gene loci in a population, though one loci may be in Hardy-Weinberg Equilibrium while other are evolving. Allelic frequencies do often change for a gene. Populations, therefore, are often evolving.

Populations can be tested to see if they are **NOT** evolving (or evolving) using the **Hardy-Weinberg Equation** & **Chi Square Test for Goodness of Fit**.

We should assume initially that a population is not evolving (our Null Hypothesis, H_0) - that no mechanism is occurring that would change p and q (and thus p^2, 2pq, and q^2).

Only, if the Null Hypothesis proves incorrect, do we support the Alternate Hypothesis (H_A) that the population is evolving.

1. **Determining what the INITIAL/EXPECTED p and q** (frequency of dominant allele / allele #1 and frequency of recessive allele / allele #2) **should be and remain if a population is in Hardy-Weinberg Equilibrium (not evolving)**. The p and q should, when possible, be calculated directly from the total alleles in the gene pool.

$$p = \frac{\text{Total \# of Dominant Alleles}}{\text{Total \# of Alleles}} \qquad q = \frac{\text{Total \# of Recessive Alleles}}{\text{Total \# of Alleles}}$$

   If the population is in Hardy Weinberg Equilibrium, meaning the population is not evolving for that gene **(meaning there is NO sexual selection, NO mutations, NO genetic drift, NO natural selection, and NO gene flow (migration) occurring)**, then this value for p and q must remain the SAME at a future time as well.

2. Based on this p and q, **calculate what the initial/THEORETICAL EXPECTED genotype frequencies p^2, q^2, & 2pq must be if the population is indeed in Hardy-Weinberg Equilibrium (not evolving).**

3. To see if the population has evolved over time, y**ou have to calculate the currently OBSERVED genotype frequencies p^2, q^2, & 2pq and, USE THESE observed genotype frequencies to calculate the OBSERVED allele frequencies p & q in order to COMPARE these calculations to the initial allele frequencies or the initial/ theoretical expected genotype frequencies.**

   To calculate the actual **OBSERVED genotype frequencies**, use the following formulas and use the # of each individual with each genotype observed in the population.

$$p^2 = \frac{\text{Total \# of Homozygous Dominant Individuals (Genotypes)}}{\text{Total number of Individuals (Genotypes)}}$$

$$q^2 = \frac{\text{Total \# of Homozygous Recessive Individuals (Genotypes)}}{\text{Total number of Individuals (Genotypes)}}$$

$$2pq = \frac{\text{Total \# of Heterozygous Individuals (Genotypes)}}{\text{Total number of Individuals (Genotypes)}}$$

   If these later/OBSERVED genotype frequencies do not match the initial/THEORETICAL EXPECTED genotype frequencies, or $p^2 + 2pq + q^2$ does not equal 1, then the population is probably evolving due to one or more of the 5 evolutionary mechanisms!!!

   Similarly, a population has evolved when its allelic frequencies (p and q) have changed!

   To find the later/ OBSERVED allelic frequencies, calculate the p and q indirectly from OBSERVED genotype frequencies. Calculate p^2 or q^2, then square root either to get the current OBSERVED p or q. If these OBSERVED allele frequencies do not match the INITIAL EXPECTED allele frequencies, or the current p + q do not equal 1, then the population is evolving and one or more mechanisms of evolution are changing the allele frequencies.

A Chi Square Test for Goodness of Fit can be performed to determine statistically if the observed # of individuals with each genotype (or # of each allele in a gene pool) differs from the expected # of individuals with each genotype (or # of each allele in a gene pool) due to chance alone (HO) or due to one or more evolutionary mechanisms (HA). *(Remember, you cannot plug frequency values into the Chi Square formula. You must calculate the total number of individuals with each genotype or the total number of each type of allele)*

23. Now that you have studied the process of using the Hardy Weinberg Equations to test that a population is **not** evolving, let's try the **Scientific Skills Exercise**: Using the Hardy-Weinberg Equation to Interpret Data & Make Predictions.

1. (As step 1 on the previous pages instructs, calculate your Hardy-Weinberg Equilibrium allele frequencies **DIRECTLY** from the total number of alleles in the Gene Pool **NOT** from the genotype frequencies you can also calculate!!!)

2. (As step 2 on the previous pages instructs, calculate your Hardy-Weinberg Equilibrium expected genotype frequencies **DIRECTLY** from the total number of alleles in the Gene Pool **NOT** from the genotype frequencies you can also calculate!!!)

3. a.

b.

4. a.

b.

5. a. Hypothesis:

b. Prediction:

Additional Tip = If you cannot calculate the p and q directly from the total alleles in a gene pool or you are only given the number of organisms with different phenotypes but no information on each of their genotypes, you will have to use the genotype frequencies to find the p or q indirectly. **In this situation, try always to use q^2 to calculate q, and then use q to calculate p** since the homozygous recessive genotype is the only genotype that leads to the homozygous phenotype. Therefore, **the frequency of the recessive phenotype in a population = the frequency for the recessive genotype in a population (q^2).**

Individuals with heterozygous or homozygous dominant genotypes will both show the dominant phenotype so if you know the number of individuals with the dominant phenotype, you still don't know how many of those are homozygous dominant or heterozygous in genotype to calculate p^2 or 2pq. **The frequency for the dominant phenotype = the frequency for the homozygous dominant genotype + the frequency of the heterozygous genotype (p^2 + 2pq)**

24. A population has 85 individuals of genotype AA, 320 of genotype Aa, and 295 of genotype aa. Calculate the allele frequencies and calculate the genotype frequencies in this population. *Show all work.* *(Check your answers by going to the Ch.23.2* ***Concept Check Question #1*** *in Appendix A of your textbook)*

Allele Frequencies:

Genotype Frequencies:

25. The frequency of allele a is 0.45 for a population **in Hardy-Weinberg Equilibrium**. What are the expected frequencies of genotypes AA, Aa, and aa. *Show all work.* *(Check your answers by going to the Ch.23.2* ***Concept Check Question #2*** *in Appendix A of your textbook)*

First you must calculate the frequency of A:

Frequency of AA:

Frequency of Aa:

Frequency of aa:

26. In a population of plants, 64% exhibit the dominant flower color (red), and 36% of the plants have white flowers. What is the frequency of the dominant allele? (**Be cautious:** **1.** Always convert % into decimals before doing your calculations. **2.** Remember the tip above, you cannot calculate p by square rooting p^2 in this problem since the dominant phenotype is not seen in only homozygous dominant individuals but heterozygotes too!). *Show all work.*

27. Suppose, in a plant population, the red flower allele is completely dominant to the white flower allele. This population is in Hardy-Weinberg Equilibrium for the flower color gene. Within the population of 500 individuals, 25% show the recessive phenotype. How many individuals would you expect to be homozygous dominant and heterozygous for this trait? *Show all work.*

Now, check your work shown and answer to question **26**.

This problem requires you to recognize that individuals with the dominant trait can be either homozygous or heterozygous. Therefore, you cannot simply take the square root of 0.64 to get p. For problems of this type, you must begin with the homozygous recessive group. So . . .

Let p = frequency of the dominant allele (R) and q = frequency of the recessive allele (r)

1. q^2 = frequency of the homozygous recessive = 36% = 0.36. Because $q^2 = 0.36$, $q = 0.6$.

2. Now, $p + q = 1$, so $p = 0.4$.

3. Notice that this problem asks for the *frequency of the dominant allele* (p), not the frequency of the homozygous dominant individuals (p^2). So, you are done . . . **the frequency of the dominant allele = 40%.**

Now, check your work shown and answer to question **27**.

Let p = frequency of the dominant allele (R) and q = frequency of the recessive allele (r).

1. q^2 = frequency of the homozygous recessive = 25% = 0.25. Since $q^2 = 0.25$, $q = 0.5$.
2. Now, $p + q = 1$, so $p = 0.5$.
3. Homozygous dominant individuals are RR or $p^2 = 0.25$, and they will represent (0.25)(500) = **125 individuals**.
4. The heterozygous individuals are calculated from $2pq$ = (2)(0.5)(0.5) = 0.5, and in a population of 500 individuals will be (0.5)(500) = **250 individuals**.

28. For a locus with two alleles (V and v) in a population at risk from an infectious neurodegenerative brain disease, 16 people had a genotype VV, 92 had a genotype Vv, and 12 had a genotype vv. Is this population evolving? **Explain**. ***Show all work.*** *(Check your answers by going to the Ch.23.2* ***Concept Check Question #3*** *in Appendix A of your textbook)*