Active Learning Exercise 8 Mendelian Genetics & the Chromosomal Basis of Inheritance Reference: Chapter 14-15 (Biology by Campbell/Reece, 8th ed.)

- Note: In addition to the genetics problems on ALE 8, you should be <u>able</u> to solve (using Punnett Squares where appropriate) the genetic problems and self quiz questions at the end of chapters 14 and 15, and all other genetic problems discussed in the laboratory and class.
- 1. a.) What is a *gene* as defined by a *Mendelian* geneticist?
 - b.) What is the relationship between a gene and an allele? <u>Support your response with an example</u>.
 - c.) What are *homologous chromosomes*? <u>Support your response with an example</u>.
 - d.) What is the relationship between *alleles* and *homologous chromosomes*? <u>Support your</u> <u>response with an example</u>.
- 2. Distinguish between an organism's *genotype* and its *phenotype*. *Give a good example of each*.
- Use the terms *domina*nt, *recessive* and *alleles* in defining the following. <u>Give an example of each</u>.
 a.) *Heterozygous*
 - b.) *Homozygous*

4. State Mendel's two laws *in your own words*. Include a simple diagram to illustrate each. *Mendel's 1st Law (Law of Segregation)*:

Mendel's 2nd Law (Law of Independent Assortment):

5. Below are the results of a few of Mendel's F₁ monohybrid crosses involving various characteristics in pea plants. Use these results to answer the questions that follow.

Character	Dominant Trait	X	Recessive Trait	F ₂ Generation Dominant : Recessive	Phenotypic Ratio
Height	Tall	х	Dwarf	787 : 277	
Pod color	Green	X	Yellow	428 : 152	
Flower color	Purple	X	White	705 : 224	

a.) Calculate to *two decimal places* the *phenotypic ratio* of the F₂ generation offspring for each cross. Record your responses in the table above.

b.) Discuss how the results of these crosses led Mendel to his first law, the *law of segregation*.

6. Below are the results from Mendel's *dihybrid cross* between *true-breeding* (i.e. homozygous) parent plants differing in two traits, flower color and height.

		Observed	Expected	
Generation	Phenotypes	Phenotypic ratio	Phenotypic ratio	
Parents	(Purple-flowered, tall) x (white-flowered, dwarf)			
F ₁	100% purple-flowered, tall	×/////////////////////////////////////	//////////////////////////////////////	
	630 purple-flowered, tall			
Б	216 purple-flowered, dwarf			
\mathbf{F}_2	202 white-flowered, tall			
	64 white-flowered, dwarf			

a.) Calculate to decimal places and enter in the table above the <u>observed</u> phenotypic ratio for the F_2 . Now enter in the table the expected phenotypic ratio for the F_2 .

- b.) Compare the expected *phenotypic ratio* for the F₂ with that obtained by Mendel in the table above. *Explain why they are different*.
- c.) Discuss how the results of these crosses led Mendel to his second law, the *law of independent assortment*.

d.) What would be the expected phenotypic ratios be for the F_1 and the F_2 if pairs of alleles segregated together (i.e. *dependent assortment*), rather than assorting independently. *Justify your response with a Punnett square of the cross that produces the* F_2 .

7. In *Drosophila*, black body color is recessive to gray body color. A geneticist had three pairs of flies with gray body color designated **A**, **B**, and **C**. The crosses she made and their results are tabulated below.

	F ₁ Generation				
Parental Cross	Gray body	Black body	Ratio		
A x B	109	0	100% Gray- bodied		
A x C	80	28	2.86:1		
ВхС	76	0	100% Gray- bodied		

a.) What are the genotypes of **A**, **B**, and **C**? *Justify your responses. Use Punnett squares where appropriate*.

b.) What are the expected phenotypic and genotypic ratios when flies **A**, **B**, and **C** are crossed with flies having black bodies? <u>Use Punnett squares where appropriate to justify your response</u>.

8. Do laws prohibiting marriages between brothers and sisters make *biological* sense? <u>Explain in</u> <u>terms of recessive traits</u> (e.g. cystic fibrosis, Tay-Sacs disease, phenylketonuria, etc.).

9. a.) What are the chances of obtaining a pure-breeding individual with the dominant trait for all four traits from the following cross: AaBbCcDd x AaBbCcDd? Use the much simpler "*probability method*," rather than a huge Punnett square to answer this problem.

b.) How could you determine if the individual really is *true-breeding* for all four traits? What kind of cross would you use? *Explain*.

10. Complete the table below for the following modes of inheritance. *Complete dominance* (i.e. heterozygotes exhibit the dominant phenotype), *incomplete dominance* (i.e. heterozygotes exhibit a blending of the dominant and recessive phenotypes), *codominance* (heterozygotes express both phenotypes) and *polygenic inheritance* (2 or more genes control a specific phenotype) differ in the amount of variation seen in the offspring and, in the case of polygenic inheritance the, in the number of genes involved.

Mode of Inheritance	possi	ber of ble phenotypes	Possible genotypes (e.g. AA, Aa, etc.)	Possible phenotypes (e.g. Dominant, recessive)	Examples
Complete dominance					
Incomplete dominance					
Codominance					
Polygenic inheritance				Continuous variation of phenotype	

- 11. Imagine a cross involving a pea plant heterozygous at the loci for flower color (white vs. purple) and seed color (yellow vs. green) with a second pea homozygous for flower color (white) and seed color (yellow). What types of gametes will the first pea produce?
 - a.) two gamete types: white/white and purple/purple
 - b.) two gamete types: white/yellow and purple/green
 - c.) four gamete types: white/yellow, white/green, purple/yellow, purple/green
 - d.) four gamete types: white/purple, yellow/green, white/white, and purple/purple
 - e.) one gamete type: white/purple/yellow/green
- 12. Imagine a genetic counselor working with a couple who have just had a child who is suffering from *Tay-Sachs disease*. Neither parent has Tay-Sachs, nor does anyone in their families. Which of the following statements should this counselor make to this couple?
 - a.) "Because no one in either of your families has Tay-Sachs, you are not likely to have another baby with Tay-Sachs. You can safely have another child."
 - b.) "Because you have had one child with Tay-Sachs, you must each carry the allele. Any child you have has a 50% chance of having the disease."
 - c.) "Because you have had one child with Tay-Sachs, you must each carry the allele. Any child you have has a 25% chance of having the disease."
 - d.) "Because you have had one child with Tay-Sachs, you must both carry the allele. However, since the chance of having an affected child is 25%, you may safely have thee more children without worrying about having another child with Tay-Sachs."
 - e.) "You must both be tested to see who is a carrier of the Tay-Sachs allele."
- 13. *Albinism* in humans occurs when both alleles at a locus produce defective enzymes in the biochemical pathway leading to melanin. Given that heterozygotes are normally pigmented, which of the following statements is/are correct?
 - a.) One normal allele produces as much melanin as two normal alleles.
 - b.) Each defective allele produces a little bit of melanin.
 - c.) Two normal alleles are needed for normal melanin production.
 - d.) The two alleles are codominant.
 - e.) The amount of sunlight will not affect skin color of heterozygotes.
- 14. In humans, alleles for *dark hair* are genetically dominant while alleles for *light hair* are recessive. Which of the following statements is/are most likely to be correct?
 - a.) Dark hair alleles are more common than light hair alleles in all areas of Europe.
 - b.) Dark hair alleles are more common than light hair alleles in southern Europe but not in northern Europe.
 - c.) Dark hair alleles are equally common in all parts of Europe.
 - d.) Dark hair is dominant to light hair in southern Europe but recessive to light hair in northern Europe.
 - e.) Dark hair is dominant to light hair in northern Europe but recessive to light hair in southern Europe.
- 15. Envision a family in which the grandfather, age 47, has just been diagnosed with *Huntington's disease*, an *autosomal dominant* neurological disorder where symptoms appear in midlife and a slow and sad death a few years later. His daughter, age 25, now has a 2-year-old baby boy. No one else in the family has the disease. What is the probability that the daughter will contract the disease?
 a.) 0%
 b.) 25%
 c.) 50%
 d.)75%
 e.) 100%

- 16. Imagine that you are the daughter in the family with *Huntington's disease* described in the previous question. You had been planning on having a second child. What would <u>you</u> do?
 - a.) I would not be genetically tested. I would have a second child and trust that I can handle whatever my life circumstances are.
 - b.) I would not be genetically tested. I would not have a second child.
 - c.) I would have myself genetically tested. If I carry the gene, I would not have a second child and I would have my 2-year-old child tested.
 - d.) I would have myself genetically tested. If I do carry the gene, I would not have a second child but I would not have my 2-year-old baby boy tested.
 - e.) I would not be genetically tested and I would adopt a second child.
- 17. Imagine a locus with four different alleles for fur color in an animal. The alleles are named D^a, D^b, D^c, and D^d. If you crossed two heterozygotes, D^aD^b and D^cD^d, what genotype proportions would you expect in the offspring?
 - a.) $25\% D^{a}D^{c}$, $25\% D^{a}D^{d}$, $25\% D^{b}D^{c}$, $25\% D^{b}D^{d}$
 - b.) $50\% D^{a}D^{b}$, $50\% D^{c}D^{d}$
 - c.) 25% D^aD^a, 25% D^bD^b, 25% D^cD^c, 25% D^dD^dD^cD^d
 - d.) $50\% D^{a}D^{c}$, $50\% D^{b}D^{d}$
 - e.) 25% D^aD^b, 25% D^cD^d, 25% D^cD^c, 25% D^dD^d
- 18. Suppose that you are a genetic counselor. A concerned young couple, Jack and Jill, visits you because <u>each had a brother that died</u> of cystic fibrosis, the most common lethal genetic disease amongst Caucasians in the United States, affecting 1 in 2500 births. Disease symptoms, and ultimately death, are due to the accumulation of mucus in the pancreas, intestinal tract, and lungs in individuals that are homozygous recessive for the cystic fibrosis allele.
 - a.) What are the *genotypes* of Jack's parents? Jill's parents? Justify your response.
 - b.) What are the chances that Jack is a *carrier* of cystic fibrosis? Jill? <u>Justify your responses</u>.
 (<u>Hint</u>: the fact that we know that Jack and Jill do <u>not</u> have cystic fibrosis must be considered!!)

c.) What are the chances that their first child will have cystic fibrosis? Be unaffected by cystic fibrosis? *Justify your responses*.

- 19. Explain what each of the following is, how they work, what they are used for, and the risks involved:
 - a.) Amniocentesis:

b.) Chorionic villi sampling:

20. After eight years of married life, during which time she had failed to become pregnant, Mrs. X met and fell in love with Mr. Y. During the ensuing five years three children were born. In the meantime the persons involved had tried to come to an understanding and wished to determine which of the two men was responsible for each child. The blood types of those involved were determined, with the results indicated below.

			_		Possible Offspring	
	Blood Group	Blood Type ¹			Blood Group	Blood Type ¹
Mr. X	0	MN	1 [
Mrs. X	0	MN		Mr. and Mrs. X		
Mr. Y	Α	Ν				
First child	0	MN				
Second child	0	М		Mr. Y and Mrs. X		
Third child	Α	Ν				

Blood types M, N, and MN are controlled by alleles M and N, the heterozygote being MN, an example of codominance.

- a. Record in the table above all possible phenotypes that could be produced by Mr. and Mrs. X, and by Mr. Y and Mrs. X
- b. What do conclude as to the paternity of each child? Justify your responses.

The Chromosomal Basis of Inheritance (Ref. Chapter 15)

- 21. Why is a *recessive sex-linked allele* always expressed in human males, but not always expressed in females?
- 22. a.) Make a diagram of an unduplicated chromosome containing two different genes, gene *A* and gene *B*. Use this diagram to explain *gene locus* and *gene linkage*.

b.) Why does *gene linkage* interfere with *independent assortment*?

- c.) Suppose the genotype of an organism is *AaBb*. If the genes *A* and *B* are <u>linked</u> on one chromosome (as in part a, above), and their recessive alleles, *a* and *b*, are on the other homologue. (You should be able makes a diagram of meiosis to show how the gametes would be produced in the following questions.)
 - i.) How many different genetic types of gametes would be produced in the *absence of* <u>crossover</u>? List them:
 - ii.) If <u>crossover occurs</u> between the loci of the linked genes A and B involving the organism above (AaBb), how many different genetic types of gametes could be produced? <u>List them</u> and estimate the approximate ratio of the different genetic types of gametes and <u>explain why those produced as a result of crossover occur much less frequently</u>:

iii.) Suppose the genotype of an organism is *AaBb*. How many different genetic types of gametes would be produced if Genes *A* and *B* are <u>not linked</u>?
 List them:

- 23. How does crossover in meiosis affect gene linkage?
- 24. a.) How does the distance between two linked genes affect the crossover frequency between those genes?
 - b. How is crossover frequency used in mapping the location of genes on chromosomes?
- 25. Human traits that are controlled by a single gene are usually inherited according to Mendel's Laws. Some defective genes causing disorders are dominant to the normal gene while others are recessive to the normal gene. Some of these genes are located on the autosomal chromosomes, some on the X chromosome, and none are known on the Y chromosome. For each mode of inheritance listed below give two human examples and a symptom for each.
 - a.) Autosomal recessive disorders:
 - b.) Autosomal dominant disorders:
 - c.) X-linked recessive disorders:
 - d.) *Y-linked inheritance*: There is no clear evidence for genetic loci on the Y chromosome other than those involved with gender determination in males. If a Y-linked disorder were discovered, what would be the gender(s) of those affected? Why?

Would it be dominant or recessive? Why?

26. a.) Make a labeled diagram to show how *nondisjunction* during meiosis I could result in an extra number 21 chromosome in the resulting egg. What is the name of resulting disorder if this egg is fertilized by a normal sperm and ultimately develops into a child?

b. Make a labeled diagram to show how *nondisjunction* during meiosis II could result in an extra number 21 chromosome in the resulting egg.

- 27. Triploid species are usually sterile (unable to reproduce) whereas tetraploids are often fertile. Which of the following are likely good explanations of these facts?
 - a.) In mitosis, some chromosomes in triploids have no partner at synapsis, but chromosomes in tetraploids do have partners.
 - b.) In meiosis, some chromosomes in triploids have no partner at synapsis, but chromosomes in tetraploids do have partners.
 - c.) In mitosis, some chromosomes in tetraploids have no partner at synapsis, but chromosomes in triploids do have partners.
 - d.) In meiosis, some chromosomes in tetraploids have no partner at synapsis, but chromosomes in triploids do have partners.

- 28. The chromosome in the figure to the right is made up of two chromatids, joined at the centromere. The two chromatids were formed by...
 - a.) DNA replication of a single chromatid.
 - b.) fertilization, bringing together a maternal and a paternal chromatid.
 - c.) one double helix of DNA.
 - d.) nondisjunction during anaphase I of meiosis.
 - e.) nondisjunction during anaphase of mitosis.
- 29. Circle whether each statement is true or false and then explain clearly your reasoning.
 - a.) <u>*True*</u> or <u>*False*</u>: A h*eterozygous* parent is equally likely to contribute the dominant or the recessive allele to an offspring.
 - b.) *True* or *False*: Over time, *dominant alleles* will tend to increase in frequency in a population.
 - c.) <u>*True*</u> or <u>*False*</u>: Dominant alleles prevent recessive alleles from being expressed in a heterozygote.

