

Genetics Supplementⁱ

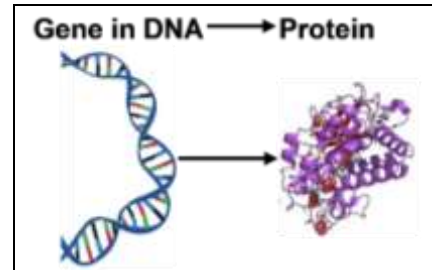
Why do parents and their children generally have similar characteristics? If siblings have the same biological mother and father, what explains any differences in the siblings' characteristics?

In this activity you will learn how genes contribute to the similarities and differences between family members.

I. How do genes influence our characteristics?

A **gene** is part of a DNA molecule that gives the instructions for making a protein.

Different versions of the same gene are called **alleles**. Different alleles give the instructions for making different versions of a protein. For example, the **A** and **a** alleles give the instructions for making different versions of a protein enzyme.



Allele	→	Protein
A	→	Normal enzyme for making melanin, a pigment molecule that gives color to our skin and hair
a	→	Defective enzyme that cannot make melanin

1. In the above table, circle each symbol or word that represents part of a DNA molecule.

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

- 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.

This chart shows how different **genotypes** (the genetic makeup of a person) can result in different **phenotypes** (the observable characteristics of a person).

Genotype	→	Proteins	→	Phenotype (characteristics)
AA	→	Normal enzyme that makes melanin	→	Normal skin and hair color
Aa	→	Normal enzyme that makes melanin and defective enzyme that can't make melanin	→	Normal skin and hair color
aa	→		→	Very pale skin and hair color (albino)

2a. Circle the heterozygous genotype in this table.

2b. Underline the version or versions of the protein enzyme that are produced in the skin and hair cells of a heterozygous person.

This example illustrates how a **dominant** allele (**A**) can determine the phenotype of a heterozygous person. The **A** allele in each skin cell and hair cell results in enough normal enzyme to make enough melanin to produce normal skin and hair color. Thus, the **recessive** allele (**a**) for the defective enzyme does not influence the phenotype of a heterozygous person.

3. Fill in the empty box in the above table to show that the **aa** genotype results in the production of a version of the enzyme which results in the albino phenotype.

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

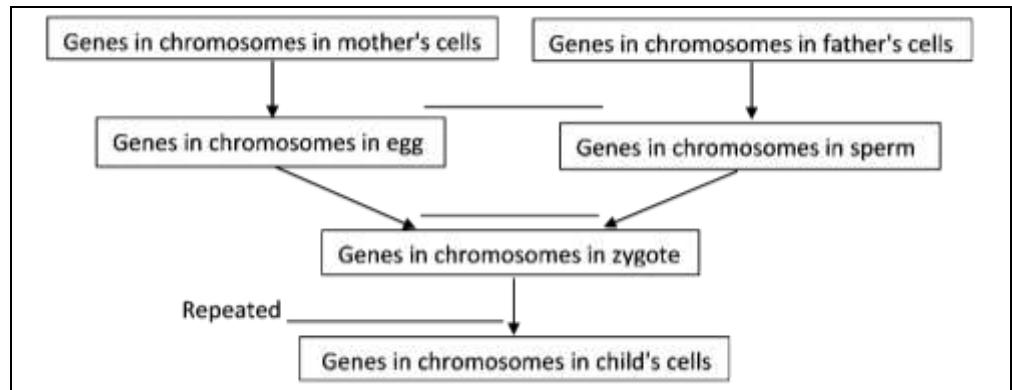
- A heterozygous person has the same phenotype as a person who is homozygous for the _____ allele.
- A _____ allele does not affect the phenotype of a heterozygous person.
- A _____ allele is represented by a capital letter.

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

Each gene is part of a DNA molecule, and each DNA molecule is contained in a chromosome. This flowchart shows how genes in chromosomes are inherited.

5. Fill in each blank with fertilization, meiosis, or mitosis.

6. Why does each cell in a child's body have the same genetic makeup as the zygote that the child developed from?



To better understand how meiosis and fertilization result in inheritance of genes, we will analyze this 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?

7a. Draw rectangles around each sister chromatid in one of the homologous chromosomes in this **Aa** father's cell.

7b. Complete the diagram to show how this pair of homologous chromosomes and sister chromatids separate during meiosis to produce sperm.

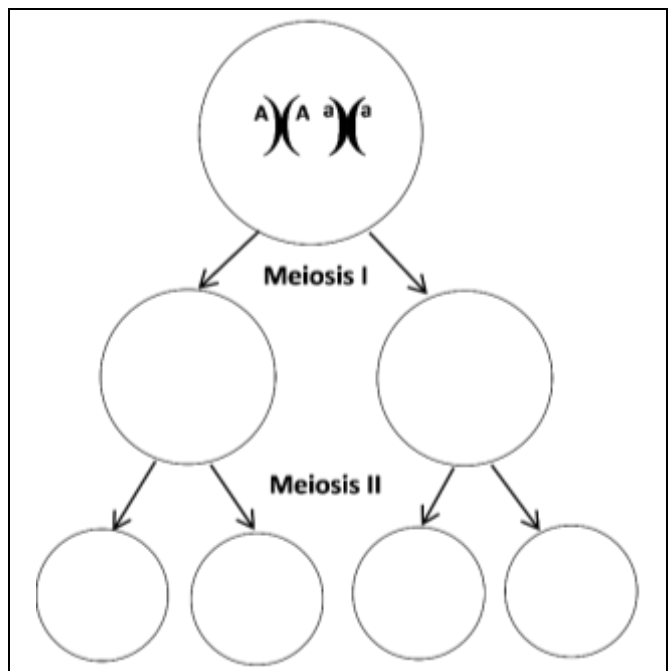
7c. How many copies of this chromosome does each sperm have?

7d. What is the genetic makeup of the different types of sperm that an **Aa** father can produce?

_____ or _____

7e. What is the genetic makeup of the different types of eggs that an **Aa** mother can produce?

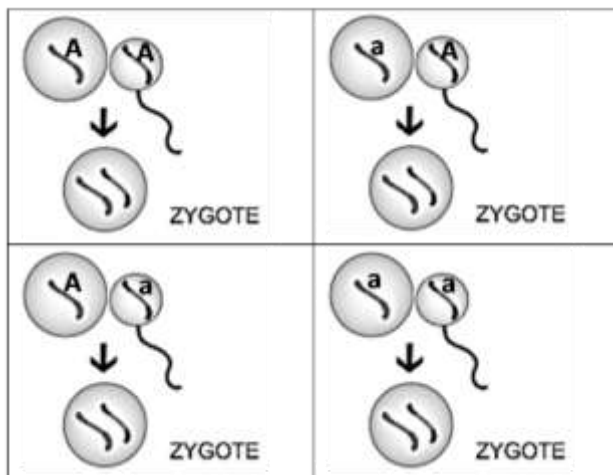
_____ or _____



8. This figure shows fertilization of each different type of egg by each different type of sperm. For each zygote, label the allele (A or a) on each chromosome.

9a. Circle the egg and sperm that would produce a zygote that would develop into an albino child.

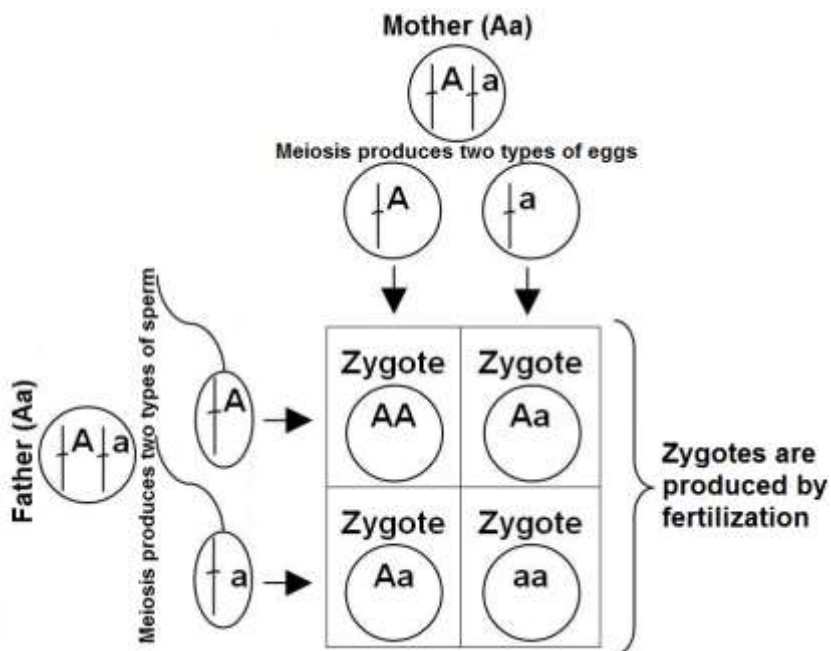
9b. Explain your reasoning.



This chart combines the results of meiosis and fertilization for an **Aa** mother and an **Aa** father. It shows the genetic makeup of the different zygotes that this couple could produce.

10a. Put an * next to the cells that were produced by meiosis in the mother.

10b. Circle the part of this chart that shows how fertilization can produce a zygote with the genetic makeup **AA**.



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

11. In this Punnett square:

- Label each letter that represents the genetic makeup of a gamete with a G.
- Label each pair of letters that represents the genetic makeup of a zygote with a Z.

12. Explain how each cell in a baby's body gets one copy of each gene from his/her mother and another copy of each gene from his/her father.

13a. A parent with normal skin and hair color has the _____ or _____ genotype. Two parents who have normal skin and hair color may have the same genotype or different genotypes. Draw three Punnett squares, one for each possible combination of these parental genotypes.

13b. Circle the genetic makeup of any zygote that will develop into an albino child.

13c. Explain how two parents with normal skin and hair color could have an albino child.

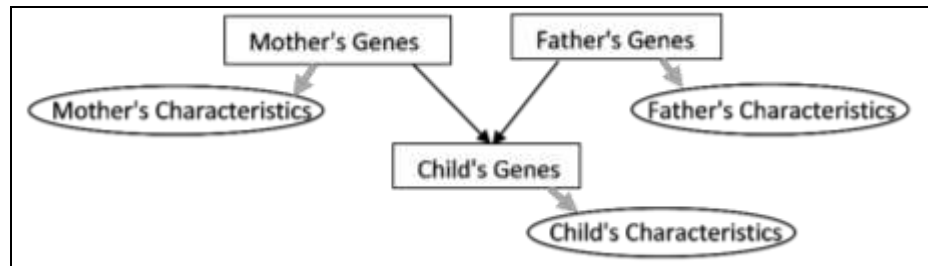
13d. For two parents who have normal skin and hair color,

- most of their children are albino.
- half of their children are albino.
- most of their children have normal skin and hair color.

14a. Draw a Punnett square for two albino parents.

14b. Explain why two albino parents will not have any children with normal skin and hair color.

This flowchart summarizes what you have learned about how genes contribute to the similarities between parents and their children.



15a. Each cell in a person's body has two copies of each gene. Where did these two copies of each gene come from?

15b. What biological processes transmit genes to a child (represented by the thin black arrows)?

15c. Explain how genes influence a person's characteristics (represented by the fatter gray arrows). A complete answer will include the words alleles and protein.

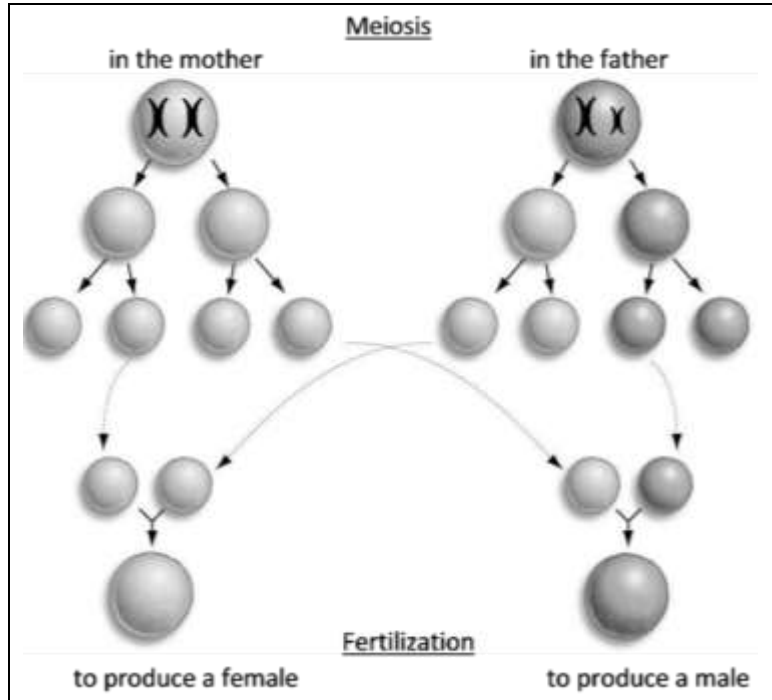
15d. Summarize the answers to questions 15a-15c in a sentence that explains why parents and their children generally have similar characteristics.

Genetics of Sex Determination

A crucial gene that stimulates the development of male anatomy is located on the **Y** chromosome. Therefore, a person with an **X** and a **Y** chromosome in each cell (**XY**) is male.

A person with two **X** chromosomes in each cell (**XX**) is female.

The top row of this figure shows two cells at the beginning of meiosis, each with a pair of sex chromosomes with sister chromatids. Since the Y chromosome is shorter than the X chromosome, the symbol for the Y chromosome is smaller.



1. In this figure:

- Put a rectangle around the four sperm produced by meiosis.
- Put an asterisk (*) next to each of the two zygotes.
- In each cell, draw the symbol(s) for the sex chromosome(s) that cell would have.

2. Complete this Punnett Square to show the inheritance of sex chromosomes. Use **X** and **Y** to indicate the genetic makeup of the father's sperm, mother's eggs, and the zygotes.

3a. Based on your Punnett square, what percent of children are expected to be male?

0% ___ 25% ___ 50% ___ 75% ___ 100% ___

3b. Explain your reasoning.

3c. If a couple's first child was male, what is the probability that their second child will be male?

0% ___ 25% ___ 50% ___ 75% ___ 100% ___

3d. Explain your reasoning.

4. To test your predictions in question 3, begin by entering the data for your mother's children in this table.

Sex of each child							Total number of children	Number of males	% males
1 st	2 nd	3 rd	4 th	5 th	6 th	7 th			

The chart below shows the sexes for the children in each family of one woman's descendants (her children, three families of grandchildren, and seven families of great-grandchildren). The ♀ and ♂ symbols indicate the sequence of male and female births in each family.

Percent male	0-14%	15-24%	25-34%	35-44%	45-55%	56-65%	66-75%	76-85%	86-100%
One child family	♀								
Two child families	♀♀				♂♀				♂♂ ♂♂
Three child families							♂♂♀		♂♂♂
Four child families	♀♀♀♀		♂♀♀♀		♀♂♂♀				
5+ child families				♀♀♂♀♂♂♀					
All 34 descendants					✓				

5. Draw the symbols for your mother's children in the appropriate location in this chart.

6. Describe how the above data fit with or deviate from your prediction about the percent male in question 3a.

7a. Explain why the percent male in individual families often differs from the Punnett square prediction. (A complete answer will include fertilizes or fertilization.)

7b. Approximately half the babies born in the US are male and half are female. Explain why the Punnett square prediction is accurate for large samples, even though the Punnett square prediction is not accurate for many individual families.

8a. Fill in each blank in these sentences with the best match from the list below.

A Punnett square can accurately predict _____ .

A Punnett square can not accurately predict _____ .

- a. the probability that the next child in a family will be male
- b. the actual sex of the next child that will be born in a family

8b. Explain your reasoning.

Sickle Cell Anemia and Sickle Cell Trait

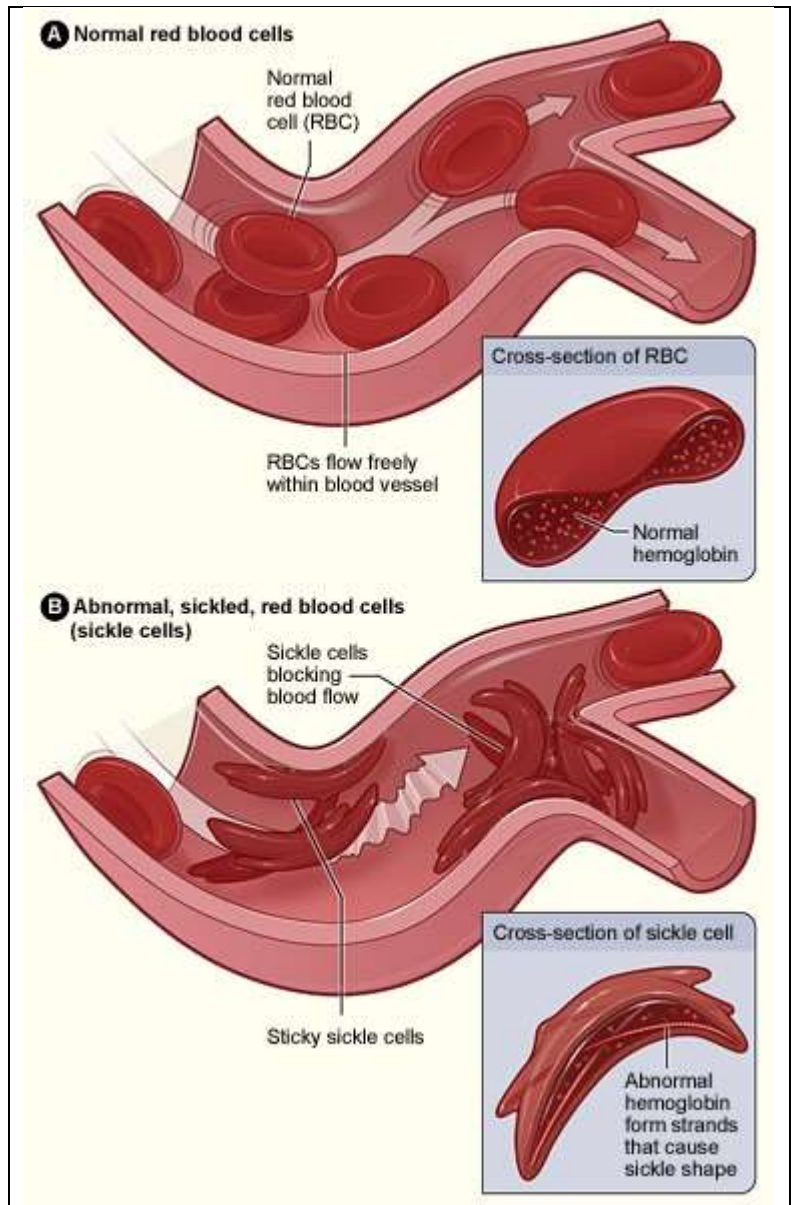
Red blood cells are full of hemoglobin, the protein that carries oxygen in the blood. One allele of the hemoglobin gene (**S**) provides the instructions to make normal hemoglobin. Another allele (**s**) provides the instructions to make sickle cell hemoglobin.



Red blood cells with normal hemoglobin are disk-shaped. These red blood cells can squeeze through the smallest blood vessels (Figure A).

In a person who is homozygous for the sickle cell allele (**ss**), sickle cell hemoglobin tends to clump into long rods that elongate red blood cells into sickle shapes (Figure B; table below).

1a. How can the **ss** genotype cause pain?

1b. How does the **ss** genotype cause anemia?



Genotype	→	Protein	→	Phenotype (characteristics)
SS	→	Normal hemoglobin dissolves in the cytosol of red blood cells. 	→	Disk-shaped red blood cells can squeeze through the small blood vessels → normal health
ss	→	Sickle cell hemoglobin can clump in long rods inside red blood cells. 	→	When sickle cell hemoglobin clumps in long rods → sickle-shaped red blood cells → block small blood vessels → reduced oxygen supply → pain, damage to body organs. Also, these red blood cells die faster than they can be replaced → anemia (low red blood cells). Person has <u>sickle cell anemia</u> .

Sickle Cell Trait

A person who is heterozygous for the sickle cell allele (**Ss**) has sickle cell trait. In a person with sickle cell trait, each red blood cell has roughly half sickle cell hemoglobin and half normal hemoglobin. The normal hemoglobin prevents the sickle cell hemoglobin from forming long rods. As a result, people with sickle cell trait almost never experience the pain, organ damage and anemia that occur in sickle cell anemia. Therefore, the **S** allele for normal hemoglobin is often described as dominant. However, heterozygous **Ss** people do not have exactly the same phenotype as homozygous **SS** people, so the **S** allele is not completely dominant.

Heterozygous **Ss** people have less severe malaria infections than homozygous **SS** people. The malaria parasite infects red blood cells. Malaria parasites are less able to reproduce in red blood cells that contain sickle cell hemoglobin. Therefore, an infected person who has sickle cell trait has fewer malaria parasites, so the illness is less severe.

In the US, people with sickle cell trait have the same life expectancy as the general population. However, people with sickle cell trait have an increased risk of some health problems. For example, people with sickle cell trait have a greater risk of sudden death during extremely strenuous exercise (e.g. Division I football or basic training in the military). These deaths are very rare, but tragic when they occur. Sudden death can almost always be prevented if the exercising person consumes adequate fluids and avoids excessive overheating. These same precautions are important for people who don't have sickle cell trait.

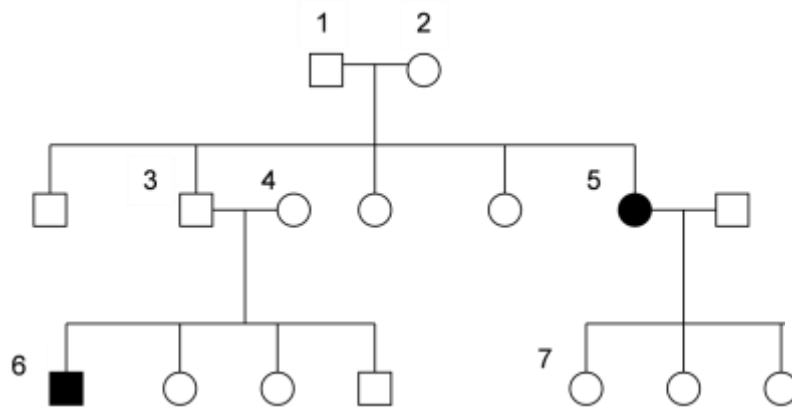
2. Often, when geneticists investigate a pair of alleles, neither allele is completely dominant or completely recessive. Complete the table to explain how each allele in a heterozygous **Ss** person influences his/her phenotype.

Allele	→	Protein	→	Effect on Phenotype
S	→		→	
s	→		→	

3. Explain why a person with sickle cell trait could easily not know that they have sickle cell trait.

4. What do you think is the best way to reduce the risk of sudden death for people with sickle cell trait? Should athletes and military recruits be tested for sickle cell trait? Explain your reasoning.

This pedigree chart shows the inheritance of sickle cell anemia in three generations of a family. In a pedigree chart, males are symbolized by a square (□) and females are symbolized by a circle (○). People who are affected by a condition or disease are symbolized by a dark square or circle. The couple labeled 1 and 2 had five children, including one daughter with sickle cell anemia (5). The son labeled 3 and his wife (4) had four children, including one son with sickle cell anemia (6). The son labeled 3 and his wife (4) had four children, including one son with sickle cell anemia (6). The son labeled 5 and his wife (7) had three children, none of whom are affected.



5a. Explain how person 5 inherited sickle cell alleles from parents who did not have sickle cell anemia. Include a Punnett square for parents 1 and 2 in your answer. (**S** = allele for normal hemoglobin and **s** = allele for sickle cell hemoglobin)

5b. In the pedigree, write the genotypes of each person who is labeled with a number.

A **model** is a simplified representation of a complex biological process. A model does not include all of the features of the process it represents; instead, the model highlights certain key features of the process. For example, pedigree charts and Punnett squares provide different types of information about inheritance.

6a. Compare pedigree charts with Punnett squares. What is an advantage of a pedigree chart as a model of inheritance?

6b. What are some advantages of Punnett Squares as a model of inheritance?

ⁱ By Drs. Ingrid Waldron, Scott Poethig, and Jennifer Doherty, Dept. Biology, Univ. Pennsylvania, © 2019. This Genetics Supplement has (1) an alternative introductory module that does not require model chromosomes, (2) Genetics of Sex Determination, and (3) Sickle Cell Anemia and Sickle Cell Trait. The Genetics Student Handout and Teacher Notes are available at https://serendipstudio.org/sci_edu/waldron/#genetics.