We all know that children tend to resemble their parents. 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?**

1. A **gene** is a segment of a __________ 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 _________________. The different versions of a protein can result in different observable characteristics (i.e. different **phenotypes**).

Each cell in your body has two copies of each gene (one inherited from your mother and one inherited from your father).
- 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** for that gene.

<table>
<thead>
<tr>
<th>Genotype</th>
<th>→</th>
<th>Protein</th>
<th>→</th>
<th>Phenotype (characteristics)</th>
</tr>
</thead>
<tbody>
<tr>
<td>AA or Aa</td>
<td>→</td>
<td>Enough normal enzyme to make melanin, the molecule that gives color to our skin and hair</td>
<td>→</td>
<td>Normal skin and hair color</td>
</tr>
<tr>
<td>aa</td>
<td>→</td>
<td>Defective enzyme that does not make melanin</td>
<td>→</td>
<td>Very pale skin and hair color (albino)</td>
</tr>
</tbody>
</table>

2. Circle the genotypes in the chart that are homozygous. Explain how these two different homozygous genotypes result in different phenotypes.

3a. In a heterozygous person, often a **dominant** allele determines the phenotype and the other **recessive** allele does not affect the phenotype. This means that a heterozygous person has the same phenotype as a person who is homozygous for the ________________ allele. (dominant/recessive)

For example, a person who is heterozygous Aa has the same phenotype as a person who is homozygous AA because skin cells that have at least one A allele produce enough melanin to result in normal skin color.

3b. For this gene, which allele is dominant? ___ A ___ a
- Which allele is recessive?  ___ A ___ a
- What evidence supports your conclusion about which allele is dominant and which is recessive?

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1 By Drs. Scott Poethig, Ingrid Waldron, and Jennifer Doherty, Dept. Biology, Univ. Pennsylvania, © 2016. This Student Handout, a Genetics Supplement with an alternative module that does not assume prior completion of "Meiosis and Fertilization – Understanding How Genes Are Inherited" and additional modules, and Teacher Preparation Notes with instructional suggestions and background information are available at http://serendip.brynmawr.edu/sci_edu/waldron/#genetics
How does a baby inherit genes from his or her mother and father?

Each gene is a part of a DNA molecule which is contained in a chromosome. 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 and fertilization.

4. Meiosis produces gametes, which are the _________ or _________. Fertilization produces a _________________.

Inheritance of Albinism

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

If both parents are heterozygous (Aa), what different combinations of A and/or a alleles could be observed in the children of these parents?

To answer this question, your group will use model chromosomes to show how meiosis and fertilization result in inheritance when each parent has a pair of homologous chromosomes, one with an A allele and the other with an a allele.

➢ One of you should use your model chromosomes to demonstrate how meiosis produces different types of eggs, and another group member should demonstrate how meiosis produces different types of sperm.

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

➢ Next, model fertilization, using the model chromosome for each type of sperm to fertilize each type of egg.

6. In this chart, record the genetic makeup (the alleles) for each type of zygote produced by fertilization.

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.

7. 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 genetic makeup of each zygote.

8. The genetic makeup of each zygote in the Punnett square represents a possible genotype of a child of this couple. Explain why the genotype of each child is the same as the genetic makeup of the zygote that he or she developed from.
9. For an Aa mother, what fraction of her eggs have an a allele? _____
- What fraction of an Aa father’s sperm have an a allele? _____
- What fraction of this couple’s children would you expect to have the aa genotype? _____
- Explain your reasoning.

<table>
<thead>
<tr>
<th>A</th>
<th>a</th>
</tr>
</thead>
<tbody>
<tr>
<td>AA</td>
<td>Aa</td>
</tr>
<tr>
<td>a</td>
<td>Aa</td>
</tr>
</tbody>
</table>

10a. Complete this Punnett square for two parents who are homozygous AA.

10b. Complete this Punnett square for two parents who are homozygous aa.

10c. Complete this Punnett square for a mother who is heterozygous Aa and a father who is homozygous aa.

11. For each of the four Punnett squares above, circle the genotype of anyone who would have normal skin and hair color.

Notice that all of the children with normal skin and hair color have at least one parent who also has normal skin and hair color. Also, almost all of the albino children have at least one albino parent. These findings fit with our general observation that children tend to resemble their parents.

- There is only one example of a child who would have a different phenotype which was not observed in either parent. Use an * to indicate this example.

12. Explain why two albino parents will not have any children with normal skin and hair color, but two parents with normal skin and hair color could have an albino child.

13. Albino children are rare in the general population. Based on this observation, what is the most common genotype for parents? Explain your reasoning.
**Coin Toss Genetics**

The way genes behave during meiosis and fertilization can be modeled by using two-sided coins, where heads represent the dominant allele (A) and tails represent the recessive allele (a). This table explains how the coin toss model of inheritance represents the biological processes of meiosis and fertilization for heterozygous (Aa) parents.

<table>
<thead>
<tr>
<th>Biological Process</th>
<th>How This Will Be Modeled in Coin Toss Genetics</th>
</tr>
</thead>
<tbody>
<tr>
<td>Meiosis in an Aa parent produces gametes. Each gamete has an equal probability of</td>
<td>You toss your coin and check for heads up vs. tails up. This represents the 50-50 chance of getting an A allele</td>
</tr>
<tr>
<td>having an A allele or an a allele.</td>
<td>or an a allele.</td>
</tr>
<tr>
<td>Fertilization of an egg by a sperm produces a zygote. Each gamete contributes one</td>
<td>Two students each toss a coin and the result of this pair of coin tosses indicates the genotype of the child</td>
</tr>
<tr>
<td>allele to the genotype of the child that develops from the zygote.</td>
<td>that develops from the zygote.</td>
</tr>
</tbody>
</table>

- Find someone to “mate” with.
- Each of you will toss your coin; record the results as the genotype of the first child in the first family of four children in the table below. Make three more pairs of coin tosses and record the genotypes for the second, third and fourth children in this family.

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

<table>
<thead>
<tr>
<th>Result for each coin toss</th>
<th>Number with each genotype</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>AA</td>
</tr>
<tr>
<td>1st</td>
<td></td>
</tr>
<tr>
<td>2nd</td>
<td></td>
</tr>
<tr>
<td>3rd</td>
<td></td>
</tr>
<tr>
<td>4th</td>
<td></td>
</tr>
<tr>
<td>Totals</td>
<td></td>
</tr>
</tbody>
</table>

- Predictions based on Punnett square:
- Class data (Percent):
  - 1/4 = 25%
  - 2/4 = 50%
  - 1/4 = 25%

- Repeat this procedure three times to determine the genotypes for three more families of four children each, and record your results in the table.
- Complete the last three columns for these four families of coin toss children, and add your results. Give your teacher the total numbers for the AA, Aa and aa genotypes.
- Use a check mark to indicate any coin toss family of 4 children that has exactly the numbers of AA, Aa and aa genotypes predicted by the Punnett square.

To understand why some of the coin toss families do not have exactly the predicted number of children with each genotype, answer these questions.

1. Does the genotype produced by the first pair of coin tosses have any effect on the genotype produced by the second pair of coin tosses? 
   - ___ yes  ___ no

2. If a coin toss family has one aa child, could the second child in this family also have the aa genotype? 
   - ___ yes  ___ no
   - Explain your reasoning.
In real families the genotype of each child depends on which specific sperm fertilized which specific egg, and this is not influenced by what happened during the fertilizations that resulted in previous children. Therefore, the genotype of each child is independent of the genotype of any previous children.

3. Suppose that a mother and father who are both heterozygous $Aa$ have two children who also are heterozygous $Aa$. If this couple has a third child, what is the probability that this third child will also be heterozygous $Aa$?
- Explain your reasoning.

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.

4. Suppose that you had data for 20 families of four children each where both parents were heterozygous $Aa$. Would each of these families have exactly one albino child, as predicted by the Punnett square? Explain why or why not.

5. Your teacher will give you the class data to enter in the last line of the table on page 5. Are the percents of each genotype in the class data similar to the predictions of the Punnett Square?

The random variation observed in small samples usually averages out in large samples. Therefore, the predictions of the Punnett Square are usually more accurate for larger samples of children.

**Genetics of Sickle Cell Anemia**
Red blood cells are full of hemoglobin, the protein that carries oxygen. One hemoglobin allele codes for normal hemoglobin, and another allele codes for sickle cell hemoglobin. In a person is homozygous for the sickle cell allele, 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. This causes a disease called sickle cell anemia.

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Hemoglobin in Red Blood Cells</th>
<th>Shape of Red Blood Cells</th>
</tr>
</thead>
<tbody>
<tr>
<td>Homozygous for alleles for normal hemoglobin</td>
<td>Normal hemoglobin dissolves in cytosol</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Homozygous for alleles for sickle cell hemoglobin</td>
<td>Sickle cell hemoglobin tends to clump in long rods</td>
<td></td>
</tr>
</tbody>
</table>

5
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 ($) or recessive (s)? Explain your reasoning. Include a Punnett Square in your answer.

The sickle cell allele illustrates some common complexities of genetics that we have ignored thus far. Read the information in this box, and then answer questions 3 and 4.

People who are **homozygous** for the sickle cell allele have sickle cell anemia, including pain and organ damage due to blocked circulation and anemia (low red blood cell levels) due to more rapid breakdown of red blood cells. People who are **heterozygous** for the sickle cell allele almost never experience these symptoms. Therefore, the allele for sickle cell hemoglobin is generally considered to be recessive and the allele for normal hemoglobin is generally considered to be dominant.

However, a heterozygous person does not have exactly the same phenotype as a person who is homozygous for the allele for normal hemoglobin. Specifically, people who are heterozygous for the allele for sickle cell hemoglobin are less likely to develop severe malaria than people who are homozygous for the allele for normal hemoglobin.

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. Malaria parasites are less able to reproduce in red blood cells that have some sickle cell hemoglobin. This explains why people who are heterozygous for the allele for sickle cell hemoglobin have less severe malaria infections than people who are homozygous for the allele for normal hemoglobin.

3. Explain how the hemoglobin gene illustrates the following generalization:
   A single gene often has multiple phenotypic effects.

4. 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.