

Genetics

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We all know that children tend to resemble their parents in appearance. Parents and children generally have similar eye color, hair texture, height and other characteristics because children inherit **genes** that control specific characteristics from their parents.

Where are genes found in our bodies?

Researchers have shown that genes are parts of DNA molecules, and DNA molecules are packaged in chromosomes in the nucleus of each cell in our body.

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

When we talk about genes being inherited from one generation to the next, we are really talking about how the gene-carrying chromosomes behave during meiosis and fertilization. For example, if you think about how the mother's and father's chromosomes behave during meiosis and fertilization, you can understand why the zygote that becomes a baby has two copies of each gene, one copy from the mother and one copy from the father.

To explain how a zygote gets one copy of each gene from the mother and one copy from the father, show what happens during meiosis and fertilization, using diagrams or sockosomes.

Inheritance of Albinism

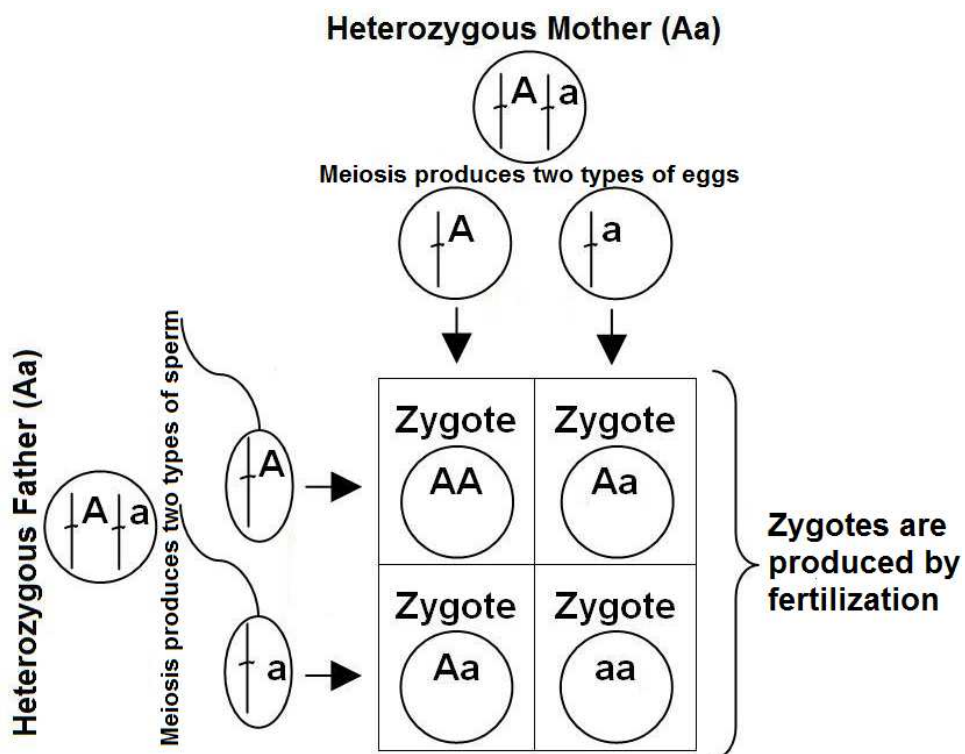
To learn more about how genetic traits are inherited, we will consider a specific example -- the gene that controls whether or not a person can produce the pigment **melanin** which contributes to the color of skin, eyes and hair. Some people have the hereditary condition, **albinism**; they are not able to produce melanin and have little or no pigment in their skin and hair. Two different versions of the same gene are called **alleles**. One allele of this gene codes for melanin production and normally pigmented skin and hair; it is symbolized by **A**. Another allele of this gene codes for albinism; it is symbolized by **a**.



¹ Teachers are encouraged to copy this student handout for classroom use. A Word file (which can be used to prepare a modified version if desired), Teacher Preparation Notes, comments, and the complete list of our hands-on activities are available at http://serendip.brynmawr.edu/sci_edu/waldron/.

1. We'll analyze inheritance for the case where 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 would you expect to observe in the children of these parents? (Again, you should think about what happens during meiosis and fertilization. You may want to use sockosomes to model meiosis and fertilization for two parents who are **Aa** or draw diagrams to represent these processes.)

Biologists use a **Punnett Square** to answer this type of question. The figure below shows more details than a typical Punnett Square. It shows that, as a result of meiosis in a mother who is **Aa**, half of her eggs will have a chromosome which carries the **A** allele, and the other half will have a chromosome with the **a** allele. Similarly, half of the father's sperm will have an **A** allele, and half will have the **a** allele. The four smaller squares within the larger Punnett Square show the possible genetic combinations in the zygotes resulting from fertilization of the two different types of eggs by the two different types of sperm.



Typically, Punnett squares exclude much of the explanatory material we have included in the above Punnett square. The simplified version of this Punnett square shown below illustrates the usual format of a Punnett square.

	A	a
A	AA	Aa
a	Aa	aa

2. What fraction of this couple's children would you expect to be **AA**? ____
3. What fraction of this couple's children would you expect to be **Aa**? ____
4. What fraction of this couple's children would you expect to be **aa**? ____

The children who have **AA** alleles will have normal pigmentation, and the children who have **aa** alleles will have albinism. These children are homozygous for the **A** allele or the **a** allele. **Homozygous** means that both copies of the gene have the same allele.

The next question is: What pigmentation will be observed for the children who have **Aa** alleles? This type of combination of two different alleles is called **heterozygous**. Often, one allele in a heterozygous pair of alleles is **dominant** and the other allele is **recessive**; this means that the dominant allele determines the observable characteristic of the heterozygous individual. Typically, the dominant allele is symbolized by a capital letter, in this case **A** for the allele for pigmentation. Thus, heterozygous (**Aa**) individuals will have normal pigmentation.

5. What fraction of this couple's children would you expect to have normal pigmentation? _____
6. What fraction of this couple's children would you expect to have albinism? _____

The **genotype** refers to the genetic makeup of an individual. The **phenotype** refers to the observable physical and physiological characteristics of an individual.

7. Give an example of two individuals who have the same phenotype, but different genotypes for the albinism gene. Explain how two individuals with the same phenotype can have different genotypes.

Biologists frequently express the fractions of different genotypes or phenotypes as ratios. For example, for the mating between two heterozygous parents, the genotype fractions are $\frac{1}{4}$ **AA**, $\frac{1}{2}$ **Aa**, $\frac{1}{4}$ **aa**, which can also be expressed as a 1:2:1 ratio.

8. For the corresponding phenotypes, the fraction with normal pigment is _____ and the fraction with albinism is _____, so the corresponding ratio is _____.
9. Suppose a father has **aa** alleles and a mother has **Aa** alleles. Complete the Punnett Square to describe this mating and determine what fraction of this couple's children would be expected to have albinism.

Coin Genetics

The way genes behave can easily be simulated using two-sided coins, where tails represent the recessive allele that controls pigment production (**a**), and heads represent the dominant allele (**A**). Suppose a parent is heterozygous (**Aa**). Then, tossing a coin and checking for tails up vs. heads up represents the 50-50 chance that an egg or sperm produced by the parent 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 the pair of coin tosses will indicate the pair of alleles contributed by an egg and a sperm to the baby that results from that mating.

1. Find someone to "mate" with.
2. 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 genetic makeup for the second, third and fourth children in this family. Record how many of these 4 children had each of the 3 possible combinations (**AA**, **Aa**, or **aa**) in the row labeled "first family of 4 children" in the table below.
3. Now make 4 more pairs of coin tosses to indicate the alleles in a second family of 4 children. Record these genotypes in the second row in the table below.
4. Do this two more times and record the results in the third and fourth rows of the table below.

Genetic makeup of
"children" produced by two
heterozygous (**Aa**) parents

	AA	Aa	aa
First family of 4 children			
Next family of 4 children			
Next family of 4 children			
Next family of 4 children			
Total			
Fractions			
Percents			
Predicted percents from Punnett square, page 2			

5. Add up your results to determine the total number of **AA**, **Aa**, and **aa** combinations in the children produced by your coin tosses. Calculate the fractions of these children who have each of the three genotypes. Convert the fractions to percents.
6. Compare the results for these children (produced by your coin toss matings between heterozygous parents) with the predictions from the Punnett Square on the second page. Are the percents of each genotype in these children similar to the predicted percents?

If there is any difference between your results and the predictions, is this difference relatively small or large?

- Next, consider the different families of 4 children each. Did you get different results for the number of **AA**, **Aa**, and **aa** combinations for the children in each family?

How do these results for the separate families compare to the predictions of the Punnett Square?

In many cases, the results for a family of four children will not match the predictions of the Punnett Square. Random variation in which particular sperm fertilizes which particular egg explains why the children in the individual families may differ considerably from the predictions based on a Punnett Square. The random variation observed in small samples usually averages out in larger samples. Therefore, the results for a large number of children from multiple pairs of parents with the same genetic makeup are usually close to the predictions of the Punnett Square.

Genetics of Sex Determination

As you probably know, human males have an X and a Y chromosome (**XY**), whereas females have two X chromosomes (**XX**). The gene that results in the development of male anatomy is located on the Y chromosome. This gene is called **SRY**, which stands for sex-determining region of the Y chromosome. A zygote must have at least one chromosome to survive. If the zygote also has a Y chromosome with the SRY gene, the embryo will develop testes and male anatomy. If the zygote does not have a Y chromosome with the SRY gene, the embryo will develop ovaries and female anatomy.

Your understanding of meiosis and fertilization provides the basis for understanding the inheritance of X and Y chromosomes. During meiosis in a female, the two X-chromosomes separate, so each egg has a single X-chromosome. In males, even though the X and the Y-chromosomes are very different, they can nevertheless pair with each other and separate from each other during meiosis. This means that males produce two kinds of sperm; half have an X chromosome and half have a Y chromosome.

- What will be the sex of a child produced when an egg is fertilized by a sperm that has a Y chromosome?

What type of sperm must fertilize an egg to result in a female child?

- Draw a Punnett Square which shows the inheritance of the sex chromosomes. Use X to indicate an egg or sperm with an X chromosome and Y to indicate a sperm with a Y chromosome.

- Based on this Punnett Square, what percent of children would you expect to be male?

4. To test this prediction, begin by writing down the initials of all the children your mother has had. Arrange these initials in order from the oldest to the youngest, indicating whether each was male or female.

5. Complete the following table.

	Total number of children	Number of males	% males
Your mother's children			
Children of the mothers of all the students in your class			
Predicted percent from Punnett square, page 5			

6. Next, compare the predicted percent male with the observed percent male for your mother's children, for the children of the mothers of the other members of your group, and for all the children in the class sample. How similar to the prediction are the observed results for individual families and for all the families combined?

Notice that the percent male children varies in different families and the percent male children in some families is very different from the predicted. To understand why, remember that each time a sperm fertilizes an egg to form a new zygote, there is random variation in whether the sperm has an X or Y chromosome. Random variation generally averages out in large samples, so the overall percent male children for all the mothers should be fairly close to the prediction.

7. Look at your group's lists of children's initials with the sex of each child to answer the following questions.

If a mother's first child is a son, is the next child necessarily a daughter?

If a mother's first child is a daughter, is the next child necessarily a son?

If a mother's first two children are sons, is the next child necessarily a daughter?

If a mother's first two children are daughters, is the next child necessarily a son?

These observations illustrate that you cannot predict the sex of the next child based on the sex of a previous child or children. Each time a sperm fertilizes an egg, the results are independent of any previous fertilizations that resulted in older brothers or sisters.

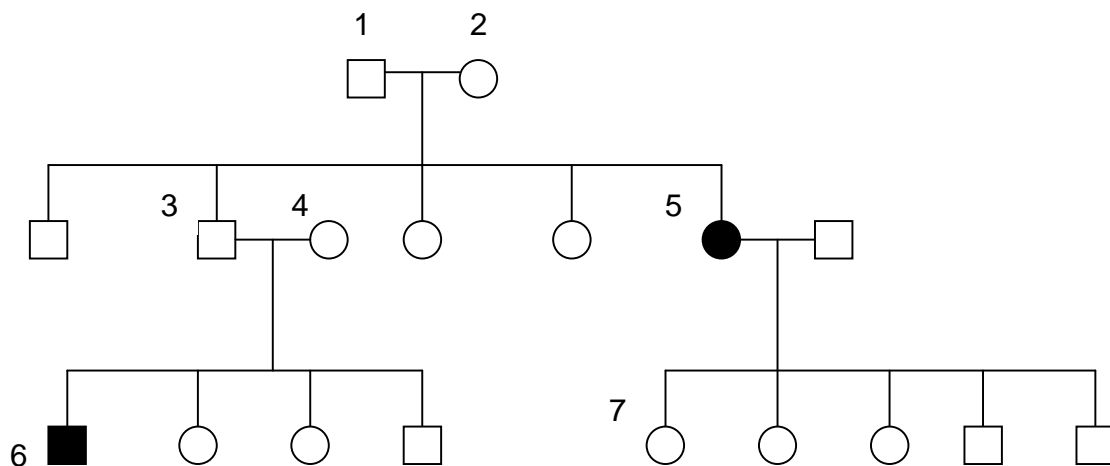
Pedigree analysis

Many traits in humans are controlled by genes. Some of these traits are common features like eye color, straight or curly hair, baldness, attached vs. free ear lobes, the ability to taste certain substances, and even whether you have dry or sticky earwax! Other genes may actually cause disease. Sickle cell anemia, muscular dystrophy, cystic fibrosis are each caused by specific alleles of a human gene, and can therefore be inherited from one generation to the next.

Human geneticists illustrate the inheritance of a gene within a family by using a pedigree chart. On such a chart, males are symbolized by a square (\square) and females are symbolized by a circle (\circ). People who are affected by a disease are symbolized by a dark circle or square.

The pedigree chart below shows inheritance of albinism. 1 and 2 represent a couple who had five children, including a son who is labeled 3 and a daughter who is labeled 5. Only one of the children, 5, was albino. The son who is labeled 3 and his wife who is labeled 4 had four children, including a son who is labeled 6.

1. In the pedigree below write the genotypes of the individuals who are labeled with numbers, using (**A**) to represent the dominant allele and (**a**) to represent the recessive allele. Start by indicating the genotypes of 5 and 6. Then use a Punnett Square to figure out what the genotypes for 3 and 4 must be. Next, determine the genotypes of 1 and 2. Finally, determine the genotype of 7.

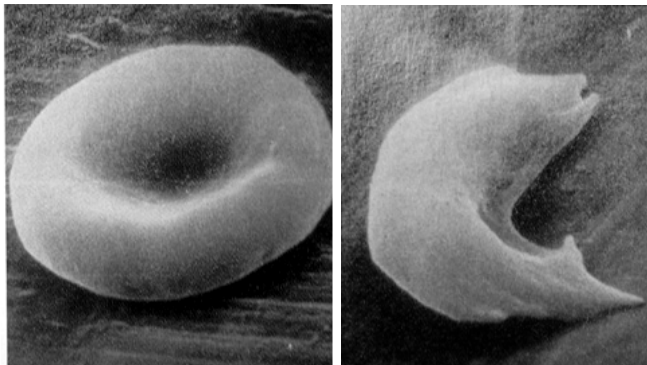


Many other genes are inherited in the same manner as this recessive allele which causes albinism. These include the genes for some genetic diseases, such as cystic fibrosis, Tay Sachs disease and phenylketonuria.

Genetics of Sickle cell anemia

For some genes, neither allele is completely dominant, so a heterozygous individual differs from either type of homozygous individual. (Remember that a heterozygous individual has two different alleles for the same gene, whereas a homozygous individual has two identical alleles for the same gene.)

The gene for hemoglobin is an example. One allele codes for normal hemoglobin, while another allele codes for altered hemoglobin, called sickle cell hemoglobin. When a person is homozygous for this sickle cell allele, this causes a serious disease called sickle cell anemia. The sickle cell hemoglobin tends to cause the red blood cells to assume a sickle shape, in contrast to the normal disk-shaped red blood cell shown on the left in the figure below.



1. What problems might be caused by the sickle-shaped red blood cells?

A person who is heterozygous for the sickle cell allele often has very few or no symptoms of sickle cell anemia. In addition, people who are heterozygous for the sickle cell allele are more resistant to malaria, an infection of the red blood cells which is transmitted by mosquitoes in many tropical countries. Thus, in areas where malaria is widespread, people who are heterozygous for the sickle cell allele are less likely to become ill and die. Because of this advantage, the sickle cell 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).

2. Suppose that a person who is heterozygous for the sickle cell allele (**Ss**) marries a person who is also heterozygous for this allele (**Ss**). Draw a Punnett Square to show the expected genetic makeup of their children, and then answer the following questions.

3. What fraction of their children will suffer from sickle cell anemia?

4. What fraction of their children will inherit the sickle cell allele, but will have very few or no symptoms of sickle cell anemia? (These children will be resistant to malaria.)

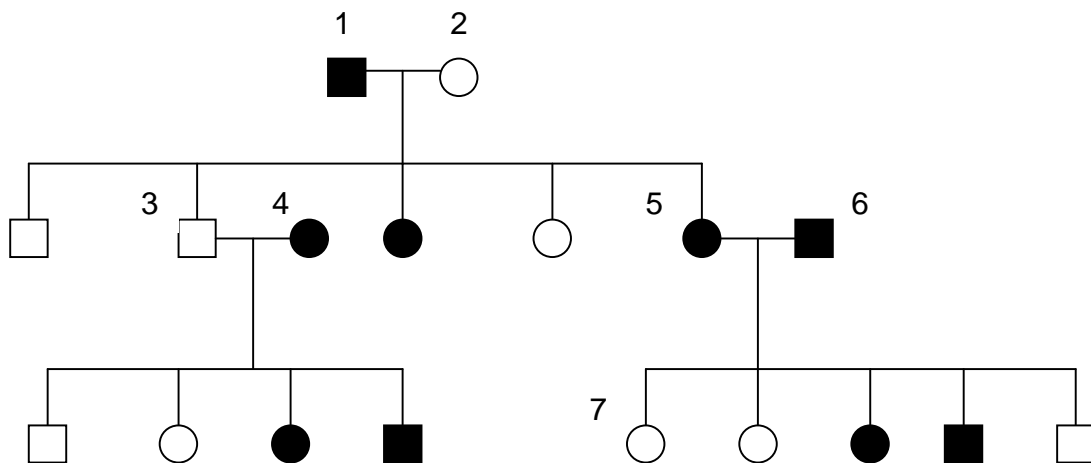
Challenge Question

Shown below is a pedigree chart for the inheritance of achondroplasia (ay-kon-druh-**play**-zhuh), a form of dwarfism. Dark circles or squares indicate individuals with achondroplasia. Examine the pedigree chart, and answer the following questions.

1. Is the gene that causes this form of dwarfism a recessive or dominant trait?

How do you know?

2. Using (**D**) to represent the dominant allele and (**d**) to represent the recessive allele, write the genotypes of the indicated individuals.



3. Based on the frequency of dwarfs among the people you have seen in your lifetime, do you think that the allele for achondroplasia is common or rare in the population?