Genetics

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This Student Handout, an alternative module that uses model chromosomes, additional modules, and Teacher Preparation Notes with background information are available at http://serendip.brynmawr.edu/sci_edu/waldron/#genetics

We all know that children tend to resemble their parents in appearance. Parents and their children tend to have similar appearance because children inherit genes from their parents and these genes influence characteristics such as skin and hair color.

How do genes influence our characteristics?

A **gene** is a segment of a DNA molecule that gives the instructions for making a protein. Different versions of the same gene are called **alleles**, and different alleles give the instructions for making different versions of a protein. This table gives an example.

Gene in DNA	→	Protein
A allele provides instructions to make normal enzyme	→	Normal enzyme for producing melanin, a pigment molecule that gives color to our skin and hair
a allele provide instructions to make defective enzyme	→	Defective enzyme that cannot make melanin

Each cell in your body has two copies of each gene (one inherited from your mother and one inherited from your father). The two copies of each gene may have the same alleles or different alleles. These alleles determine which version(s) of the protein are made by your body's cells, and the different versions of the protein can result in different characteristics. In other words, **genotype** (the genetic makeup of a person) determines which version(s) of the protein are made and the proteins influence the **phenotype** (the observable physical and physiological characteristics of a person).

Genotype	→	**Protein**	→	**Phenotype** (characteristics)	
AA or **Aa**	→	Enough normal enzyme to make melanin in skin and hair	→	Normal skin and hair color	
aa	→	Defective enzyme for melanin production	→	Very pale skin and hair color (albino)	

If both copies of a gene have the same allele, the person is **homozygous** for that gene. If the two copies of a gene have different alleles, the person is **heterozygous**.

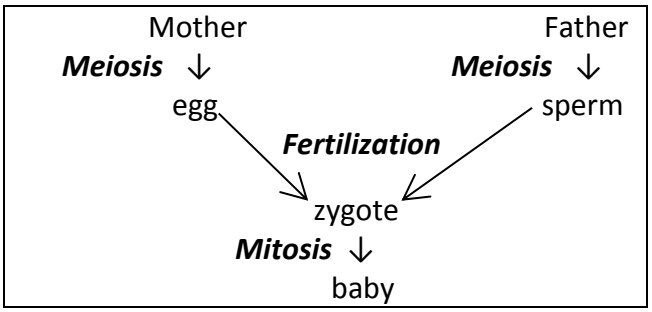
Often, in a heterozygous individual a **dominant** allele determines the observable characteristic and the other **recessive** allele does not affect the phenotype. Thus, a heterozygous person has the same phenotype as a person who is homozygous for the dominant allele. In our example, the **A** allele is dominant because it codes for normal, functional enzyme and, even in a heterozygous individual, there is enough of this normal, functional enzyme to produce enough melanin to result in normal skin and hair color. The **a** allele is recessive because it codes for a non-functional enzyme which does not affect skin or hair color in a heterozygous individual.

1. Circle the genotypes in the table that are homozygous. Explain how the two different homozygous genotypes result in different phenotypes.

2. What are the two different genotypes for the albinism gene that result in the same phenotype?
- Explain how two people with different genotypes can have the same phenotype.

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

As you know, each gene is a part of a DNA molecule. Each DNA molecule is contained in a **chromosome**. You will see that we can understand how a baby inherits genes from his or her mother and father by understanding how the gene-carrying chromosomes move during **meiosis** to form gametes and **fertilization** to form the zygote that develops into a baby.

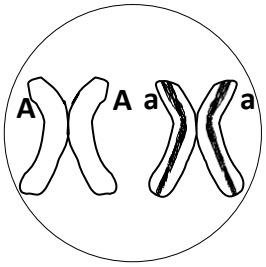


Inheritance of Albinism

To learn more about how genetic traits are inherited, we will start with a specific question:

> If each parent has one **A** allele and one **a** allele (i.e. both parents are **Aa**), what different combinations of **A** and/or **a** alleles could be observed in the children of these parents?

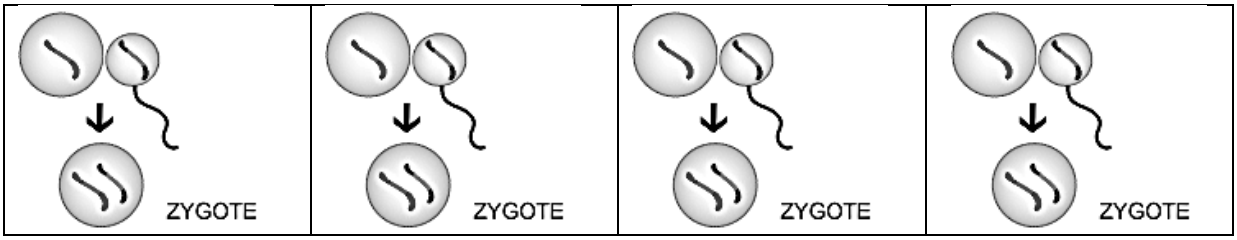
3. Begin by completing this diagram to show how meiosis produces sperm in the father.



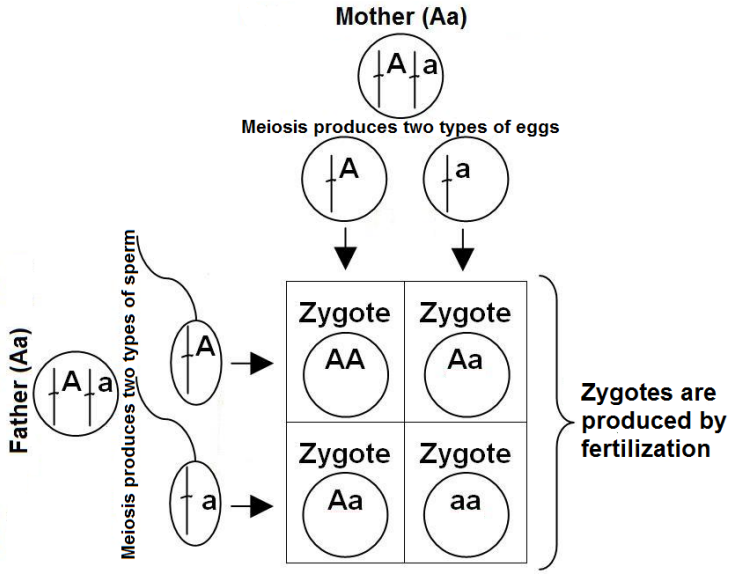
What is the genetic makeup of the different types of sperm that an **Aa** father can produce? ____ ____

What is the genetic makeup of the different types of eggs that an **Aa** mother can produce? ____ ____

4. Complete these diagrams to show how fertilization of each different type of egg by each different type of sperm produces different combinations of alleles in the different zygotes. Label the allele on each chromosome.



This chart combines the results of meiosis and fertilization for an **Aa** mother and **Aa** father to show the possible genetic combinations in the zygotes. Each zygote undergoes repeated mitosis to become a child, so the child has the same genetic makeup as the zygote.



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

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

5. In this Punnett square, circle each symbol which represents the genetic makeup of a gamete produced by the heterozygous mother or father.
- Use an * to indicate the genotype of each zygote.

6. The genotypes of the zygotes in the Punnett square represent the possible genotypes of the children of this couple. Explain why each child will have the same genotype as the zygote he developed from.

7. For a heterozygous mother, what fraction of her eggs have an **a** allele? _____
- For a heterozygous father, what fraction of his sperm have an **a** allele? _____
- What fraction of this couple's children would you expect to be **aa**? _____ Explain your reasoning.

8. Complete this Punnett square for two parents who are homozygous **AA**.	Complete this Punnett square for two parents who are homozygous **aa**.	Complete this Punnett square for a mother who is heterozygous **Aa** and a father who is homozygous **aa**.

9. For each of the four Punnett squares on this page, indicate the phenotypes of each parent and each child (N = normal skin and hair color or O = albino).

Notice that most of the children have the same phenotypic characteristic as one or both parents. Circle the only example of a child who has a phenotypic characteristic that is not observed in either parent.

Coin Toss Genetics

The way genes behave during meiosis and fertilization can be simulated using two-sided coins, where heads represent the dominant allele (**A**) that results in normal skin and hair color and tails represent the recessive allele (**a**). Suppose a parent is heterozygous (**Aa**). Then, tossing a coin and checking for heads up vs. tails up represents the 50-50 chance that an egg or sperm produced by meiosis will include an **A** allele or an **a** allele. To simulate a mating between two heterozygous (**Aa**) parents, two students will each toss a coin and the result of this pair of coin tosses will indicate the alleles contributed by an egg and a sperm to the baby that results from that mating.

- Find someone to "mate" with.
- Each of you will toss your coin, and this pair of coin tosses will indicate the pair of alleles in the first child produced by a mating of two heterozygous (**Aa**) parents. Make three more pairs of coin tosses to determine the genotypes for the second, third and fourth children in this family. Record your results in the row labeled "first family of 4 children" in this table.

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

	Result for each coin toss				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							
Predictions based on Punnett square					1/4 = 25%	2/4 = 50%	1/4 = 25%
Class data – Percents (Total # children = ____)							

- Repeat this procedure three times to determine the genotypes for three more families of four children and record your results in the table.
- Add up your results to determine the total number of children from your coin tosses who had **AA**, **Aa**, and **aa** genotypes. Give your numbers to your teacher to include in the class data.

Notice that, even if a family already has an **aa** child or an **AA** child, a later child may have the same genotype as an earlier child. This occurs because the genetic makeup of each child depends on which specific sperm fertilized which specific egg, and this is independent of what happened during fertilizations that resulted in previous children. Thus, for each child, the probability of an **aa** genotype and an albino phenotype is 1/4, independent of the genotype and phenotype of any previous children.

1. Two **Aa** parents have two children with the **Aa** genotype and one child with the **aa** genotype. If these parents have a fourth child, what is the probability that the fourth child will have the **AA** genotype?

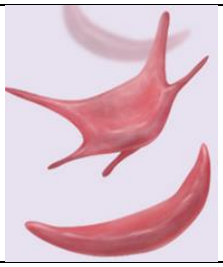
2. For each family of 4 children produced by your coin toss matings, compare your results with the predictions from the Punnett square. Use a check mark to indicate any family that has exactly the numbers of **AA**, **Aa**, and **aa** genotypes predicted by the Punnett square. Explain why some of the families do not have exactly the number of **AA**, **Aa**, and **aa** genotypes predicted by the Punnett square.

3. Your teacher will give you the class data to enter in the last line of the table on the previous page. Are the percents of each genotype in the class data similar to the predictions of the Punnett Square?

As a result of random variation in which particular sperm fertilizes which particular egg to form a zygote, the proportions of each genotype and phenotype vary in different families, and the observed proportions of each genotype and phenotype often do not match the predictions of the Punnett square. The random variation observed in small samples usually averages out in large samples. Therefore, the results for a large number of children from multiple pairs of parents with the same genotypes are usually close to the predictions of the Punnett Square.

Genetics of Sickle Cell Anemia

Some alleles of certain genes can cause disease. An example is the gene for hemoglobin, the protein that carries oxygen in red blood cells. One allele codes for normal hemoglobin, and another allele codes for sickle cell hemoglobin. When a person is homozygous for the sickle cell allele, this causes a disease called sickle cell anemia. Sickle cell hemoglobin tends to clump into long rods that cause the red blood cells to assume a sickle shape or other abnormal shapes, instead of the normal disk shape.

Genotype	→	**Hemoglobin in Red Blood Cells**	→	**Shape of Red Blood Cells**
Homozygous for alleles for normal hemoglobin	→	Normal hemoglobin dissolves in cytosol 	→	
Homozygous for alleles for sickle cell hemoglobin	→	Sickle cell hemoglobin tends to clump in long rods 	→	

1. Normal disk-shaped red blood cells can barely squeeze through the capillaries (the tiniest blood vessels). What problems might be caused by red blood cells that are sickle-shaped or have other abnormal shapes?

2. Most children with sickle cell anemia have parents who do not have sickle cell anemia. Explain how a person can inherit sickle cell alleles from parents who do not have sickle cell anemia. Is the sickle cell allele dominant (**S**) or recessive (**s**)? Explain your reasoning. Include a Punnett Square in your answer.

3. Rasheda and Craig do not have sickle cell anemia, but their first child has sickle cell anemia. What is the probability that their second child will have sickle cell anemia? Explain your reasoning.

Read the information in the box and then answer the questions on the bottom half of the page.

The sickle cell allele illustrates some common complexities of genetics that we have ignored thus far.

People who are heterozygous for the sickle cell allele almost never experience the symptoms of sickle cell anemia that are observed in people who are homozygous for the sickle cell allele. These symptoms include pain and organ damage due to blocked circulation and anemia (low red blood cell levels) due to more rapid breakdown of red blood cells.

Because people who are heterozygous for the sickle cell allele almost always have normal health, the allele for normal hemoglobin is generally considered to be dominant and the allele for sickle cell hemoglobin is generally considered to be recessive. However, a heterozygous person does not have exactly the same phenotype as a person who is homozygous for the allele for normal hemoglobin.

People who are heterozygous for the sickle cell allele are less likely to develop severe malaria than people who are homozygous for the normal hemoglobin allele. Malaria is caused by a parasite that infects red blood cells. The red blood cells of heterozygous individuals have both sickle cell and normal hemoglobin, and the presence of sickle cell hemoglobin inhibits the reproduction of the malaria parasite. Therefore, malaria infections are generally less severe in individuals who are heterozygous for the sickle cell allele.

Malaria infections are common in many tropical countries where there are lots of the type of mosquitoes that transmit the malaria parasite. In areas where malaria is widespread, people who are heterozygous for the sickle cell allele are less likely to become seriously ill and die. Because the sickle cell allele contributes to increased survival of heterozygous individuals, this allele became relatively common in regions like West Africa where malaria is common. Since African-Americans are descended from populations in which the sickle cell allele was relatively common, African-Americans have relatively high rates of the sickle cell allele (approximately 8% are heterozygous for this allele and 0.16% are homozygous).

4. Explain how the hemoglobin gene illustrates the following generalization:

A single gene often has multiple phenotypic effects.

5. Often, when geneticists investigate a pair of alleles, neither allele is completely dominant or completely recessive. In other words, the phenotype of a person who is heterozygous for these two alleles is different from the phenotypes of people who are homozygous for either allele. Explain how this general principle is illustrated by the sickle cell and normal alleles for the hemoglobin gene.