BIOL 109L
Laboratory five Fall 2018
Understanding Genetics and Inheritance

Student full name: _____

Introduction:

Chromosomes are like a set of books that provide instructions for building and operating an individual. You already know the alphabet used to write those books: the four letters A, T, G, and C, for the four nucleotides in DNA—adenine, thymine, guanine, and cytosine. During the lectures of this course we investigate "words" that can be made from those letters, and "sentences" that can be made from the words.

The nature of information represented by the sequence of nucleotides in a DNA molecule occurs in hundreds or thousands of units called genes.

How do genes contribute to the similarities and differences between parents and their children? This question can be divided into two parts:

- How do genes influence our characteristics?
- How are genes inherited by children from their parents?

During this lab activity we will address many genetic scenarios which will answer both of these questions to help you develop an understanding of human genetics and inheritance.

This "dry lab" on Genetics and Inheritance was first developed as an educational tool by Drs. Ingrid Waldron and Jennifer Doherty, University of Pennsylvania.

It is provided by *Serendip Studio* and was initially founded by Bryn Mawr College and the Bryn Mawr College Trust in Diversity and Exchange (TIDE), which was launched in 1990 at Massachusetts Institute of Technology by John R. Bemis. This wonderful educational resource was made possible by support from the Howard Hughes Medical Institute through an Undergraduate Science Education program grant.

How do genes influence our characteristics?

To begin, we will review and extend an analysis of the genetics of albinism.

Genotype	→	**Protein**	→	**Phenotype** (characteristics)	
AA	→	Normal enzyme that makes melanin, the pigment that gives color to skin and hair	→	Normal skin and hair color	
Aa	→	Normal enzyme that makes melanin and defective enzyme that does not make melanin	→	Normal skin and hair color	
aa	→	Defective enzyme that does not make melanin	→	Very pale skin and hair color (albino)	

1) Why does each genotype have two letters? What do these two letters represent?

- A person is **homozygous** for a gene if both alleles for that gene are the same.
- A person is **heterozygous** for a gene if the two alleles are different.

2) Circle the genotypes in the chart that are homozygous and put a square around the genotype that is heterozygous.

3) For a person who is heterozygous **Aa**, the phenotype is:

a. the same as the phenotype of a homozygous **AA** person.
b. the same as the phenotype of a homozygous **aa** person.
c. Intermediate – halfway between the phenotypes of the two homozygous individuals.

4) Explain why a person with the **AA** or **Aa** genotype has normal skin and hair color, but a person with the **aa** genotype is albino.

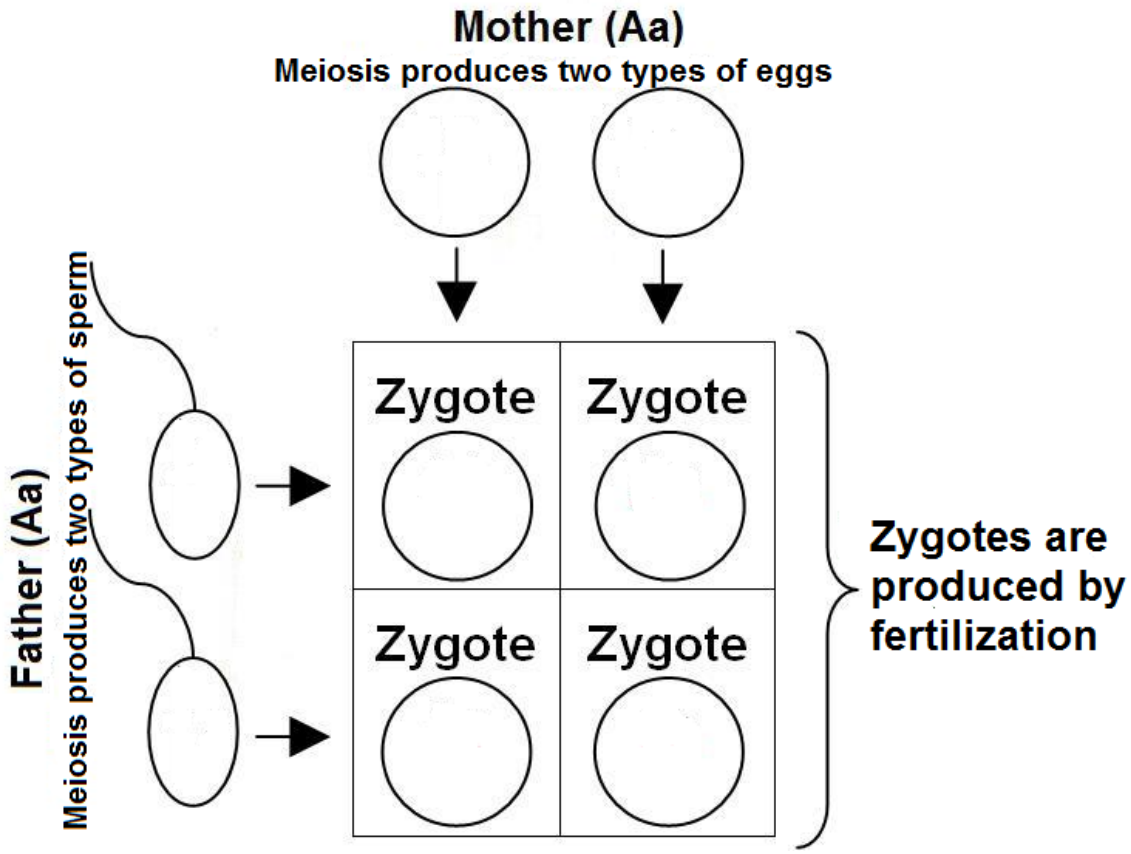
This example illustrates how a **dominant** allele (**A**) can determine the phenotype of a heterozygous person, and a **recessive** allele (**a**) does not affect the phenotype of a heterozygous person.

5) Based on the example on the previous page, fill in each blank with dominant or recessive.

- A heterozygous person has the same phenotype as a person who is homozygous for the _____ allele.
- A _____ allele does not affect the phenotype of a heterozygous person.
- A _____ allele is represented by a capital letter.

How does a child inherit genes from his or her mother and father?

In this chart, record the allele in each type of egg produced by meiosis, and record the allele in each type of sperm produced by meiosis

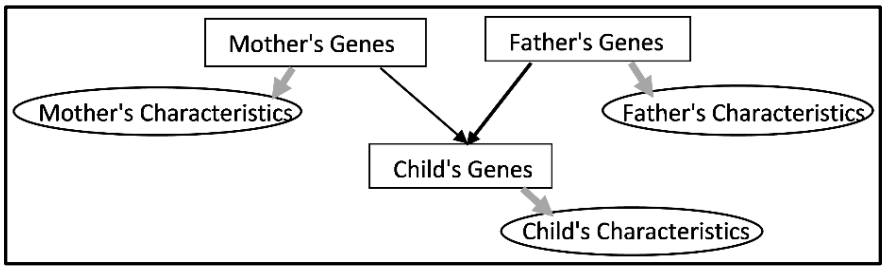


6) Biologists use a similar chart to analyze inheritance However, biologists omit much of the detail and use a simplified version called a **Punnett Square**

	A	a
A	**AA**	**Aa**
a	**Aa**	**aa**

In this Punnett square:
What fraction of the mother's eggs have an a allele? _____
What fraction of the father's sperm have an a allele? _____
What fraction of this couple's children would you expect to be aa?_____Explain your reasoning

This flowchart shows how genes contribute to the similarities between parents and their children.



7) What biological processes are represented by the thin black arrows? In other words, which biological processes transmit genes from parents to their children?

8). Explain how genes influence a person's characteristics (represented by the fatter gray arrows). Include the words alleles and protein in your answer.

9) Explain how genes contribute to the similarities between parents and their children

Coin Flip Genetics

The way genes behave during meiosis and fertilization can be modeled by flipping coins. You and a partner will be heterozygous **Aa** parents. When you flip your coins, heads will represent the **A** allele and tails will represent the **a** allele. This table explains how you will model meiosis and fertilization.

Biological Process	How You Will Model This
Meiosis in an **Aa** parent produces gametes. Each gamete has an equal probability of having an **A** allele or an **a** allele.	Flip your coin and check for heads up vs. tails up. This represents the equal probability that a gamete will have an **A** allele or an **a** allele.
Fertilization of an egg by a sperm produces a zygote. Each gamete contributes one allele to the genotype of the child that develops from the zygote.	You and your partner each flip a coin to determine the alleles that the egg and sperm contribute to the zygote that develops into a child.

When you flip a coin, half the time you will get heads and half the time you will get tails. If you and your partner each flip a coin, the probability of getting two tails is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$.

10) What is the probability that a child of two heterozygous parents will be albino?

_____

11) Explain your reasoning.

12) If two **Aa** parents have a family of 4 children, on average, the predicted number of children with each genotype is _____ **AA**, _____ **Aa**, and _____ **aa**.

	A	**a**
A	**AA**	**Aa**
a	**Aa**	**aa**

Work with a partner to make a family of four coin flip children. Each of you should flip your coin to determine the genotype of the first child. Record the genotype in the box for the first child in the first family in the table below. Make three more pairs of coin flips and record the genotypes for the second, third and fourth children in this family.

Repeat this procedure three times to determine the genotypes for three more families of four children each. Record your results in the table. Complete the last three columns for each family of coin flip children. Calculate the totals for each genotype.

Genotypes of coin flip children produced by two heterozygous (Aa) parents

	Genotype for each child				Number with each genotype		
	1st	2nd	3rd	4th	AA	Aa	aa
First family of 4 children							
Next family of 4 children							
Next family of 4 children							
Next family of 4 children							
Totals							

13) How many of your coin flip families had exactly the predicted numbers of **AA**, **Aa** and **aa** coin flip children? 0 ___ 1 ___ 2 ___ 3 ___ 4 ___

14) Why didn't all of your coin flip families have exactly the predicted number of children with each genotype?

15) In your coin flip families, did the genotype produced by the first pair of coin flips have any effect on the genotype produced by the second pair of coin flips? yes ___ no ___

In real families the genotype of each child depends on which specific sperm fertilized which specific egg. This is not influenced by what happened during the fertilizations that resulted in any previous children. Therefore, the genotype of each child is independent of the genotype of any previous children.

16) Think about real families where both parents are heterozygous **Aa**. If the first child is albino, what is the probability that the second child will be albino? 0% ___ 25% ___ 50% ___ 75% ___

17). Explain your reasoning.

18) Fill in each blank in these sentences with the best matches from the list below.

A Punnett square can accurately predict _____ and _____.
A Punnett square can <u>not</u> accurately predict _____ or _____.

a. the average percent albino in a large sample of the children of two heterozygous parents
b. the percent albino in a specific family of children of two heterozygous parents
c. the probability that the next child in a family will be albino
d. whether the next child in a family will be albino

19) Explain your reasoning.

20) The Punnett square for two heterozygous **Aa** parents predicts that one quarter of the children will be albino. Why aren't one quarter of the children in your school albino?

The Genetics of Skin Color

Skin color is influenced by other genes besides the gene that can result in albinism. In this section, you will learn how multiple genes and the environment influence skin color. You will also learn why parents and children often, but not always, have similar skin color.

To begin we will consider two alleles of a gene that influences the amount of melanin produced in skin cells. Notice that, for this gene, a heterozygous individual has an intermediate phenotype, halfway between the two homozygous individuals. This is called **incomplete dominance**.

Genotype	**Phenotype** (skin color)
BB	dark brown
Bb	light brown
bb	tan

21) How does incomplete dominance differ from a dominant-recessive pair of alleles? (Hint: Think about the phenotypes of heterozygous individuals.)

22) For each pair of parents listed in this table, indicate what skin color or colors you would expect to observe in their children.

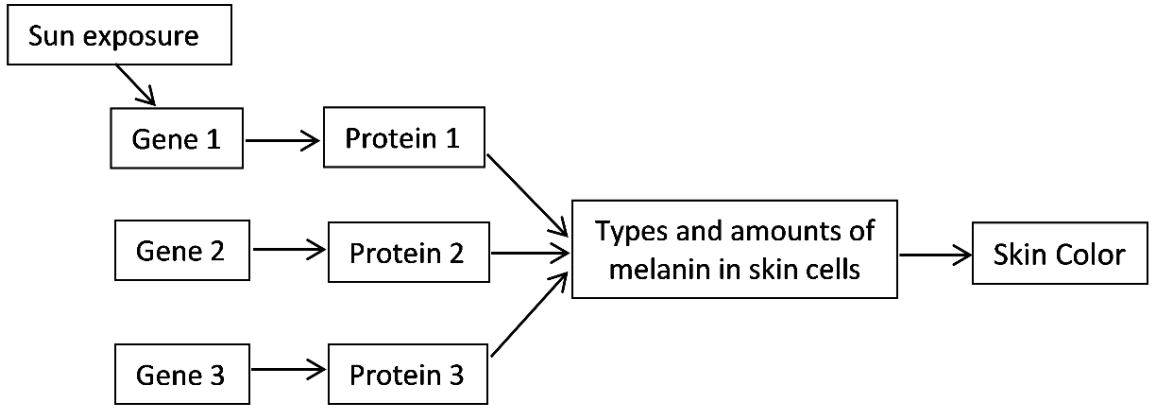
Parents	Expected Skin Color or Colors for Their Children
2 **BB** parents with dark brown skin	
2 **Bb** parents with light brown skin	
2 **bb** parents with tan skin	

23) To test whether your expectations are correct, show the Punnett squares for each pair of parents in this table and write in the skin colors of their children.

2 **BB** parents with dark brown skin	2 **Bb** parents with light brown skin	2 **bb** parents with tan skin

24) Notice that, in general, parents and children tend to have similar skin color, but there are exceptions. Explain how two parents with light brown skin could have a child with dark brown skin and a child with tan skin.

Obviously, people have many different skin colors, not just dark brown, light brown, or tan. This wide variety of skin colors results from the effects of multiple genes and also environmental factors such as sun exposure. This flowchart shows how genetic and environmental factors influence skin color.



25) This flowchart is based on the scientific findings that are summarized in A-D below. Use the letter for each scientific finding to label the part or parts of the flowchart that represent this scientific finding.

A. Different skin colors result from differences in the types and amounts of the pigment melanin in skin cells.

B. Several different proteins influence the production and processing of melanin molecules in skin cells. Different versions of these proteins result in different types and amounts of melanin in skin cells.

C. For each protein, there is a gene that gives the instructions to make that protein. Different alleles of a gene result in different versions of the protein.

D. Exposure to sunlight can change the activity of one or more of these genes and increase the amount of melanin in skin cells.

26) This information indicates that the table on the top of the previous page is oversimplified. Multiple factors influence skin color, so two people who both have the **Bb** genotype can have different skin colors. Suppose that two people both have the **Bb** genotype, but one has darker skin color than the other. Give two possible reasons for this difference in skin color.

Skin color is a good example of the complexity of genetics. Many pairs of alleles do not show simple dominant/recessive effects on phenotype. Most of our characteristics are influenced by multiple genes and the environment. For example, your height, weight, blood pressure, and risk of diabetes are each influenced by multiple genes and the environment.

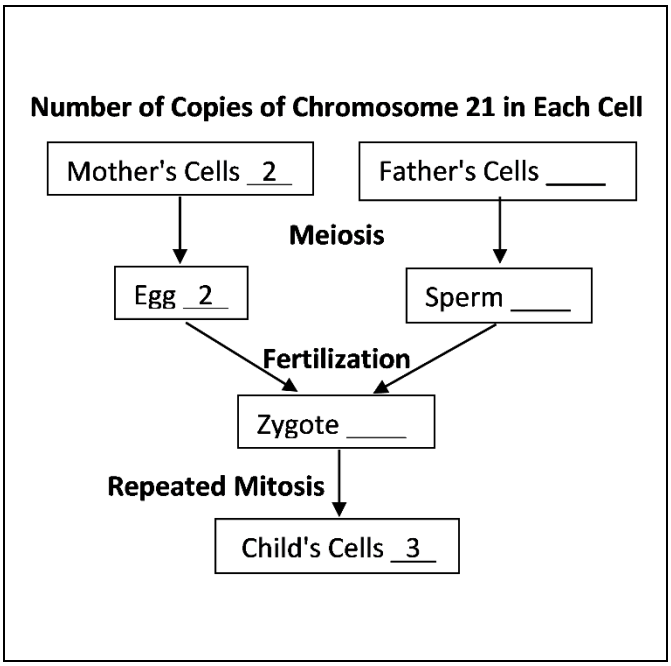
Some genetic conditions are not inherited.

Sometimes, mistakes occur in the biological processes we have been discussing. This can result in a child having different characteristics than either of his or her parents.

Down syndrome is caused by three copies of chromosome 21 in each cell. This chart shows one way that two normal parents can have a child with Down syndrome.

27) Fill in the blanks to show normal meiosis in the father and the results of fertilization.
Use an * to mark the arrow that represents the biological process where the mistake occurred that resulted in the child's Down syndrome.

Notice that the child has a genetic condition that was not inherited.



Achondroplasia is another example of a genetic condition that often is not inherited. Achondroplasia is a type of dwarfism with very short legs and arms. Achondroplasia is caused by the **D** allele of a gene that provides the instructions to make a protein that regulates bone growth.

Genotype	→	**Protein**	→	**Phenotype (characteristics)**
dd	→	Normal regulator protein that stops bone growth after a person has reached adult height	→	Person has normal height and mortality risk.
Dd	→	Both normal regulator protein and overactive regulator protein	→	Person is very short and has somewhat elevated mortality risk; person has achondroplasia.
DD	→	Overactive regulator protein that excessively inhibits bone growth	→	Abnormalities are so severe that fetus dies before birth or baby dies shortly after birth.

28) The **D** allele is considered dominant because a heterozygous person is very short and has achondroplasia. However, the **D** allele is not completely dominant. If the D allele were completely dominant, then **DD** and **Dd** individuals would have the same phenotype. Describe one difference in the characteristics of **DD** and **Dd** individuals.

29) A child has achondroplasia, so his genotype is **DD** ___ **Dd** ___ **dd** ___.

30) Neither parent has achondroplasia, so each parent has the genotype, **DD** ___ **Dd** ___ **dd** ___.

31) Neither parent had achondroplasia. Nevertheless, the **D** allele was present in the sperm that fertilized the egg to produce the zygote that developed into the child. During sperm production, a

mistake in DNA replication resulted in a mutation that changed the **d** allele to a ____ allele.

32) Based on the frequency of dwarfs among the people you have seen in your lifetime, what genotype do most people have?

The End