Unit 5: It's In Your Genes

Biology in a Box

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Unit 5: It's In Your Genes
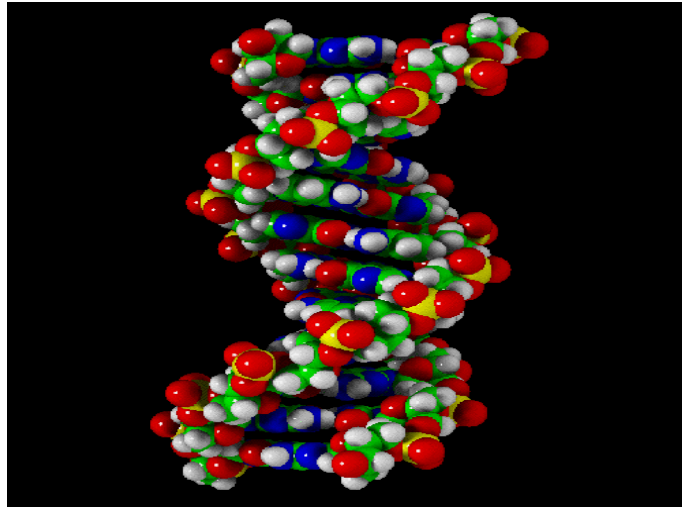## Materials List

- Molecular model building kit
- DNA puzzle
- Expandable DNA model
- Blindfold
- 2 boxes (F, M) each containing
  - 6 thin red chips
  - 6 thin white chips
- 1 box (GP) containing
  - 6 blue chips,
  - 6 red chips, and
  - 6 white chips
- 2 boxes (IF, IM) each containing
  - 3 thin red chips,
  - 3 thick red chips,
  - 3 thin white chips
  - 3 thick white chips
- 1 plastic Petri dish (C)
- 5 ears of corn:
  - 2 parental ears (1 each of two different colors)
  - 1 F1 generation ear (with kernels all of a single color)
  - 1 F2 generation ear (with kernels of two different colors)
  - 1 corn ear of unknown parentage

Unit 5: It's In Your Genes
Table of Contents

Unit 5: It's In Your Genes

Introduction

Perhaps the most important thing an organism does is to reproduce, or make copies of itself. Parents pass the traits they possess such as eye color, bone structure, and metabolic rate on to their offspring through a process called heredity. The materials and exercises in this box will help you to understand the process of heredity. This unit is divided into three sets of exercises, with **Exercise 1** covering the topic of genes as segments of DNA, **Exercise 2** covering the basics of Mendelian patterns of inheritance, and **Exercise 3** considering polygenic inheritance, in which multiple genes interact in the determination of a given trait.

Exercise 1: Genes are segments of a DNA molecule

The instructions for building an organism are encoded in the organism's **DNA**. This exercise deals with the replication of DNA and its importance to the process of heredity.

- Locate the cylindrical coil in the trunk and stretch it out by twisting the two ends in opposite directions.

The cylindrical coil is a model of a DNA molecule. The model is many orders of magnitude larger than an actual DNA molecule which can only be seen through an electron microscope that magnifies objects 10,000+ times. Each person has an enormous amount of DNA. Stretched out into a single line, a person's DNA would have a total length of about 1.7 meters! Human DNA, however, is tightly coiled so that it fits inside the cell's nucleus (its computer). In humans, DNA is separated into 46 linear strands called **chromosomes**.

Genes are chemical codes that determine specific traits such as the shape of your chin, the color of your eyes, your metabolic rate and personality. They are positioned along the rungs of DNA molecules and are composed of molecules called **nucleotide bases.** These nucleotide bases form pairs, and each **nucleotide base pair** forms one rung of the DNA ladder. A single gene is typically composed of a sequence of about 100,000 of these base pairs, while each chromosome (DNA strand) contains about 2×10^8 nucleotide base pairs. That makes a lot of genes!

Q1. Approximately how many genes are on a chromosome?

- Check your answer on the answer sheet under **Exercise 1, Q1.**

When an organism reproduces, it donates copies of its DNA to its offspring. Through this DNA, the offspring inherits its parents' traits. This process is called **heredity**. Typically, animals and plants have two copies of their genes (are **diploid**). One of these copies comes from the animal's mother and the other copy comes from the animal's father. The genes one parent passes on to its offspring may be the same or different than the ones that the other parent provides. This is because genes come in many varieties. The chemical variants of a gene are called **alleles**. The chemical differences between alleles occur as differences in the order of the nucleotide base pairs that make up the gene and differences between individuals of the same species and among all organisms, in fact, are determined by the sequence of nucleotide base pairs on the rungs of each DNA molecule.

- Examine the section of the DNA molecule you have expanded through twisting out of it coiled mode into a ladder. The sides or "backbone" of the ladder are constructed of sugar molecules (white) and phosphate groups (black). Four nitrogen bases form the rungs of the ladder and represent the genetic code or library, which has only four letters: A for adenine (red), G for guanine (green), T for thymine (blue) and C for cytosine (yellow). Figure 1 will help you to understand the structure you are looking at.

Figure 1.1. One side of a DNA ladder: sugar-phosphate backbone with a sequence of bases sticking off

Phosphate

Sugar—Base

Phosphate

Sugar—Base

Phosphate

Sugar—Base

Phosphate

and so on...
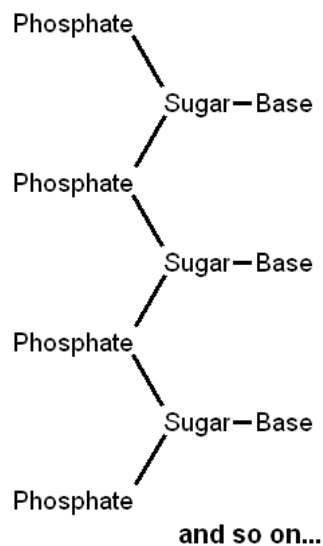
The nitrogen bases in a DNA molecule are ring-like structures that consist of carbon, hydrogen, nitrogen and oxygen atoms (Fig. 3). Cytosine (C) and thymine (T) are the smaller, single ring bases. They are called **pyrimidines**. The **purine** nitrogenous bases are Adenine (A) and guanine (G). These are the larger, double ring bases.

- Using the molecular model set (in the box labeled "**Exercise 1: Molecular Model Kit**"), construct each pyrimidine base and each purine base to confirm the difference in the sizes of the two types of bases. Use the diagrams in Figure 1.2 as blueprints for this construction. Do not dismantle your work until the end of this exercise.

Figure 1.2. Blueprints for nitrogen bases that form the rungs of the DNA ladder.

PYRIMIDINES (single ring bases)
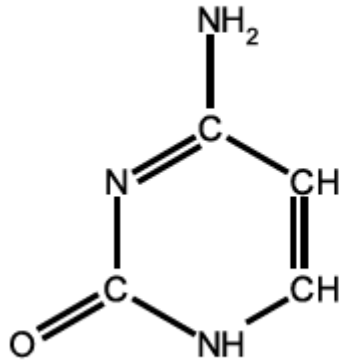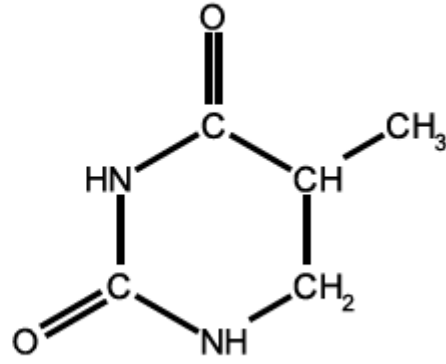
Cytosine (C)   Thymine (T)

PURINES (double ring bases)
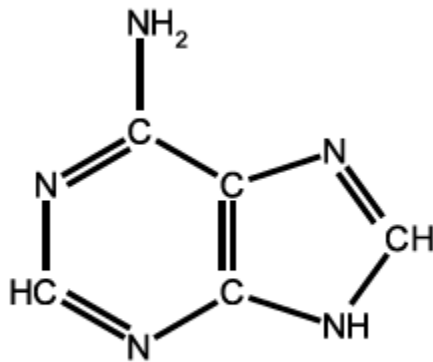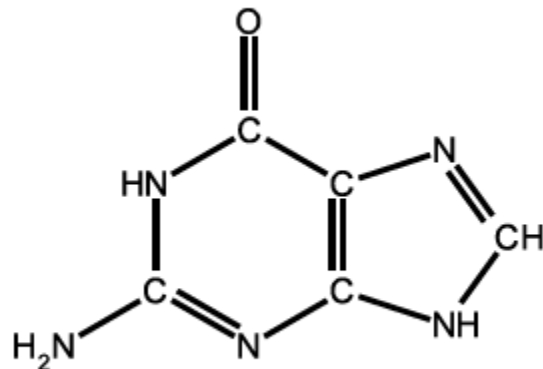
Adenine (A)   Guanine (G)

Each of the lines connecting the carbon (C), nitrogen (N), oxygen (O), and hydrogen (H) atoms to other atoms in Figure 1.2 represent **covalent bonds**, which are bonds formed when atoms share a pair of electrons. Double lines between two atoms denote two shared pairs of electrons between those two atoms. Covalent bonds are very important within DNA molecules, as they not only hold the atoms making up each nitrogenous base together, but also are the types of bonds that keep the DNA "backbone" together.

Exercise 1a: Replication of DNA: Making Exact Copies

Each pyrimidine base pairs with one of the larger purine bases.

- Looking at your model section of a DNA molecule, determine which single-ringed base pairs with which double-ringed base. Check a number of rungs to confirm the fact that a given base is always paired with a particular other base due to the chemical composition of the respective bases. The fact that each base can pair only with one other base is called the **Principle of Complementarity**, and is important to DNA replication.

Q2. What are the complementary base pairs?

- Check your answer against the correct answer which can be found on the answer sheet under **Exercise 1, Q2.**

- Locate the puzzle of a section of DNA molecule (in the box labeled "**Exercise 1. DNA Puzzle**"). Piecing the puzzle together will help you to understand why each nitrogen base can only pair with its complementary base.

Consulting the puzzle you have put together, position each of the two models of base pairs in the proper orientation and connect them. The types of bonds holding complementary base pairs together on the two strands of DNA that form the "ladder" are different from the covalent bonds discussed earlier. This class of covalent bond is called the **hydrogen bond**. Two hydrogen bonds are formed between adenine and thymine (A & T), and three hydrogen bonds are formed between cytosine and guanine (C & G). Though hydrogen bonds do a good job of holding the two complimentary strands of DNA together when necessary, they are much weaker than other covalent bonds. The reason that this is important will be made clear in just a bit, when we discuss the process of DNA **replication**.

- Now answer the following questions.

Q3. The number of A, G, C, and T bases in a gene will vary between DNA molecules, but one base will always be present in the same amount as A (adenine). Which base is it?

- Check your answer under **Exercise 1, Q3.**

Q4. Which base is present in the same amount as G (guanine) in the DNA molecule?

- Check your answer on the answer sheet under **Exercise 1, Q4.**

Q5. Here's a more difficult question. You are a real scholar if you get it right! Let
A = the number of adenine molecules in a molecule of DNA,
C = the number of cytosine molecules in a molecule of DNA,
T = the number of thymine molecules in a molecule of DNA,
G = the number of guanine molecules in a molecule of DNA.

- ○ Now see if you can complete the following equation:

$$A + C = \underline{\quad} + \underline{\quad}$$

The Principle of Complementarity is extremely important to the duplication (replication) of genes during reproduction. Figure 1.3 demonstrates how DNA duplicates itself by making two exact copies from one. Briefly, the two strands (sides of the ladder) spread apart, or "unzip", separating the two complementary bases on each rung. (This is where the relatively weak hydrogen bonds between complimentary base pairs is important: complimentary strands of DNA need to be able to separate when it is time to replicate!) Each base then calls in a new complement base, thus forming a new strand. The new strand attaches to the side of the original DNA ladder from which it was formed. The new DNA molecule twists into the characteristic helix. In this way, each side of the original ladder creates a DNA molecule, so that two DNA molecules are formed from the original molecule.

- You can demonstrate this process yourself on a piece of paper using an 8 base pair section of your DNA model. Complete steps 1 and 2 from Figure 1.3 for the DNA sample you have chosen.

Figure 1.3. During reproduction, DNA copies itself relying on the complementarity of its bases (After Gonick, L.& M. Wheels.1991. *The Cartoon Guide to Genetics*. HarperCollins)

Step 1. The two sides of the ladder (strands of DNA) split apart.
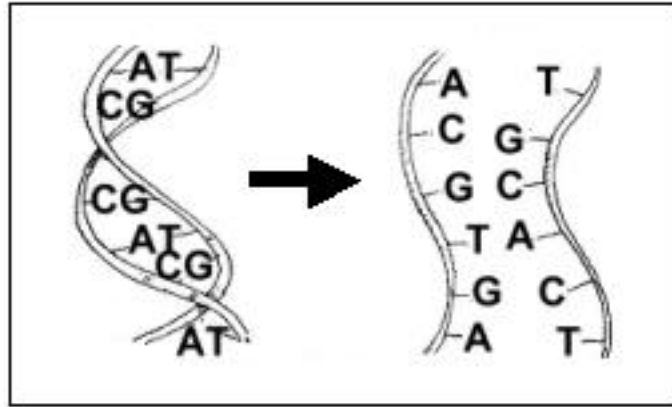
Step 2. Along each strand, a new strand forms in the only possible way.
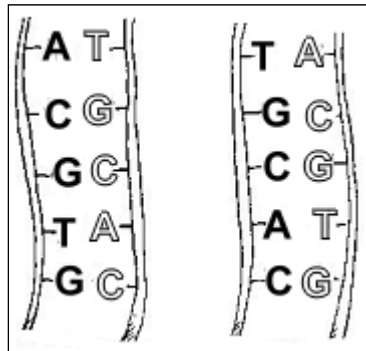
Step 3. The ladder twists into a helix and we wind up with two copies of the original DNA molecule.
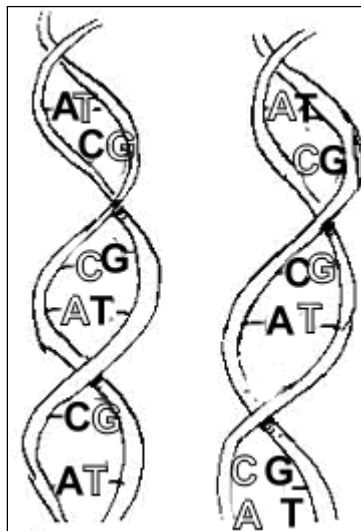
Function of DNA

DNA can be read again and again, just like a library book. During an organism's lifetime, each gene is read millions of times. Genes contain instructions for building molecules called polypeptides (enzymes and other important proteins) that perform vital biological functions. Each gene contains the "code" for the synthesis (production) of one or more polypeptides. In short:

thousands of nitrogen bases = one gene → one or more polypeptides

Exercise 1b: Accidents (mutations) do happen.

While DNA replication is very accurate, accidents do happen. To demonstrate this, complete the following experiment.

- Divide the class into two teams.
- Your teacher will read a piece of genetic code from one side of your DNA model that is 6 letters long (e.g. A T G T A C) and write it down on a piece of paper.
- He/she will whisper the six letter code to a student at each side of the classroom and ask them to whisper it to another classmate, and so on, until all of the students in teams 1 & 2 have heard the code.
- The last person to hear the code in each team will write it down and turn it in to the teacher
- The teacher will now put the original code on the board at the front along with the two codes recorded at the end of the replication process (whispered code through the sequence of students)..
- Is it the same?
- If not, how many letters have changed? Have deletions occurred (code is now less than 6)?

Each change represents a mutation. A mutation is a chemical change to a gene. Mutations may alter a gene's output. For example, mutations may alter the structure of the enzyme the gene makes, alter the amount of enzyme the gene makes, or cause the gene to produce no enzyme at all. Mutations create new alleles of a gene, which may lead to the expression of new traits. We calculate the rate of mutation as the number of base changes in a sequence divided by the total number of bases in the sequence.

- What are the rates of mutation in your experiment?

You can multiply your answer by 100 to obtain the percent change. For example, if two bases were incorrect, your answer would be 2/6 or 1/3. The mutation rate would thus be about 0.33, and the percent change would be about 33%.

- Calculate each group's percent change.

Exercise 2: Patterns of Trait Inheritance

Chromosomes are strands of nuclear DNA that contain specific genes. The nucleus of a plant or animal cell contains two versions of most of its chromosomes. One version comes from each parent. These pairs of like chromosomes are called **homologous pairs**. If a chromosome belongs to a homologous pair it is called an **autosome**. If a gene is located on an autosome, then the nucleus contains two alleles of the gene. One allele is found on each of the autosomes in the homologous pair. For example, suppose that the gene that determines human eye color is located on an autosome, and that B and b are the alleles of the gene. Since every person receives one allele from their father and one allele from their mother, a person has one of three possible allele combinations. In particular, a person could have any of the following allele combinations, BB, Bb, bB or bb, where the first allele is the maternal allele (the one donated by the mother), and the second allele is the paternal allele (the one donated by the father). However, which parent donated which allele does not alter the expression of the trait that the allele determines, so scientists treat the allele combinations bB and Bb as equivalent. Scientists call the set of alleles that a person has for a gene (or group of genes) the individual's **genotype**. In the eye color example above there are three possible genotypes BB, Bb or bb.

The chromosomes are usually tangled together so as to be indistinguishable from one another. When the cell prepares to divide, however, the chromosomes appear as distinct x-shaped bodies called **duplicated chromosomes** (See Figure 1). A duplicated chromosome consists of two identical copies of the same chromosome joined together at a central location. The identical copies of the chromosome are called **sister chromatids**. The point where the two chromatids join together is called the **centromere**. Figure 2.1 depicts duplicated chromosomes in the nucleus of an animal cell.

9

- Carefully examine Figure 2.1.

Figure 2.1. A cell's nucleus showing its chromosomes (DNA molecules). Genes positioned along these chromosomes determine an organism's traits.
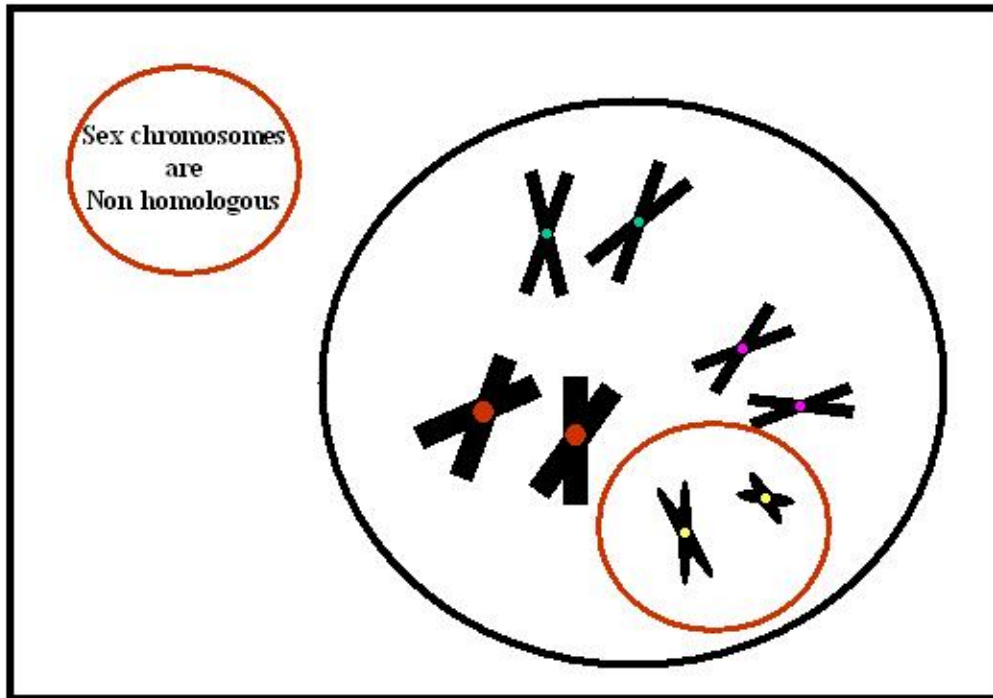
- The pairs of matching chromosomes are the homologous pairs. Although both of the autosomes in a homologous pair carry the same genes, they may carry different alleles of these genes.
- Note that two of the chromosomes do not look like any of the other chromosomes. These two chromosomes form a non-homologous pair. They are called the **sex chromosomes**, because they govern the sexual characteristics of male and female offspring. Locate the sex chromosomes in the model cell.
- Now try to answer the following questions:

   Q1. How many homologous pairs are in the nucleus?

   Q2. How many autosomes are in the nucleus?

   Q3. How many sex chromosomes are in the nucleus?

   Q4. How many chromatids are in the nucleus?

- Check your answers in the Answer section at the end of the workbook.

Cells called **gametocytes /germ or sex cells** can undergo a special type of cell division (**meiosis**) in which the nuclear DNA replicates itself and then divides twice. Through this process a gametocyte produces four **gametes** (eggs or sperm). Gametes are special because they carry only one version of each chromosome, and therefore, only one allele of each gene. Sexual reproduction is the process through which a gamete from one individual joins with a gamete from a second individual to form a new organism/offspring. The genotype of an individual produced via sexual reproduction is determined by the laws of probability.

Exercise 2a: Basic Probability

To every experiment there corresponds a set of possible outcomes, called the **sample space** of the experiment. We will denote the sample space of an experiment by the letter S. In our case, the experiment of interest is sexual reproduction. The sample space of this experiment is the set of all allele combinations that the resulting offspring could have.

Q1. Suppose that a man and a woman with genotypes Bb and BB reproduce. What is the sample space of this experiment?

Q2. Suppose that a six sided die is rolled. What is the sample space of this experiment?

When the experiment of interest is sexual reproduction, we may use a special table called a **Punnett square** to find the sample space. To construct a Punnett square, draw a two-by-two table like the one pictured below. Label the columns of the table with the father's alleles (possible gametes [sperm in animals]) and the rows of the table with the mother's alleles (possible gametes [eggs in animals]). In each cell, record the allele at the beginning of the cell's row and the allele at the top of the cell's column. For example if a man with genotype Bb and a woman with genotype Bb reproduce, then the corresponding Punnett square is the one pictured below. By convention, the male's gametes are presented across the top of the table and the female's down the left side as shown below.

	B	b
B	BB	Bb
b	bB	bb

Q3. Draw the Punnett square for the experiment from question **Q1.**

Note that the genotype Bb appears twice in the Punnett square from the example on the previous page. This is because there are two ways for these parents to produce such an offspring: the offspring will have genotype Bb if the father contributes a B allele and the mother contributes a b allele, or if the father contributes a b allele and the mother contributes a B allele.

The Punnett square shows all of the genotypes that the offspring may have (the sample space) *and* the number of ways that each genotype can be produced.

Q4. Use your Punnett square to find the number of ways that each of the offspring genotypes from question **Q1** can be produced.

A subset of the sample space is called an **event**. To be more specific, an event is a set that contains some (possibly all) of the experiment's outcomes. In the example above, the set $E = \{Bb, BB\}$ is an event. Note that the order in which we list the elements in the set does not matter. This means that the sets $\{Bb, BB\}$ and $\{BB, Bb\}$ are considered to be the same. There are many ways to describe the event E with words. For example, we might say that E is the event that the child has at least one B allele.

Q5. Can you think of any other ways to describe the event E?

The sets $\{Bb\}, \{BB\}, \{bb\}, \{BB, bb\}, \{Bb, bb\}$ and $\{Bb, BB, bb\}$ are other possible events for this experiment. Events such as $\{bb\}$, which contain a single element are called **elementary events**. If two events contain none of the same elementary events then they are said to be **mutually exclusive** events. For example, $\{Bb, BB\}$ and $\{bb\}$ are mutually exclusive events.

Q6. Suppose that you roll a six sided die. Let E denote the event that you roll less than a five. Write down all of the elements that belong to the event E.

Q7. Let B be the event that you roll 1, 4, or 6, that is, let $B = \{1, 4, 6\}$. Are B and E mutually exclusive? If not, which elementary events belong to both B and E?

Q8. Find three possible events of the experiment in question **Q1.** Also find a pair of mutually exclusive events.

The **probability** that any specific event occurs measures the likelihood that the event occurs. If E is an event, then we denote the probability that E occurs by $P(E)$. Sometimes the probability that an event occurs can be determined through intuition. At other times it may be determined experimentally.

There are three basic axioms (rules) of probability that are used to determine the probability that an event occurs.

Axioms of Probability
1) If S is the sample space of an experiment, then $P(S) = 1$
2) If E is any event, then $0 \leq P(E) \leq 1$
3) If A and B are mutually exclusive events, then $P(A \text{ or } B) = P(A) + P(B)$

Or, expressed verbally,

1) The sample space S of an experiment is the set of all possible outcomes. One of the outcomes in the sample space will definitely occur.
2) The likelihood of a particular event ranges from impossible to absolutely definite. (Probabilities are usually expressed as fractions, or more commonly, decimals, ranging from 0, or 0% likely, to 1, or 100% likely.)
3) If an outcome can be one of two alternatives (but not both), the probability of either event occurring is equal to the sum of the likelihood of each event's occurrence.

A couple of very useful rules follow directly from the axioms of probability.

Rule 1:
If all of the outcomes in the sample space are equally likely to occur, then the probability that any specific event, E, occurs is the ratio of the number of outcomes that belong to E divided by the total number of outcomes in the sample space.

For example, suppose that you roll a six-sided die. Since the sides of the die all have the same size and shape, the die is equally likely to land on any of its sides (we are assuming that the die is a fair one), so all of the experiment's outcomes are equally likely to occur. Therefore, we can use Rule 1 to find the probability that any event occurs. For example, the probability that the event $B = \{1,4,6\}$ occurs is

$$P(B) = \frac{number\ of\ outcomes\ in\ E}{total\ number\ of\ possible\ outcomes} = \frac{3}{6} = \frac{1}{2}.$$

13

Note that it is the symmetry of the die that allows us to apply Rule 1. That is, we could apply Rule 1 because all of the die's sides were the same size and shape (the die is a fair one).

Q9. Suppose that there are three pennies in your pocket. One of the pennies was made in 1991, another was made in 1985, and the third penny was made in 2006. Now suppose that you pull one penny from your pocket (without looking). Are you equally likely to pick any of the pennies? Explain why or why not.

Q10. What is the probability that you pick the penny from 1985? What is the probability that you don't pick the penny from 1985?

We already know that if a gene is located on an autosome, then an animal has two alleles of this gene. We also know that when an animal reproduces, it gives only one of these alleles to its offspring. In fact, the animal is equally likely to give either of these alleles to its offspring. As a result, when we draw a Punnett square, each cell of the square is equally likely to happen. Therefore, we can determine the probability that the offspring has a specific genotype by completing a three step procedure.

1) Draw the Punnett square.
2) Count the number of cells in which the genotype of interest appears.
3) Divide the number from Step 2 by the total number of cells in the Punnett square.

Example: Suppose that a woman and a man with genotypes Bb and Bb reproduce. What is the probability that their child has genotype Bb?

First we draw the Punnett square.

	B	b
B	BB	bB
b	Bb	bb

Next we count that the genotype Bb appears in 2 cells. (Note that the order in which the alleles are listed does not affect the genotype, that is, the genotype Bb is the same as bB.)

Finally we see that probability that their child has genotype Bb is $\frac{2}{4} = \frac{1}{2}$.

Q11. Suppose that a woman and a man with genotypes Bb and BB reproduce. What is the probability that their child has genotype BB?

Suppose that E is an event. The event that E does not occur is denoted by $\bar{E}$.

Rule 2:

If E is an event then $P(\bar{E}) = 1 - P(E)$.

In other words, the likelihood that an event does **not** occur is equal to 1 (100%) minus the chance that the event **does** occur.

Q12. Let E be the event from **Q11.** Use Rule 2 to find $P(\bar{E})$.

There is one more concept from probability that is very important to heredity. This is the concept of independence. Two events are **independent** if the occurrence of one event does not affect the probability that the other event will occur.

For example, if we roll two dice then the outcome of the first roll does not affect the outcome of the second roll. Therefore if we let A be the event that the first die lands on a 6, and B be the event that the second die lands on a 3 then the events A and B are independent.

Q13. Suppose two coins are tossed. Let A be the event that the first coin is heads, and B be the event that the second coin is heads. Are A and B independent?

Q14. Suppose that two children attend the same daycare. Let A be the event that the first child catches a cold and B be the event that the second child catches a cold. Are the events A and B independent?

When two events are independent, the probability that both events occur is equal to the product of the probabilities that each event occurs.

If A and B are independent events, then $P(A \ and \ B) = P(A)P(B).$
In fact, the converse is also true. That is, if the probability that both events occur is equal to the product of the probabilities that each event occurs, then the events are independent.

If $P(A \ and \ B) = P(A)P(B)$, then A and B are independent events.

Q15. Find the probability that *A* and *B* occur, where *A* and *B* are the events from Q13.

In carefully controlled experiments with pea plants in the 1800s, the Austrian monk Gregor Mendel learned much of what we know today about the process of heredity. We will use plastic disks and ears of corn to explore trait inheritance patterns.

Exercise 2b: Law of Segregation – Offspring are equally likely to inherit either of their parents' alleles.

The process of meiosis described earlier produces four gametes, each of which carries only one allele of every gene. These gametes are produced in such a way that if we consider any specific gene, two of the gametes carry the maternal allele (the one that was donated by the mother) and two of the gametes carry the paternal allele (the one that was donated by the father). As a result, when a gamete is chosen to create an offspring, it is equally likely to carry either of the parent's alleles. In addition, when a gamete produced by a male unites with a gamete produced by a female, the alleles that the two gametes carry are independent of each other. This is known as the Law of Segregation.

This experiment models how offspring inherit alleles from their parents. Red and white chips will be used to represent the alleles of a gene that determines the color of an organism. Red chips represent the allele of the gene that creates red color, and white chips represent the allele of the gene that creates white color. On paper we will denote the red allele by C, and the white allele by C'. We assume that the color gene is located on an autosome, so that each individual has two alleles of the gene.

- Find **plastic boxes F** (for female) and **M** (for male) holding red and white chips in them. These boxes hold the alleles that each parent has to give to its offspring. In this simulation, both of the parents have genotype CC', so each box contains an equal mix of 6 red and 6 white chips.
- Count out your chips and make sure that there are equal numbers of red and white chips in each box.
- Place the female parent box (F) to your left and the male parent box (M) to your right.
- Divide the plastic container C into two sections, Cf and Cm. In a moment, you will place alleles from the female parent into the Cf section, and alleles from the

16

male parent into the Cm section, so place each section in front of the appropriate parent box.

- Now put on the blindfold and remove one chip from each box and put it into the dish in front of the box.

Q1. What is the probability that you drew a red allele from the female parent box? Explain how you came to this answer.

- Take off your blindfold. What allele did each parent offer? Record the first offspring's genotype on a sheet of paper or on the blackboard under the heading 'Offspring of Heterozygous Parents'.

The genotype of your first offspring may be $CC, C'C', CC'$, or $C'C$. However, $C'C$ and CC' are identical, because it does not make any difference which parent donated a particular allele. If both of an individual's chromosomes have the same allele for a trait, then the individual is said to be **homozygous** for that trait. Your offspring with genotypes CC or $C'C'$ are homozygous. If an individual has two different alleles for a trait, then the individual is said to be **heterozygous** for the trait. The parents in our experiment had one white allele and one red allele, so they were heterozygous for the trait.

- Now put the chips back in the boxes from which you removed them, and repeat this process another 15 times. Each time, record the offspring's genotype on the board and return the chips to the boxes from which they were drawn.

Q2. Suppose that when choosing the genotype of the first offspring you drew a red chip from the female parent's box, and did not replace this chip before the second draw. What is the probability that the second offspring receives a red allele?

We replace the chips after each selection because an individual is equally likely to receive either of his or her mother's alleles and either of his or her father's alleles. If a parent has one chromosome for each color, then each time that parent reproduces, the probability that he or she will donate a red allele is ½, and the probability that he or she will donate a white allele is ½. **In general, each time an organism reproduces, it is equally likely to donate either of its alleles for a given gene to its offspring.**

- Draw a Punnett square for this experiment and use it to complete the fourth column of Table 2.1 below.

Table 2.1. Cross between Heterozygous Parents

Genotype	Frequency of Genotype	Fraction of Offspring with Genotype	Probability that an Offspring has Genotype
$C'C$			
$C'C'$			
CC			

Q3. Based on the probability that an individual has genotype CC, how many offspring would you expect to have genotype CC? What about $C'C'$ and $C'C$?

- Count the number of times that each genotype appears in your data, that is, find the frequency of each genotype. Record your answers in Table 1.

Q4. How close are the actual frequencies to the frequencies that you predicted based on probability? Why wouldn't they be the same?

- Find the fraction of the offspring that have each genotype. Record your answers in Table 1. **(Note that the fraction of the offspring that have a particular genotype is equal to the number of offspring with that genotype divided by the total number of offspring.)**

Q5. Find the probability that an individual offspring is not homozygous white.

- Repeat this experiment using one parent that is homozygous red for color and one parent that is homozygous white for color. Complete a Punnett square analysis for this homozygous cross and record your findings in Table 2.2.

Table 2.2. Cross between Homozygous Red and Homozygous White

Genotype	Frequency of Genotype	Fraction of Offspring with Genotype	Probability that an Offspring has Genotype
$C'C$			
$C'C'$			
CC			

- Repeat this experiment using one parent that is homozygous for the red chip color and the other parent heterozygous . Complete a Punnett square analysis for this homozygous-heterozygous cross and record your findings in Table 2.3.

Table 2.3. Cross between Homozygous Red and Heterozygous

Genotype	Frequency of Genotype	Fraction of Offspring with Genotype	Probability that an Offspring has Genotype
$C'C$			
$C'C'$			
CC			

- Put all of your chips back in the appropriate boxes and seal each parent box with the rubber bands provided. There should be 6 red and 6 white chips in each box.

We need to consider one more thing before leaving Mendel's Law of Independent Assortment. Genes do not come in just two alleles (chemical variants). Human blood types have three alleles: $A, B,$ and O. Eye color in fruit flies has many more alleles (Fig. 3).

Figure 3. Multiple alleles for eye color in fruit flies. Each of the colors below is produced by a different allele of the gene for eye color.
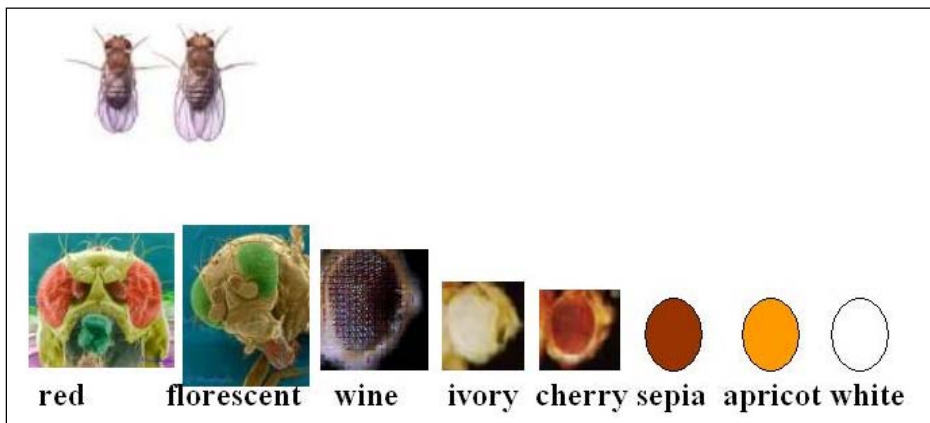
How is eye color inherited when there are multiple alleles? In this experiment, you will need to determine the male and female parents' genotypes before producing your offspring.

- Find the box GP, which represents a population gene pool. This includes all of the alleles of the color gene that are available to the next generation of your species. It has 8 chips of each of three colors (red C, white, C' and blue C''). Spread these chips out on the table.

- Put on your blindfold and pick out two chips and place them in the dish (Cf) corresponding to the female parent.
- Remove your blindfold and record the female parent's genotype.
- Put the chips back in the GP box.
- Put on your blindfold and pick out two more chips and place them in the dish (Cm) corresponding to the male parent.
- Remove your blindfold and record the male parent's genotype.
- Construct a Punnett square for this cross.
- Use the Punnett square to find the probability that an offspring produced by this cross will have each genotype.
- Repeat this experiment several times.
- Be sure to put all 8 chips of each color back in the GP box and close it with the rubber band provided when you are finished.

What have you learned? Despite the fact that the gene pool may contain many different alleles of a gene, each parent possesses only two alleles. This is because each parent has only two chromosomes which carry an allele of the gene.

Exercise 2c: Mendel's Law of Independent Assortment

Our Austrian monk Mendel also experimented with pea plants to see if different traits such as plant height and flower color were inherited together (linked) or were inherited independently. To help you understand Mendel's question, pretend for a moment that you are shopping for a car. You enjoy traveling with your large family, so you need a car with a lot of space for passengers and luggage. At the same time you are a thrifty person, and would like a car with very good gas mileage. This presents a problem because the gas mileage of the car is linked to the size of the car, as the larger the car, the poorer the gas mileage.

Q1. Can you think of two car features that are not linked?

According to Mendel's Law of Independent Assortment, alleles for different genes are inherited independently. This means that the allele you inherit from your mother for eye color is independent of the allele that you inherit from your mother for hair color. Mendel's law of independent assortment holds most of the time. However, it may not hold for genes that are found close together on the same chromosome. When genes are located very near one another on a particular chromosome, they are often said to be linked. This means that during meiosis, alleles of genes that are found close together are more likely to go together into the

newly-formed gametes, and traits represented by those alleles are more likely to be inherited together.

Now we will use a chip experiment to demonstrate the Law of Independent Assortment. We will use red chips and white chips to represent the alleles for color, and large and small chips to represent the alleles for size. The alleles of the color gene are **C** (red) and **C'** (white). The alleles of the thickness gene are **T** (thick) and **T'** (thin).

Below is an example of how to complete a Punnett square for the inheritance of two traits. The genotype of the both parents is **CC'TT'**. Remember, each of these genotypes reflects the pairs of alleles that each parent has for two different traits: color and thickness. When completing Punnett squares involving multiple traits, it is important to remember that **each parent contributes one and only one allele for each gene considered to each offspring**. In other words, a gamete produced by either parent would only have one allele from each gene (in this case, one allele for color, and one allele for size). **Thus, when creating a Punnett square, the column and row headings should represent all of the different types of gametes that each parent could produce!**

		Possible gametes produced by male parent			
		CT	**CT'**	**C'T**	**C'T'**
Possible gametes produced by female parent	**CT**	*CCTT*	*CCTT'*		
	CT'	*CCTT'*			
	C'T	*CC'TT*			
	C'T'	*CC'TT'*			

- Finish filling out this Punnett square.

NOTE: For a Punnett square involving two traits, there may be as many as 16 cells in the Punnett square. This is because the maximum number of types of gametes a parent can produce is equal to 2^n, where *n* is equal to the number of traits being considered. If both parents could produce this many different types of gametes, the Punnett square would have 2^n rows, and 2^n columns, resulting in a total of $2^n \times 2^n = 2^{2n}$ cells in the square.

- **What is the maximum number of cells for a Punnett square involving three traits?**

- **ANSWER:** A Punnett square involving three traits would have a maximum of $2^{2n} = 2^{(2\times3)} = 2^6 = 2 \times 2 \times 2 \times 2 \times 2 \times 2 = 64$ cells.

Exercise 2c.1: The counting principle and tree diagrams
In exercises 2a and 2b, we used probability to analyze the way an offspring inherits a *single* trait. Those previous experiments consisted of only two steps or *tasks*. The first task was to pick the allele that came from the female parent, and the second task was to pick the allele that came from the male parent. It was easy to list and count all of the possible outcomes. In considering how an offspring inherits *multiple* traits, in which many more tasks (with many more outcomes) are involved, using tree diagrams and the counting principle is another helpful way to determine the probability of an event, and an alternative to drawing a Punnett square.

The counting principle
- If there are x ways to perform one task, and y ways to perform a second task, then there are xy ways to perform both tasks.
- Suppose for example that we flip two coins. Then there are 2 possible results from the flip of the first coin (heads or tails), and 2 possible results from the flip of the second coin, so there are 2×2 = 4 possible results when flipping both coins.

Q2. Suppose that you have three shirts (green, orange, and yellow) and two pairs of pants (blue and white). How many different outfits can you assemble from these clothing items?

Tree diagrams
- A tree diagram is a visual aid that helps us find and count all the possible outcomes of an experiment. For example, we can construct a tree diagram for the experiment from **Q1**.

```
        blue ——→ green, blue
  green <
        white ——→ green, white

        blue ——→ orange, blue
  orange <
        white ——→ orange, white

        blue ——→ yellow, blue
  yellow <
        white ——→ yellow, white

  shirt        pants       outcome
```
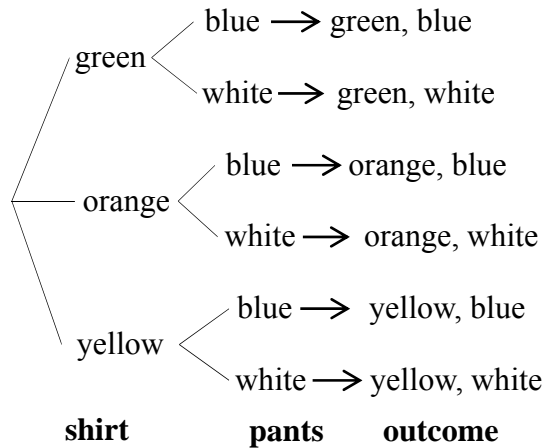
- A tree diagram can also help us to find the probability that an event occurs. For example, suppose that when you get up in the morning, you grab a shirt and a pair of pants from your closet without turning on the light. **What is the probability that your shirt is yellow?**

- **ANSWER:** Since you don't turn on the light, you are equally likely to grab any of the shirts or pants in your closet. Therefore, the probability that your shirt is yellow is the quotient of the number of outcomes with a yellow shirt divided by the total number of outcomes. Looking at the tree diagram we see that there are two outcomes with a yellow shirt and six outcomes total, so the probability that your shirt is yellow is 2/6 = 1/3.

Q3. What is the probability that your shirt is yellow or orange?

What if you got another green shirt as a birthday gift? This would change the probabilities of each outcome, but we can still use a tree diagram to help us do so, using the rules of probability. Now you would have four shirts instead of three, so the probabilities of getting each possible outfit would change, since the probabilities of grabbing a shirt of a particular color are no longer all equal.

- **What are the probabilities of getting each color shirt now that you have received this gift?**

ANSWER: The probability that you would draw a green shirt at random now would be equal to 2/4 = 1/2, since you now have four shirts, and two of them are green. The probability of drawing an orange shirt would now be equal to 1/4, and the probability of drawing a yellow shirt would also be 1/4.

Though you could have reflected this by drawing another tree diagram, and adding an additional branch to represent the new green shirt, another way of calculating the probabilities of the overall outcome (the final outfit) without having to draw extra branches would be to write the probabilities of the result of each task on our tree diagram. We can also then use these to calculate the probabilities of each of the outcomes. Look at the new tree diagram below for an example.

blue (P=1/2) ⟶ green, blue

green (P=1/2)

white (P=1/2) ⟶ green, white

blue (P=1/2) ⟶ orange, blue

orange (P=1/4)

white (P=1/2) ⟶ orange, white

blue (P=1/2) ⟶ yellow, blue

yellow (P=1/4)

white (P=1/2) ⟶ yellow, white

shirt   **pants**   **outcome**
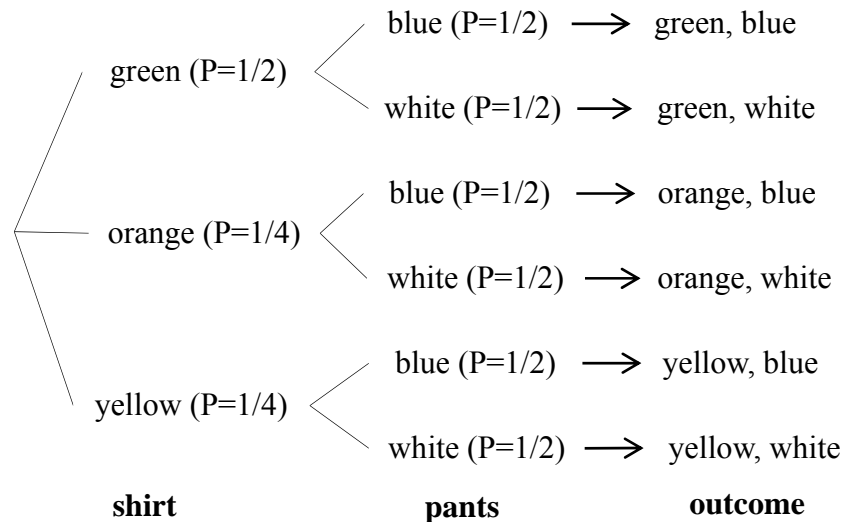
Recall that if events A and B are independent, $P(A \ and \ B) = P(A)P(B)$. Since the shirt that you grab from your closet at random has no effect on the pants that you grab at random (these events are independent!), you can calculate the probabilities of each outcome on your tree diagram by simply multiplying the probabilities of the branches along the branches leading to that particular outcome, as shown below:

blue (P=1/2) ⟶ green, blue (P=1/2 × 1/2 = 1/4)

green (P=1/2)

white (P=1/2) ⟶ green, white (P=1/2 × 1/2 = 1/4)

blue (P=1/2) ⟶ orange, blue (P=1/4 × 1/2 = 1/8)

orange (P=1/4)

white (P=1/2) ⟶ orange, white (P=1/4 × 1/2= 1/8)

blue (P=1/2) ⟶ yellow, blue (P= 1/4 × 1/2 = 1/8)

yellow (P=1/4)

white (P=1/2) ⟶ yellow, white (P=1/4 × 1/2 = 1/8)

shirt   **pants**   **outcome**
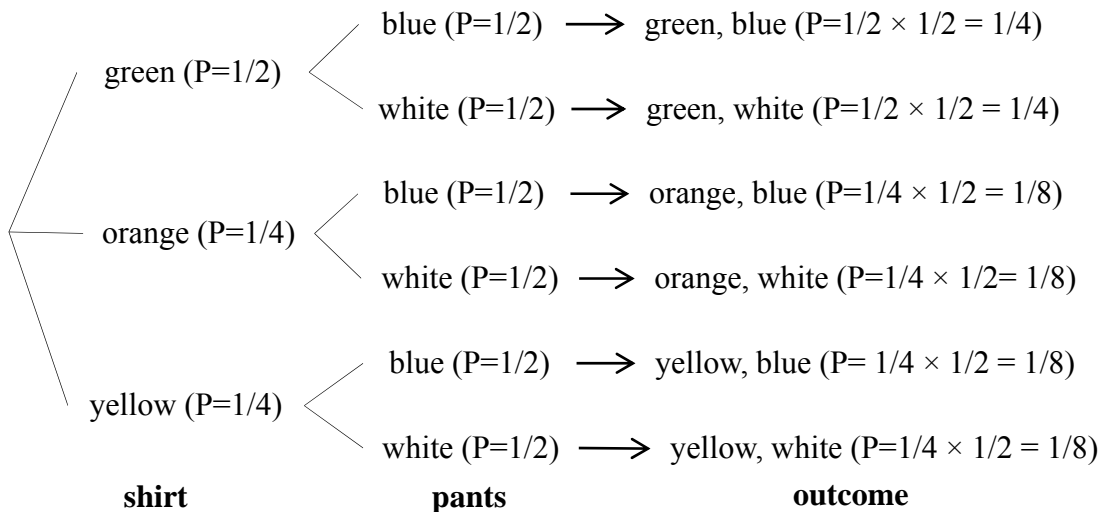
Now, let's apply this information to the topic at hand, the inheritance of multiple traits. Remember our Punnett square from earlier? To refresh your memory, this was of a cross between two parents, both with the genotype *CC'TT'*.

Q4. How many different tasks are involved in determining the genotype of an offspring in this cross? List each of these tasks.

Q5. In how many ways can each of the tasks from the previous question be performed?

Q6. Construct a tree diagram to show all of the possible outcomes of this cross.

Q7. How many outcomes are possible for this experiment?

Q8. How many possible genotypes could the offspring produced by this cross have? (Remember that the genotype is the set of alleles that an individual has with no regard to which parent donated which allele.)

Exercise 2c.2: Simulating independent assortment

- Now we will simulate the inheritance of multiple traits by an offspring produced by a cross between parents of randomly determined genotypes.
- Find the boxes labeled **IF** (representing the female parent) and **IM** (representing the male parent).
- Make sure each box has equal numbers of thick red, thin red, thick white, and thin white chips (3 of each)..
- Put on your blindfold and find your parent's genotypes by picking two chips each from the respective **IF** and **IM** boxes. Record the genotypes of the parents on a sheet of paper.
- Repeat this experiment two more times, simulating new parent genotypes for each cross as described above.
- Answer **Q4-Q8** above for each of your crosses.

Q9. Let *A* be the event that the offspring has at least one *C* allele, and *B* be the event that the offspring has at least one *T* allele. Use each of your tree diagrams to find $P(A)$, $P(B)$, $P(A \text{ and } B)$, and $P(A)P(B)$ from each of your crosses. Do your answers support Mendel's Law of Independent Assortment? That is, do they support the hypothesis that alleles for the color and thickness genes are inherited independently? Explain why or why not.

Q10. (Critical thinking!) Why was it important that you made sure that both the IF and IM boxes contained equal numbers of thick red, thin red, thick white, and thin white chips?

Exercise 2d: There are genotypes and then there are phenotypes.

Thus far, we have talked only about the genes that underlie traits and how these genes are inherited. The **genotype** of an individual refers to the specific alleles of a gene that the individual possesses. Genes are not observable without the use of molecular tools. The observable traits of an individual are referred to as the individual's **phenotype**. The phenotype of a homozygous red individual (CC) is red, and the phenotype of a homozygous white individual ($C'C'$) is white. But what is the phenotype of an individual who is heterozygous (CC')? If the traits are of equal influence, we might expect the individual to exhibit some intermediate phenotype between red and white, as in pink. However, Mendel discovered that when he crossed peas with red flowers with peas with white flowers, the offspring all had red flowers. An allele that masks the presence of another allele is called **dominant**. An allele that has its presence masked is called **recessive**. We denote dominant alleles with capital letters, and recessive alleles with lower case letters. So from now on, C will denote the allele for red color, and c will denote the allele for white color. Thus in our chip system, the genotypes CC and Cc result in the red phenotype, and the genotype cc results in the white phenotype.

To demonstrate how genes that have dominant and recessive alleles are expressed, we will examine the progeny (offspring) of corn plants. Here the progeny are the individual kernels on an ear of corn. Each of these kernels could potentially give rise to a new plant. The trait we will be examining in this wild corn (species = *Zea mays*) is kernel color, which may exhibit various coloration.

- Find the parental ears. The kernels on these ears of corn will grow into the parent plants. The kernels on the ear of one color will become female parents and the kernels on the ear of the other color will become the male parents. Furthermore, we know that each parent is homozygous for the color it exhibits.
- Find the ear marked as *F1* offspring. The kernels on this ear of corn are the offspring of a cross between a corn plant that grew from one of the kernels from a parental ear of one color and a corn plant that grew from one of the kernels from an ear of a parental plant of the second color.

Q1. What color are the kernels on the *F1* ear of corn?

Q2. Why are they this color?

Q3. What are the genotypes of the *F1* offspring?

Q4. What are the phenotypes of the *F1* offspring?

A cross between two of the *F1* offspring produces the ear labeled *F2*. The kernels on this ear of corn are the second generation of offspring. The original parental cross was $CC \times cc$. The *F2* generation was produced by the cross $Cc \times Cc$.

- What do you see?

- Count 8 rows of kernels to see how many of the kernels are of each color. Record your answers.

Q5. What fraction of the kernels that you counted is of the first color? What fraction of the kernels that you counted is of the second color?

Q6. What is the ratio of kernels of the first color to kernels of the second color?

- Complete a Punnett square analysis of the cross between two of the *F1* kernels. Crossing two individuals of the *F1* generation produces offspring of the *F2* generation.

Q7. Based on your Punnett square, what fraction of the kernels of the *F2* generation would you expect to be of the first color? What fraction of the kernels would you expect to be of the second color? What is the **expected** ratio of kernels of the first color to kernels of the second color?

What have we learned from examining the inheritance of traits that are determined by genes with dominant and recessive alleles? Because of the dominant/recessive relationship that exists between the alleles, recessive traits are not exhibited by heterozygous generations. However, the alleles that produce recessive traits are still present, so that the recessive trait may resurface in a later generation.

Exercise 3: Epistasis – Interactions Between Genes (*For high school/advanced students)*

In the previous exercise, you examined inheritance of kernel color in corn (*Zea mays*). You may have deduced, based on the sample ears of corn representing the parental, *F1*, and *F2* generations that color might be controlled by a single gene, with a simple system of dominance of one allele over another. However, like many traits in other organisms, kernel color in corn is determined by **epistasis**, or the interaction of multiple genes. Kernels in corn may be red, purple/blue, yellow, or white due to the interactions among four pigment genes.

Exercise 3a: Polygenic inheritance and corn kernel color

- Examine the diagram below and locate the parts of a corn kernel.
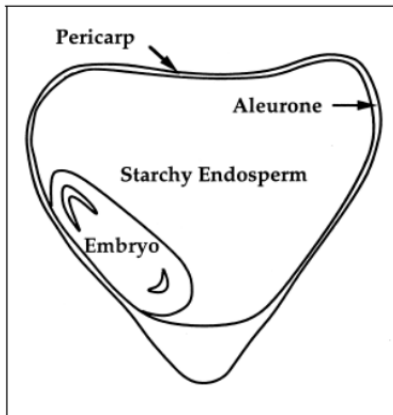
Figure 3.1. Structure of a kernel of corn (*Zea mays*). From Ford, R.H. 2000. Inheritance of kernel color in corn: Explanations & investigations. *The American Biology Teacher* 62(3):181-188.

The **pericarp**, or outer covering of the kernel, is colorless. Rather pigment production occurs in both the **aleurone,** the layer below the pericarp and in the **endosperm**, the nutrient supply for the developing corn plant embryo.

Three genes determine the color (pigmentation) of the aleurone layer:
- **Red Aleurone 1** gene, which has two alleles - ***P & p***
- **Colored Aleurone 1** gene, which has three alleles - ***C, C', & c***
- **Colored1** gene, which has two alleles- ***R & r***

If a kernel is homozygous recessive for any one of these genes, or if a kernel has at least one *C'* allele, the aleurone of the kernel is also colorless, like the pericarp.

A fourth gene affects pigmentation in the endosperm:
- o **White 1** gene, which has two alleles - *Y & y*

- Examine the tree diagram on the following page, following which shows how phenotype (kernel color) is determined by the interactions among genotypes of the four genes listed above. We present it here to show you how complex **polygenic** (epistasis involving multiple genes) trait inheritance can be.

Figure 3.2. Illustration of epistatic interactions determining kernel color in corn (*Zea mays*).

- Using the information on the previous pages, answer the following questions:

Q1. In Exercise 2d, you may have guessed that kernel color in corn involves a single gene with two alleles, and a system of simple dominance. In fact, using the parental, *F1*, and *F2* offspring ears, this appears to be the easiest explanation. Explain, in terms of genotypes and phenotypes, what may have led you to this original conclusion.
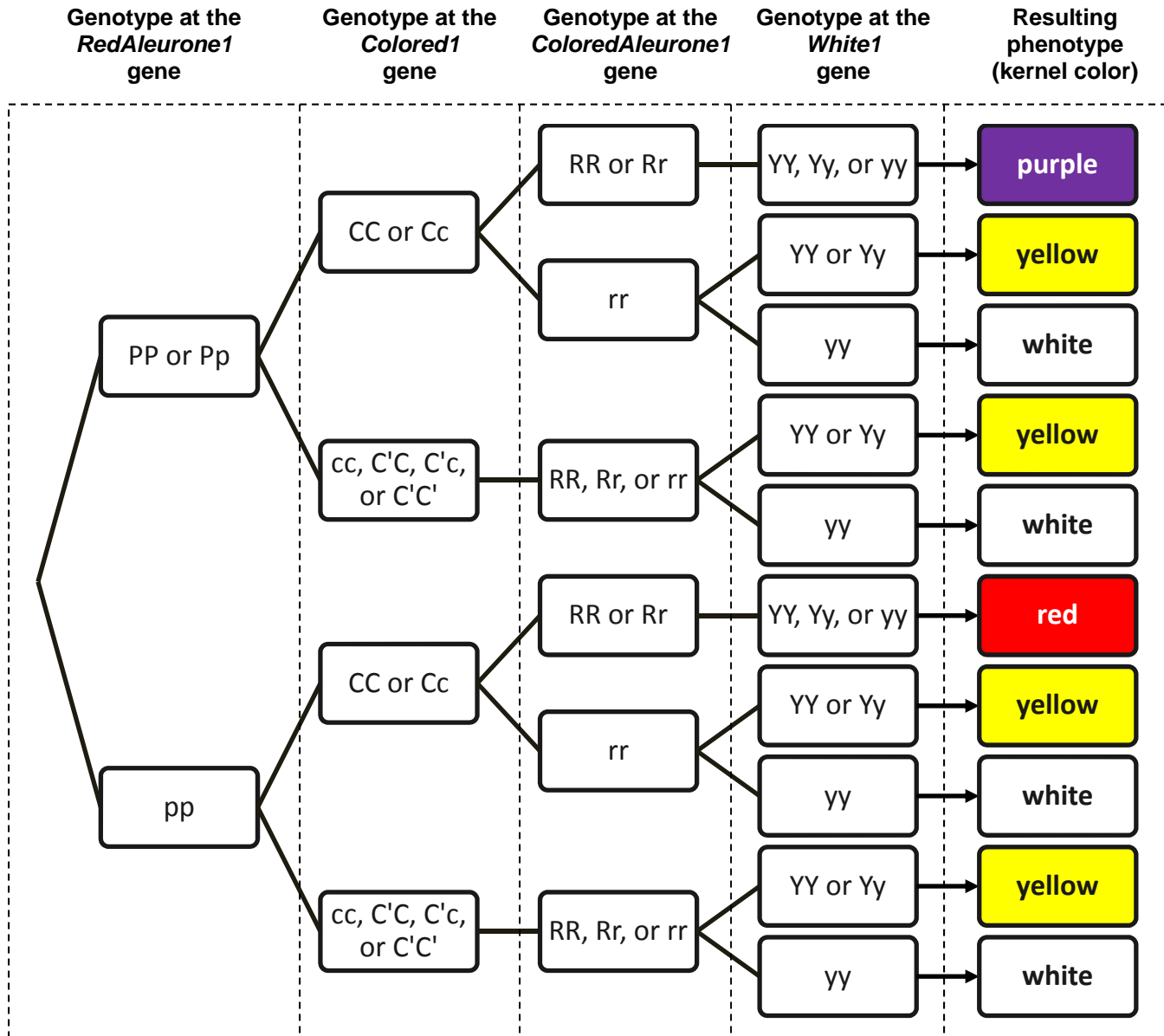
Q2. Using the tree diagram on the previous page, calculate the total number of genotypes that will result in each phenotype (kernel color). **HINT: Count the number of pathways one could take to achieve the kernel color of white, red, yellow and purple. (Note that some cells have multiple genotypes in them!)**

- Knowing what you now know about the epistatic determination of kernel color in corn, take a look at the ears of corn representing the three generations from the last exercise: parental, *F1*, and *F2* generations. Using the tree and information provided on the 4 genes, answer the following questions:

Q3. Since you know what generation a particular corn ear belongs to in Exercise 2d, try to determine its genotype for each of the 4 kernel pigment alleles. How many pathways might lead to this color: for the parental type? For the *F1* generation? For each kernel color in the *F2* generation ear?

Q4. Can you think of some ways to help determine potentially unknown genotypes at certain genes of a particular corn kernel, aside from actually examining the kernel's DNA?

- Now examine the additional ear of corn marked "Mystery Parents".

Q5. What can you deduce about the parental phenotypes and genotypes that produced the ratio of kernel colors observed in this ear? You may wish to again consult Figure 5.2, as well as play around with a few Punnett squares to help you with this question. Don't get discouraged if you can't figure out everything about the possible genotypes of the parents, as the nature of epistatic interactions among genes in determining phenotypes can make things complicated! Just list everything you can determine **for sure!**

Q6. A true-breeding plant is one that, when crossed with itself, always produces offspring with the same phenotype. In corn, there are actually multiple genotypes that can be true-breeding for each color. Using the tree diagram presented in Figure. 3.2, determine the number of genotypes that are true-breeding for each color, red, purple, yellow and white.

Q7. Calculate the expected phenotype frequencies of the offspring resulting from a tetrahybrid cross in which both parents are heterozygous for all genes: *PpCcRrYy* × *PpCcRrYy*). HINT: There is an easier way to do this without drawing a humongous Punnett square! Use Figure 3.2, what you have learned about expected offspring phenotype ratios from a monohybrid cross (a cross between two parents heterozygous for a single gene), and the rules of probability from earlier in this unit!

- Check your answers to these questions in the answer sheets in the back of this book!

Exercise 3b: Optional Computer Exercises

Exercise 3b.1: Different Types of Epistasis

As you have seen so far, several genes interact to determine the phenotype (color) of a kernel of corn. With so many different interactions going on, it is difficult to determine what contribution a single gene makes to the color of a corn kernel or the contribution of a particular interaction between two genes. Geneticists use hybrid crosses to separate the effects of gene interactions of interest from the full set of epistatic interactions.

In the **hybrid cross,** each parent is heterozygous (has two different alleles) for the gene of interest. For example, let's consider a simple **monohybrid cross** (involving just a single gene), with a dominant *A* allele, and a recessive *a* allele. A monohybrid cross would involve crossing two parents both with the genotype *Aa*, and would result in an expected phenotype ratio of 3 offspring displaying the dominant phenotype to 1 offspring displaying the recessive phenotype (and an expected genotype ratio of **1*AA*:2*Aa*:1*aa***).

To obtain an understanding of the interactions between just two of our four pigment genes determining corn color, we can simulate crosses between parents heterozygous for the genes of interest, either through the use of

Punnett squares, or, as we will do in this exercise, through the use of formulas incorporated into a spreadsheet program. The spread sheet analysis will permit us to examine the results of **dihybrid crosses** (involving parents heterozygous for two different genes), while holding the genotypes at the genes in which we are not interested constant (both of our simulated parents will have either homozygous dominant or homozygous recessive genotypes for these latter genes).

- Divide into groups. Your teacher should have provided each group with a Microsoft Excel file with the name "***CornTestCrosses.xls***", or "***CornTestCrosses.xlsx***".
- Double click on the icon representing this file on your computer's desktop.
- Once this file opens, you should see a spreadsheet containing instructions on how to code for the genotypes of hypothetical parents.
- Your teacher will assign each group one or more of the gene interactions of interest (from the table on the following page) to examine.
- Follow the instructions in the spreadsheet to code for the parent genotypes for the crosses in the table.
- Once you have coded the genotypes of two parents, you will see that in the area below your entry, the spreadsheet calculates the expected frequencies of each phenotype (kernel color) in the offspring produced by that cross.
- Record the expected phenotype frequencies of each phenotype for each cross considered, in a copy of the table on the following page, which your teacher should provide as a handout.
- **Remember, in considering the interactions between a set of genes:**
    1) Both parents should be coded as heterozygous (having two different alleles) for those genes,
    2) Both parents will possess the same genotype for the genes of interest.
    3) The parents should be homozygous for those genes in which you are not interested: homozygous dominant in one test cross, and homozygous recessive in a second test cross.

33

- After conducting your group's test crosses and filling out the table for those crosses, research the types of epistasis that exist (interaction between genes). The website "Epistasis and Its Effects on Phenotype" at http://www.nature.com/scitable/topicpage/Epistasis-Gene-Interaction-and-Phenotype-Effects-460 is a good starting point.
- Discuss what type(s) of epistatic interactions your cross(es) represent(s) within your group.
- After each group has finished their crosses, each group should present a brief summary of their results to the rest of the class, so your table can be filled out with information from each group's crosses.

Gene Interactions of Interest	Parent Genotypes	Genetic Background (Held Constant)	Expected Frequency of Purple Offspring	Expected Frequency of Red Offspring	Expected Frequency of Yellow Offspring	Expected Frequency of White Offspring	Type(s) of Epistasis Exhibited
RedAleurone1 & Colored1	PpCc	RRYY					
		rryy					
RedAleurone1 & Colored1	PpC'C	RRYY					
		rryy					
RedAleurone1 & Colored1	PpC'c	RRYY					
		rryy					
RedAleurone1 & Colored Aleurone1	PpRr	CCYY					
		ccyy					
RedAleurone1 & White1	PpYy	CCRR					
		ccrr					
Colored1 & ColoredAleurone1	CcRr	PPYY					
		ppyy					
Colored1 & ColoredAleurone1	C'CRr	PPYY					
		ppyy					
Colored1 & ColoredAleurone1	C'cRr	PPYY					
		ppyy					
Colored1 & White1	CcYy	PPRR					
		pprr					
Colored1 & White1	C'CYy	PPRR					
		pprr					
Colored1 & White1	C'cYy	PPRR					
		pprr					
ColoredAleurone1 & White1	RrYy	PPCC					
		ppcc					

Exercise 3b.2: Optional Computer Exercise 2 – Conducting Additional Test Crosses

Your teacher may assign several other test crosses for your group to conduct.

- Use the spreadsheet as described under Exercise **3b1** to calculate the expected phenotype ratios in a cross between each set of parental genotype pairings suggested by your teacher.

Exercise 3c: Open-ended Explorations /Group Project Ideas

Exercise 3c.1: Exploration/Project Idea #1

Your ears of corn in your trunk or those that you have encountered at the grocery store or in a fall display may have had some kernels which were mottled (speckled with a second color), or had some other color pattern on them. You may have also noticed shade variation among kernels in the white or yellow ears. Besides color, an ear of corn may have kernels of differing textures as in smooth versus wrinkled.

- Divide into teams and some online and/or library research to learn what might be the genetic basis behind these phenomena. Each team should focus on one of the alternatives.
- In the end, the teams researching a particular topic can get together and summarize their results for presentation to the class.

Exercise 3c.2: Exploration/Project Idea #2

- Working together in small groups of 3-4, purchase two ears of Indian corn (commonly used for fall decoration).
- Using the information that you have learned so far, as well as other techniques, such as dissecting several purple and/or red kernels from the ears (if present) to determine endosperm color, your group should construct a poster presenting an analysis of the potential genotypes of the parents that produced each ear (batch of offspring). Feel free to get creative, even attaching samples of the corn kernels to your poster!

- If you have computer access, you may also wish to use the spreadsheet from **Exercise 3b** to conduct some test crosses of potential parent genotypes to help you figure out how closely your observed phenotype ratios match up to expected phenotype ratios from a particular parental cross!

Exercise 3c.3: Exploration/Project Idea #3

Your teacher may provide your class with a list of several other organisms besides corn in which epistatic interactions determine phenotype.

- Divide the class into teams with each selecting an organism from the list.
- Using internet and/or library research, each group should find out what phenotype(s) in this organism are commonly known to have an epistatic basis, and give a brief report to the rest of your class on your findings.

Exercise 4: Human Genetics

NOTE TO TEACHERS: *Many human traits often used as examples to illustrate simple Mendelian (dominant/recessive) inheritance, such as the ability to roll the tongue, eye color, hair color, attached versus free earlobes, a widow's peak, dimples, a cleft chin, bent little fingers, a hitchhiker's thumb, and mid-digital hair, among others, are now known to have more complex mechanisms of inheritance, such as being the results of multiple genes (see the exercise on epistasis in this unit!), among other complex inheritance patterns. Unfortunately, however, many of these examples are still presented in the classroom as being simple, single-locus Mendelian traits. For a list of these traits, as well as many others, and some evidence refuting the myth of a Mendelian pattern of inheritance, check out the website* Myths of Human Genetics *in the "Links" section at the end of this workbook.*

NOTE TO TEACHERS #2: *This exercise represents a great opportunity for students to learn more about genetics, while also getting to spend time with and get to better know members of their families. If you ask your*

students to do this exercise, please be sensitive to the possibility of students who may be adopted, difficult family relationships, etc. In either case, the point is for students to learn more about genetics through an activity that also allows them to bond with important members of their families. Emphasize this to students in order to minimize student anxiety, allowing them to only make their pedigrees as extensive as they can without undue stress to themselves. Alternatively, you could allow students to pick a celebrity from a well-known or famous family, and have them do a pedigree of that individual.

Exercise 4a: Inherited Traits: a Genetic Coin Toss?

Do you have freckles? Whether or not you have freckles, as well as many other traits you possess, are the results of **dominant** and **recessive alleles** (different forms of the same **genes**) that are inherited from your parents and determined before you are born. A dominant allele is one that is always expressed (observed if at least one copy of it is present), and a recessive allele is one whose trait is only observed if a child gets two copies of the recessive allele (one from each parent). If a child gets a dominant allele from one parent, and a recessive allele from the other parent, the trait "coded" by the dominant allele "masks" the trait coded for by the recessive allele. For example, freckles are the result of a dominant allele, while a lack of freckles is a recessive trait. Many traits that humans possess are inherited. So, how likely is it that a person will end up with a certain trait?

In this lesson, you are going to learn how inherited traits are determined. You are going to toss coins to help you understand how some people have certain traits and others in the same family might have different traits depending on the combination of genes from the parents. Tossing coins results in different combinations depending on how the coins land. If you are tossing two coins, and each coin can land on either heads or tails, how many different combinations are possible? If you said 3, you are exactly right! You can have one coin land on heads and the other on heads. You could have one coin land on heads and the other tails, or you might have one coin land on tails and the other one tails, too.

- Find a partner to work with on this exercise.
- You and your partner will be tossing two coins at a time onto a flat surface and recording how they land.

- You will need to make a table so you can show tally marks in the correct column to indicate how the coins landed for each toss. The table should look something like the one below:

Heads-Heads	Heads-Tails	Tails-Tails
%	%	%

- Toss the pair of coins 50 times and record your results for each toss with a tally mark in the appropriate column. You may wish to toss the coins 25 times while your partner records the results, and then switch duties.
- If your first toss gives you a heads-tails combination, put a tally mark in the heads-tails column, and repeat that procedure for each of your 50 tosses.
- When you have tossed the pair of coins a total of 50 times, find the total number of tallies for each column. The three totals should have a sum of 50.
- Find the percentage of each combination of tosses.

For example, if 14 out of the 50 tosses were heads-heads, divide 14 by 50 to get 0.28 or 28%, meaning 28% of the total tosses were heads-heads.

Example: 14 out of 50 = 14 ÷ 50 = 0.28 = 28%

- Do the same for each total number of tosses for each of the three possible combinations.

Once you have your three percentages, the sum of those percentages should equal 100% to represent the whole set of tosses.

- Answer the following questions:
    - How do your percentages for the different combinations compare?
    - Do the three percentages vary much in size?
    - How do your percentages compare to other groups' percentages?
    - Are they similar?

In examining these percentages, what we are looking at is the **probability** of tossing a certain combination of heads and tails. In other words, we are getting an idea of how likely is it that your two coins will land on any of the three possible combinations of heads and tails on any given toss of the two coins.

The coin toss activity is related to inherited traits because whether or not you possess a certain trait depends on the combination of alleles from your parents, and how those alleles combined to give you that certain trait.   For any given trait, an individual has two alleles for that trait, inheriting one of these alleles from their mother, and the other allele from their father. You may get two dominant alleles, two recessive alleles, or one dominant and one recessive allele, depending on which alleles your parents have. For example, if one parent has only two recessive alleles, they can only pass on a recessive allele to each child they have. To link this to the coin toss activity, this would be like using a coin with two tails to represent that parent's possible allele contributions. However, if a parent has one dominant and one recessive allele, they could pass on either of those two to each child. This is why, for example, if you have one parent with freckles, and one parent without, you may have freckles while a brother or sister does not. The **probability** of having a particular trait depends on the combination of genes for that trait from your parents.

Exercise 4b: Comparing Family Traits

In this activity, you are going to explore the inheritance of traits by creating a description of yourself according to several traits. You are also going to list whether these traits are present in two other members of your family, like parents, grandparents, or other relatives. You will then compare the description of yourself to the descriptions of your family members, and use this information to help you think about how those traits might be inherited.

- On a blank sheet of paper, divide the sheet into four columns and draw lines down the paper to separate the columns.
- The first column will be the inherited traits you will compare between family members. Write "Traits" at the top of this column.
- Write your name at the top of the second column.
- Write the name of one family member at the top of the third column.

- Write the name of the other family member at the top of the fourth column.
- Under the column labeled "Traits," list several easily-observed traits that are of interest to you. Try to think of at least 3-5 traits to examine in yourself and your chosen family members.
- If you or a family member have a particular trait, write "yes" in the table in the row representing that trait.
- If you or a family member do not possess a certain trait, write "no" in the row representing that trait.
- Alternatively, if you are examining a trait that might take several forms (like hair color or eye color), you may wish to assign various simple descriptor words for these traits.
- Complete as much of the table as possible in class, then take the list home to complete for family members since you may need to be with the family members to examine their traits.
- Compare your findings with the class and share unusual or interesting things you found out about your family's traits.
- Use your results to help you think about the following questions:
  - Are there similarities among traits in your family?
  - What differences did you find?
- Now let's look back at the earlier example of freckles.
- Your teacher will poll the class to find out the number of students in the class with and without freckles.
- Look first at the number of students with freckles.
  - Does the number represent most of the class, or a small portion of the class?
- Using a ratio, compare the number of students with freckles to the total number of students.

For instance, if there are 28 students in the class and 7 of those students have freckles, the ratio will be 7:28.

- Using that ratio, find the percentage of students in your class that have freckles.

Example: Using the 7:28 ratio, we would divide 7 by 28, for a quotient of 0.25, which is 25%, meaning 25% of the students in your class have freckles.

- Now answer the following questions. You may need to do a bit of library and/or internet research to find the answers!

Q1. Are dominant traits always the most common traits in a population?

Q2. Why do you think this might be the case?

Q3. Did your results suggest that the traits you selected are inherited in simple dominant/recessive fashion, or do they possibly indicate a more complex pattern, or environmental influences?

Q4. How might it be difficult to answer the previous question above based on your results?

Q5. Can you think of a way that you could get a better idea of how a particular trait might be inherited?

- Check your answers in the answer section of this book to see if you were right!

Exercise 4c: Constructing a Pedigree (*Open-ended exploration*)

How do scientists know whether a trait is dominant or recessive, or if the trait is even inherited in a simple dominant/recessive way? One way of doing this is by constructing a **pedigree**. A pedigree is simply a type of chart that illustrates the presence or absence of a particular trait in individuals, as well as in members of their family.

Typically, pedigrees are tree-like diagrams, with different shapes (often circles and squares) representing individuals of different genders. Many pedigrees use circles to represent females, and squares to represent males. On a pedigree, some of the shapes are joined by lines to show relationships. A horizontal line connecting two shapes typically represents a mating between two individuals. A vertical line descending from the horizontal line representing the mating shows the next generation, made up of the mated couple's offspring. When examining a specific trait, the circles and squares representing individuals possessing a particular trait are usually colored in. By examining the occurrence of traits within families in a pedigree, scientists can often get a clue as to whether a trait is dominant or recessive, or if it is inherited in a different way. Below is an example of a pedigree.

First generation (grandparents)

Second generation (parents)

Third generation (grandchildren)
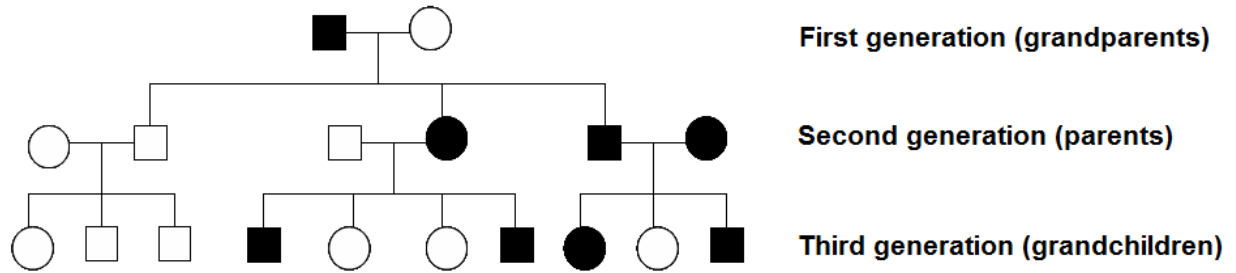
How does a pedigree give us clues about how a trait is inherited? As an example, in the pedigree pictured above, the trait of interest is likely a dominant trait, if it is inherited with a simple Mendelian basis. Notice that the trait of interest does not skip any generations. Also, if the trait was recessive, the parents represented by the dark square and dark circle could not have had the daughter without the trait.

- Pick a trait that is easily observed in individuals.
- Draw your own pedigree, showing the occurrence of your chosen trait in your family.
- Do not worry if you can't figure out information from too many generations back, or if it would be difficult for you to get information on particular family members. Families are all very different, and as a result, some students' pedigrees may be more extensive than others. Just do as much as you can, and with which you feel comfortable.
- Some creative ways of helping you fill in your pedigree might be to talk to your oldest living relatives, looking at old family photos, etc.
- After constructing your pedigree, look over it and examine the patterns of occurrence of your chosen trait over the generations represented in your chart. You may wish to use information from several exercises in this book to help you with your genetic detective work!
- Try to answer the following questions:
  - o Do you think your chosen trait is dominant or recessive, or do you think it might be inherited in a more complex way?
  - o Can you find other pedigrees (online, in the library, etc.) that show similar patterns that support your conclusions?
- If your chosen trait seems to be one that is inherited in a more complex way, do a little additional research on that particular mode of inheritance.
- Put together what you learned, and share it with the rest of your class in a brief presentation.

ANSWER SHEETS

ANSWERS FOR EXERCISE 1

Q1. Approximately, how many genes are on a chromosome?
  200,000,000/100,000 = 2000 genes

Q2. What are the complementary base pairs?
  Adenine and Thymine
  Guanine and Cytosine

Q3. The number of A, G, C, and T bases in a gene will vary between DNA molecules, but one base will always be present in the same amount as A (adenine). Which base is it?
  Thymine

Q4. Which base is present in the same amount as G (guanine) in the DNA molecule?
  Cytosine

Q5. Here's a more difficult question. You are a real scholar if you get it right! Let
  $A = the\ number\ of\ adenine\ molecules\ in\ a\ molecule\ of\ DNA$,
  $C = the\ number\ of\ cytosine\ molecules\ in\ a\ molecule\ of\ DNA$,
  $T = the\ number\ of\ thymine\ molecules\ in\ a\ molecule\ of\ DNA$,
  $G = the\ number\ of\ guanine\ molecules\ in\ a\ molecule\ of\ DNA$.
  Complete the following equation:
$$A + C = T + G$$
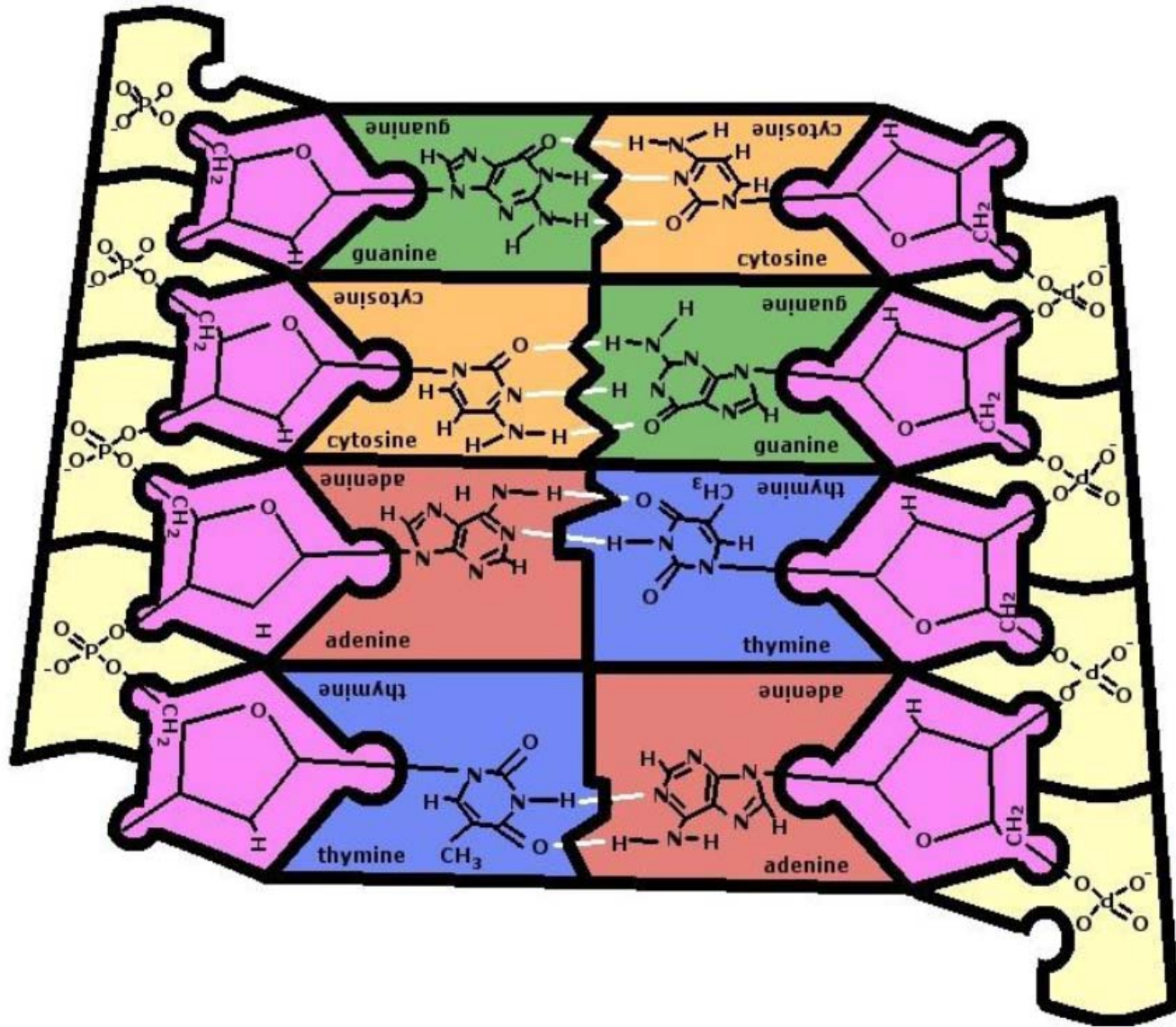
ANSWERS FOR EXERCISE 2

Q1. How many homologous pairs are in the nucleus?
There are three homologous pairs in the nucleus.

Q2. How many autosomes are in the nucleus?
There are six autosomes in the nucleus.

Q3. How many sex chromosomes are in the nucleus?
There are two sex chromosomes in the nucleus.

Q4. How many chromatids are in the nucleus?
There are 16 chromatids in the nucleus.

Exercise 2a: Basic Probability

To every experiment there corresponds a set of possible outcomes, called the **sample space** of the experiment. We will denote the sample space of an experiment by the letter S. In our case the experiment of interest is sexual reproduction. The sample space of this experiment is the set of all allele combinations that the resulting offspring could have.

Q1. Suppose that a man and a woman with genotypes Bb and BB reproduce. What is the sample space of this experiment?
$$S = \{BB, Bb\}$$

Q2. Suppose that a six sided die is rolled. What is the sample space of this experiment?
$$S = \{1,2,3,4,5,6\}$$

Q3. Draw the Punnett square for the experiment from question Q1.

	B	b
B	BB	Bb
B	BB	Bb

Q4. Use your Punnett square to find the number of ways that each of the genotypes from question Q1. can be produced.
There are two ways that the genotype BB can be produced, and two ways that the genotype Bb can be produced.

Q5. Can you think of any other ways to describe the event E?
E is also the event that the child has at most one b allele.

The sets $\{Bb\}, \{BB\}, \{bb\}, \{BB, bb\}, \{Bb, bb\}$ and $\{Bb, BB, bb\}$ are other possible events for this experiment. Events such as $\{bb\}$ that contain a single element are called **elementary events**. If two events contain none of the same elementary events then they are said to be **mutually exclusive** events. For example, $\{Bb, BB\}$ and $\{bb\}$ are mutually exclusive events.

Q6. Suppose that you roll a six sided die. Let E denote the event that you roll less than a five. Write down all of the elements that belong to the event E.
$$E = \{1,2,3,4\}$$

Q7. Let B be the event that you roll $1, 4$ or 6, that is, let $B = \{1, 4, 6\}$. Are B and E mutually exclusive? If not, which elementary events belong to both B and E.
No E and B are not mutually exclusive because the elementary events $\{1\}$ and $\{4\}$ belong to both E and B.

Q8. Find three possible events of the experiment in question Q1. Also find a pair of mutually exclusive events.
$\{BB\}, \{BB, Bb\}$ and $\{Bb\}$ are three events that belong to the experiment in question Q1. $\{BB\}$ and $\{Bb\}$ are mutually exclusive.

Q9. Suppose that there are three pennies in your pocket. One of the pennies was made in 1991, another was made in 1985, and the third penny was made in 2006. Now suppose that you pull one penny from your pocket (without looking). Are you equally likely to pick any of the pennies? Explain why or why not.
Yes, you are equally likely to pick any of the pennies, because they all feel the same. That is, they have the same size, shape, and texture.

Q10. What is the probability that you pick the penny from 1985? What is the probability that you don't pick the penny from 1985?

The probability that you pick the penny from 1985 is $\frac{1}{3}$. The probability that you don't pick a penny from 1985 is $\frac{2}{3}$.

Q11. Suppose that a woman and a man with genotypes Bb and BB reproduce. What is the probability that their child has genotype BB?

$$\frac{1}{2}$$

Q12. Let E be the event from **Q11.**   Use Rule 2 to find $P(\bar{E})$.

$$P(\bar{E}) = 1 - P(E) = 1 - \frac{1}{2} = \frac{1}{2}$$

Q13. Suppose the two coins are tossed. Let A be the event that the first coin is heads and B be the event that the second coin is heads. Are A and B are independent?

Yes, because the first coin does not affect the second coin in any way.

Q14. Suppose that two children attend the same daycare. Let A be the event that the first child catches a cold and B be the event that the second child catches a cold. Are the events A and B independent?

No they are not independent. If one child gets a cold the other child is more likely to catch a cold too because colds are contagious and the children attend the same daycare.

Q15. Find the probability that A and B occur, where A and B are the events from Q13.

$$P(A \ and \ B) = \frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$$

Exercise 2b: Law of Segregation – Offspring are equally likely to inherit either of their parent's alleles.

Q1. What is the probability that you drew a red allele from the female parent box? Explain how you came to this answer.

You choose a red allele with probability $\frac{1}{2}$ (which is equal to 0.5) because half of the chips in the female box are red.

Q2. Suppose that when choosing the genotype of the first offspring, you drew a red chip from the female parent's box, and did not replace this chip before the second draw. What is the probability that the second offspring receives a red allele?

The second offspring receives a red allele with probability $\frac{5}{11}$.

Q3. Based on the probability that an individual has genotype CC, how many offspring would you expect to have genotype CC? What about $C'C'$ and $C'C$?

Since ¼ of the offspring are expected to have genotype CC, and there are 16 offspring total, you would expect $\frac{1}{4}(16) = 4$ of the offspring to have genotype CC. Similarly you would expect 4 of the offspring to have genotype $C'C'$, and 8 of the offspring to have genotype $C'C$.

Q4. How close are the actual frequencies to the frequencies that you predicted based on probability? Why wouldn't they be the same?

Your actual frequencies are probably not equal to the predicted frequencies because the probability that an outcome occurs only measures how likely it is that the outcome occurs, not what fraction of the time the outcome actually will occur.

Q5. Find the probability that an individual offspring is not homozygous white.

$$\frac{3}{4}$$

Answers to Exercise 2c: Mendel's Law of Independent Assortment

Q1. Can you think of two car features that are not linked?

Color and gas mileage, or color and size are two possible examples.

Exercise 2c.1: Tree diagrams and the counting principle.

Q2. Suppose that you have three shirts (green, orange and yellow) and two pairs of pants (blue and brown). How many different outfits can you assemble from these clothing items?

$$3 \times 2 = 6$$

Q3. What is the probability that your shirt is yellow or orange?

$$\frac{4}{6} = \frac{2}{3}$$

Q4. How many different tasks are involved in determining the genotype of an offspring in this cross? List each of these tasks.
There are four tasks involved in the determination of an offspring's genotype, as follows:
1. The female parent must donate an allele for color.
2. The female parent must donate an allele for size.
3. The male parent must donate an allele for color.
4. The male parent must donate an allele for size.

You could have also said that there were two tasks involved: the production of a gamete by the female parent, and the production of a gamete by the male parent. However, just keep in mind that production of each gamete includes the donation of an allele for both color **and** size, which would mean that each of the "gamete production" tasks could have more possible ways that they could be performed, each involving a combination of size and color alleles!

Q5. In how many ways can each of the tasks from the previous question be performed?
1. Female parent's donation of color allele: 2 ways (*C* or *C'*)
2. Male parent's donation of color allele: 2 ways (*C* or *C'*)
3. Female parent's donation of size allele: 2 ways (*T* or *T'*)
4. Male parent's donation of size allele: 2 ways (*T* or *T'*)

If you considered this to be two tasks, the production of a gamete by each parent, the total number of ways each task can be performed are as follows:
1. Female parent's gamete produced: 4 ways (*CT*, *CT'*, *C'T*, *C'T'*)
2. Male parent's gamete produced: 4 ways (*CT*, *CT'*, *C'T*, *C'T'*)

Q6. Construct a tree diagram to show all of the possible outcomes of this cross.

```
                            T→CCTT
                     T <
                            T'→CCTT'
              C <
                            T→CCTT'
                     T' <
                            T'→CCT'T'
       C <
                            T→CC'TT
                     T <
                            T'→CC'TT'
              C' <
                            T→CC'TT'
                     T' <
                            T'→CC'T'T'
C <
                            T→CC'TT
                     T <
                            T'→CC'TT'
              C <
                            T→CC'TT'
                     T' <
                            T'→CC'T'T'
       C' <
                            T→C'C'TT
                     T <
                            T'→C'C'TT'
              C' <
                            T→C'C'TT'
                     T' <
                            T'→C'C'T'T'
```
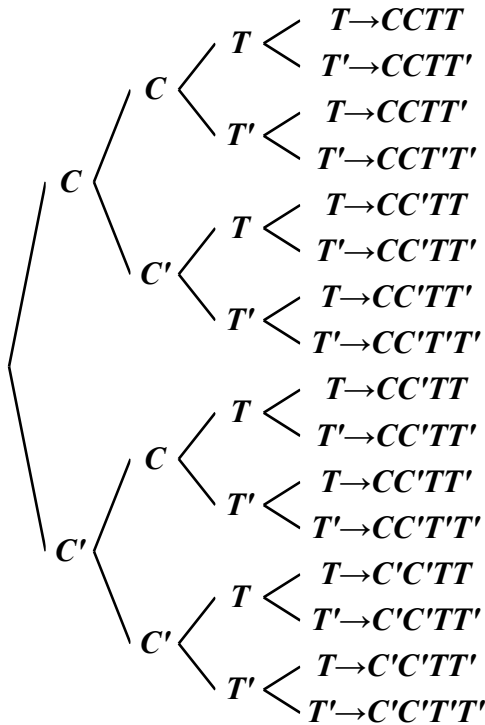
In the tree diagram above, the first branch point represents the possible alleles for color donated by one parent. The next set of branches represents the possible alleles for color donated by the other parent, given the allele for color donated by the first parent. The next set of branches represents possible alleles for thickness donated by one parent, given the alleles donated by both parents for color, and the final set of branches represents the possible alleles for thickness donated by the other parent, given the alleles donated by both parents for color, and the allele for thickness donated by the first parent. Finally, the outcomes after the arrows represent the resulting genotypes of the offspring in each scenario.

Q7. How many outcomes are possible for this experiment?
There are a total of 16 possible outcomes for this experiment.

Q8. How many possible genotypes could the offspring produced by this cross have? (Remember that the genotype is the set of alleles that an individual has with no regard to which parent donated which allele.)
As you can see in the tree diagram above, even though there are sixteen possible outcomes, some of those outcomes are the same, but just represent

different ways that those outcomes could have occurred. There are only nine possible offspring genotypes (**CCTT**, **CCTT'**, **CCT'T'**, **CC'TT**, **CC'TT'**, **CC'T'T'**, **C'C'TT**, **C'C'TT'**, **C'C'T'T'**) from the cross of two parents both with the genotype **CC'TT'**, but some of these outcomes are more likely than others!

Q9. Let A be the event that the offspring has at least one C allele, and B be the event that the offspring has at least one T allele. Use the tree diagram to find $P(A)$, $P(B)$, $P(A \text{ and } B)$, and $P(A)P(B)$. Do your answers support Mendel's Law of Independent Assortment? That is, do they support the hypothesis that alleles for the color and thickness genes are inherited independently? Explain why or why not.
You should have found in all cases that $P(A)P(B) = P(A \text{ and } B)$. Recall that this is equivalent to the statement that the events A and B are independent.

Q10. (Critical thinking!) Why was it important that you made sure that both the IF and IM boxes contained equal numbers of thick red, thin red, thick white, and thin white chips?
The reason that it was important for there to be equal numbers of each of these types of chips is because if there had been, for example, one less thick white chip, this would have represented a situation where the two traits (color and size) did NOT assort independently, which was not the point of this exercise! In other words, if you were one thick white chip short, this would have represented that the red (C) and thick (T) alleles are more likely to go together during gamete production (which, if you recall, does happen sometimes with certain genes!).

Exercise 2d: There are genotypes and then there are phenotypes.

Q1. What color are the kernels on the F1 ear of corn?
*Answer may vary, based on the colors of corn provided in this unit!

Q2. Why are they this color?
They are all this color because they are heterozygous, and the allele for the color they display is dominant over the allele for the color encoded by the allele donated from the parent of the other color.

Q3. What are the genotypes of the F1 offspring?
$$Cc$$

Q4. What are the phenotypes of the F1 offspring?
  The phenotypes of all F1 offspring are the same color as one of the parents (which was homozygous for the allele representing the dominant color).

Q7. Based on your Punnett square, what fraction of the kernels would you expect to be of the first color? What fraction of the kernels would you expect to be of the second color? What is the expected ratio of kernels of one color to kernels of the other color?

$$\frac{3}{4}, \quad \frac{1}{4}, \quad 3:1$$

ANSWERS FOR EXERCISE 3

Q1. In the previous exercise, you may have guessed that kernel color in corn involves a single gene with two alleles, and a system of simple dominance. In fact, using the parental, *F1*, and *F2* offspring ears, this appears to be the easiest explanation. Explain, in terms of genotypes and phenotypes, what may have led you to this original conclusion.
  In your sample *F1* ear of corn, you should have noticed that all of the offspring (individual kernels) were all of the same color. This should have led you to believe that each of the parents were homozygous: one homozygous for the dominant allele for one color, and one homozygous for the recessive allele for the other color. Additionally, in your *F2* ear, which possesses offspring (individual kernels) of both colors, you might have noticed that the ratio of kernels of the "dominant" color to the "recessive" color were present in a ratio of approximately 3:1, which are the phenotype ratios we would expect in the outcome of a monohybrid cross (a cross between two *F1* individuals, which would be heterozygous for the allele for color).

Q2. Using the tree diagram on the previous page, calculate the total number of genotypes that will result in each phenotype (kernel color).
  Purple and red are the easiest to calculate, since they only appear once on the diagram, so let's calculate those first. There are two different genotypes that a purple kernel can have at the *RedAleurone1* gene (**PP** or **Pp**), two genotypes a purple color can have at the *Colored1* gene (**CC** or **Cc**), 2 genotypes a purple color can have at the *ColoredAleurone1* gene

(**RR** or **Rr**), and 3 genotypes a purple color can have at the *White1* gene (**YY**, **Yy**, or **yy**). Thus, the total number of genotypes that would result in the purple phenotype is equal to $2 \times 2 \times 2 \times 3 = 24$ possible genotypes.

We would calculate the total number of genotypes resulting in the red phenotype in the same way. However, there is only one possible genotype at the *RedAleurone1* gene resulting in a red phenotype (**pp**), so there are $1 \times 2 \times 2 \times 3 = 12$ genotypes that would result in a kernel being red.

For the yellow phenotype, we calculate the answer in a very similar manner. However, we also have to do a bit of adding, since the yellow color appears more than once on our diagram. Starting with the *RedAleurone1* gene having a genotype of **PP** or **Pp** (2 different possibilities) AND the *Colored1* gene having a genotype of either **CC** or **Cc** (2 different possibilities), there is only one possible genotype at the *ColoredAleurone1* gene (rr), and two possible genotypes at the *White1* gene (**YY** or **Yy**) that would result in a yellow phenotype, so following that "path" on our tree diagram, there are $2 \times 2 \times 1 \times 2 = 8$ possible genotypes. However, we also have to consider all the other possible "paths" leading to a yellow phenotype. If a kernel also has either genotype **PP** or **Pp** at the *RedAleurone1* gene (2 possibilities), AND a genotype of either **cc**, **C'C**, **C'c**, or **C'C'** at the *Colored1* gene (4 possibilities), there are three possible genotypes at the *ColoredAleurone1* gene (**RR**, **Rr**, or **rr**), and 2 possible genotypes at the *White1* gene (**YY** or **Yy**) resulting in a yellow phenotype, for a total of $2 \times 4 \times 3 \times 2 = 48$ additional genotypes resulting in a yellow kernel. We can also do this for the paths leading to the yellow phenotype if the genotype at the *RedAleurone1* gene is pp: $1 \times 2 \times 1 \times 2 = 4$, AND $1 \times 4 \times 3 \times 2 = 24$. Adding all of these numbers up, we can see that there are $8 + 48 + 4 + 24 = 84$ possible genotypes that would result in a yellow phenotype. Similarly, for the white phenotype, there are $(2 \times 2 \times 1 \times 1) + (2 \times 4 \times 3 \times 1) + (1 \times 2 \times 1 \times 1) + (1 \times 4 \times 3 \times 1)$, or $4 + 24 + 2 + 12 = 42$ possible genotypes that would result in a kernel that is white in color. The table on the following page presents a summary of the number of possible genotypes resulting in each color.

Phenotype (kernel color)	Number of possible genotypes
Purple	24
Red	12
Yellow	84
White	42

In addition, we can check these answers to make sure that all possible genotypes are represented. There are a total of 3 possible genotypes (*PP*, *Pp*, or *pp*) at the *RedAleurone1* gene, six possible genotypes at the *Colored1* gene (*CC*, *Cc*, *cc*, *C'C*, *C'c*, or *C'C'*), three possible genotypes at the *ColoredAleurone1* gene (*RR*, *Rr*, or *rr*), and three possible genotypes (*YY*, *Yy*, and *yy*) at the *White1* gene, for a total possible number of overall genotypes of $3 \times 6 \times 3 \times 3 = 162$ possible overall genotypes. The number of possible genotypes resulting in each phenotype should add up to this same number if we have represented all possible genotypes resulting in each phenotype (and have not accidentally counted duplicates), and we can see that it does: $24 + 12 + 84 + 42 = 162$.

Q3. Imagine that you are given a single kernel of corn of a particular color. Based on its phenotype, can you determine its genotype (at each of the four previously discussed genes)? For which genes would you be able to certainly know the exact genotype for kernels of a particular color? Are there any genes for which you would be unable to completely deduce both alleles making up the genotype for that gene for each particular color? Why is this the case?
Normally, if we were examining a trait controlled by only one gene (let's use a gene with two alleles, *A* and *a*, as an example), we would only know the genotype of the organism for that gene if it displayed the phenotype resulting from being homozygous recessive for that gene, because then we would know that the organism has to have two recessive alleles for that gene (genotype *aa*). If the organism displayed the dominant phenotype, we know that it would have at least one *A* allele, so it could actually be either genotype *AA* or *Aa*. We could thus write its genotype, showing our level of uncertainty as *A?*, where the question mark shows that we are not certain what the other allele possessed by the organism actually is.

Things are slightly more complicated with corn, since kernel color is controlled by interactions between multiple genes. If we were to

examine a single kernel of corn of a particular color, it would be impossible to know its entire genotype for each of the genes mentioned in this exercise. The only genotypes we would be able to know for certain would be the genotype at the *RedAleurone1* gene for a red kernel (*pp*), or the genotype at the *White1* allele for a white kernel (*yy*), because the only way that either of these colors would be expressed would be if they had the homozygous recessive genotypes at the previously mentioned genes. The red kernel could have several genotypes at the *Colored1* and *ColoredAleurone1* genes, but cannot be homozygous recessive for either gene, or cannot have a *C'* allele for the *Colored1*, and may have any genotype at the *White1* gene. Thus, we could write the possible genotypes of a red kernel as *ppC?R???*. The white kernel could have any genotype at the *RedAleurone1* gene, and several different genotypes at the *Colored1* and *ColoredAleurone1* genes (as long as it is either homozygous recessive for either of those genes, AND/OR has at least one *C'* allele for the *Colored1* allele. Thus, we could write the possible genotypes for a white kernel as *??????yy*. All we would know about a yellow kernel is that it has at least one *Y* allele for the *White1* gene (and could be homozygous recessive at the *Colored1* gene, *cc*, and/or the *ColoredAleurone1* gene, *rr*, AND/OR have at least one *C'* allele for the *Colored1* gene). We could thus denote what we know for certain about a yellow kernel's genotype as *??????Y?*. A purple kernel would have to have at least one *P* allele, at least one *C* allele, AND at least one *R* allele. We could thus write what we know about its genotype as *P?C?R???*.

Q4. Can you think of some ways to help determine potentially unknown genotypes at certain genes of a particular kernel of corn, aside from actually examining the kernel's DNA?
One possibility would be to look not only at the kernel of interest, but also at its siblings, or other kernels from the same cob, if they were available. By examining the phenotypes that are present in that particular batch of offspring, as well as the ratios in which those phenotypes were present, you could get a better idea of what alleles were contributed to each offspring by the parent plants that produced the ear, and thus a better idea of the possible genotypes of the offspring of each particular color.

However, if you were given only a single corn kernel, you could grow an adult plant from the kernel, and when mature, cross it with some other

corn plants of known genotypes. It would be particularly helpful if you were to cross the plant you grew with a plant that "breeds true" for a color that is expressed only when kernels are homozygous recessive at the allele resulting in the production of that color (such as red, which has the genotype *pp* at the *RedAleurone1* gene, or white, which has the genotype *yy* at the *White1* gene). By conducting this **test cross**, you would be able to tell if your mystery kernel had at least one recessive allele for either of those genes if you saw either red or white kernels in the offspring produced. You could also further conduct crosses between two individuals from the offspring, cross the offspring with either of the parent plants, or conduct other crosses with plants of known genotypes, and look at the phenotypes (and their ratios) in the resulting offspring to help you figure it out!

Also, if your kernel was either purple or red, you would not know anything about its genotype at the *White1* gene, because the purple or red pigmentation in the aleurone masks the color of the endosperm inside. You could actually dissect the kernel to see what color its aleurone happened to be, at least letting you narrow down the possible genotypes of the kernel at that gene (and giving you a definite answer if the endosperm was white!).

Q5. Examine the ear of corn labeled "Mystery Parents" in your box. Based on the phenotypes of the kernels present, what can you deduce about the genotypes of the plants that were crossed to produce this ear? You may wish to consult Figure 4.2, as well as play around with a few Punnett squares to help you with this question! Don't get discouraged if you can't figure out everything about the possible genotypes of the parents, as the nature of epistatic interactions between genes in determining phenotypes can make things complicated! Just figure out everything that you are able to know for sure!
This answer will vary, based on the phenotypes present on your "Mystery Ear" of corn!

Q6. A true-breeding plant is one that, when crossed with itself, always produces offspring with the same phenotype. In corn, there are actually multiple genotypes that can be true-breeding for each color. How many genotypes would be true-breeding for each color?

Red and purple true-breeding plants have the fewest number of total possible genotypes, with three possible genotypes each. For a plant to be true-breeding for red kernels, it has to have the genotype *pp* at the RedAleurone1 gene, *CC* at the *Colored1* gene, and *RR* at the *ColoredAleurone1* gene. However, it can be either homozygous dominant (*YY*), heterozygous (*Yy*), or homozygous recessive (*yy*) at the *White1* gene. The genotype at the *White1* gene does not matter, because the effects of the homozygous dominant condition at all of the other genes determining color would always produce red pigment that masked the color of the aleurone.

The same holds for true-breeding purple plants, except they must have the genotype **PPCCRR** at the previously mentioned genes. True-breeding purple plants could not have the genotype *Pp* at the RedAleurone1 gene, because if so, when crossed with itself, a plant with such a genotype would have the potential to produce red offspring (*pp*).

White and yellow true-breeding plants have 30 possible genotypes each. White true-breeding plants must be homozygous recessive at the *White1* gene. They could have any genotype (*PP*, *Pp*, or *pp*) at the *RedAleurone1* gene, but would have to have either two recessive alleles or two *C'* alleles at the *Colored1* gene, AND/OR two recessive alleles at the *ColoredAleurone1* gene. The possible genotypes at these genes would thus be **ccRR**, **ccRr**, **ccrr**, **C'C'RR**, **C'C'Rr**, **C'C'rr**, **CCrr**, **Ccrr**, **C'Crr**, or **C'Crr**. Any of these genotypes, when crossed with themselves, would always result in offspring that are homozygous recessive for either (or both) of the two genes, or of having at least one *C'* allele, all of which would result in no production of red or purple pigment. Since there are 10 different genotype combinations at these two genes, and three possible genotypes at the *RedAleurone1* gene, there are a total possible of $10 \times 3 = 30$ possible genotypes for a true-breeding white plant.

This would be similar for a true-breeding yellow plant, except we know that the true-breeding yellow plant would have to have the genotype *YY* at the *White1* gene for the offspring of a cross with itself to always be yellow.

Q7. Calculate the expected phenotype frequencies of the offspring resulting from a tetrahybrid cross (both parents are heterozygous

for all genes: *PpCcRrYy* × *PpCcRrYy*). HINT: There is an easier way to do this without drawing a humongous Punnett square! Use Figure 4.2, what you have learned about expected offspring phenotype ratios from a monohybrid cross (a cross between two parents heterozygous for a single gene), and the rules of probability from earlier in this unit!

Using what we know about a monohybrid cross (expected genotype frequencies of 0.25 homozygous dominant, 0.50 heterozygous, and 0.25 homozygous recessive, which are all **probabilities!**), as well as the rules of probability outlined earlier in this unit:

(P(A **and** B) = P(A) × P(B), if A and B are not mutually exclusive
　　AND
P(A **or** B) = P(A)+P(B)),

We can combine this information with the technique used in **Q2** to get the answer. For example, for an offspring to be red, we know it has to be homozygous recessive for the *RedAleurone1* gene (*pp*, probability = 0.25), However, it would **also** have to be either homozygous dominant (probability = 0.25) **OR** heterozygous (probability = 0.50) at the *Colored1* gene, AND would have to be either homozygous dominant (*RR*, probability = 0.25) **OR** heterozygous (*Rr*, probability = 0.50) for the *ColoredAleurone1* gene, **AND** could be either homozygous dominant (*YY*, probability = 0.25), **OR** heterozygous (*Yy*, probability = 0.50), **OR** homozygous recessive (*yy*, probability = 0.25) for the *White1* gene. Thus, the expected frequency of the red phenotype in the offspring from a tetrahybrid cross would be equal to (0.25) × (0.25 + 0.5) × (0.25 + 0.50) × (0.25 + 0.50 + 0.25), which reduces to (0.25) × (0.75) × (0.75) × (1.00) = 0.140625 (which is equal to $\frac{9}{64}$ in fractional form). Using similar techniques (which involves a bit of adding for the yellow and white phenotypes, since they occur multiple times in our tree diagram), you should arrive at the following answer:

Phenotype (Color)	Proportion	Percent	Fractional Form	Ratio
Purple	≈ 0.4219	42.19%	27/64	27
Red	≈ 0.1406	14.06%	9/64	9
Yellow	≈ 0.3281	32.81%	21/64	21
White	≈ 0.1094	10.94%	7/64	7

ANSWERS FOR EXERCISE 4

Q1: Are dominant traits always the most common traits in a population?
Though it is a common misconception, dominant traits are not always necessarily more common than recessive traits. Many people are confused by the terms "dominant" and "recessive", and assume that "dominant" traits are ones that are always most common. However, these terms refer ONLY to how the traits are inherited, and not how prevalent they are in a population!

Q2: Why do you think this might be the case?
If a recessive allele is very common in a population, then the recessive trait will most likely be more frequently observed than the dominant trait. Also, consider certain diseases, or abnormalities caused by deleterious (harmful) mutations. Many of these are encoded by dominant alleles. However, the frequency of the dominant allele is then typically reduced dramatically due to selection against it. In this way, a trait "coded for" by a dominant allele can still be very rare in a population!

Q3: Did your results suggest that the traits you selected are inherited in simple dominant/recessive fashion, or do they possibly indicate a more complex pattern, or environmental influences?
Your answer might vary substantially here, depending on the traits you chose, as well as the family members whose traits you chose to examine.

Q4: How might it be difficult to answer the previous question above based on your results?
Though the small amount of data you collected on your chosen traits can be informative, it is unlikely to be enough evidence to support a description of the mechanisms of inheritance of your chosen traits.

Q5: Can you think of a way that you could get a better idea of how a particular trait might be inherited?
Though modern knowledge and technological advances in genetic analysis definitely facilitate further determination of how traits are inherited, one way that this question has been addressed for many years is through the construction of a **pedigree**, or a chart that shows relationships of individuals (usually of several generations), and the occurrence/absence of those traits indicated on the individuals represented in the chart. Constructing pedigrees

can be very informative, and can help deduce such things as whether a person possessing a dominant trait was homozygous or heterozygous for the trait, whether a trait is inherited in a simple Mendelian dominant/recessive fashion, whether a trait is sex-linked, among many other things. In Exercise 4c, which is an open-ended exploration, you are given further information on how pedigrees are constructed, and are given the chance to construct pedigrees of your own family, and/or of a celebrity.

SUGGESTED READING

Grades K-3
You're Full of Genes - Claudia Zylberberg
Gregor Mendel: The Friar Who Grew Peas - Cheryl Bardoe

Grades 4-7
Have a Nice DNA - Frances R. Balkwill and Mic Rolph
Double Talking Helix Blues - Joel Herskowitz & Judy Cuddihy (Illustrator)
National Geographic Investigates: Genetics: From DNA to Designer Dogs - Kathleen
     Simpson
Amazing Schemes Within Your Genes - Frances R. Balkwill and Mic Rolph (Illustrator)
Have a Nice DNA – Frances R. Balkwill and Mic Rolph (Illustrator)
Crime Scene: True-life Forensic Files #1: Dusting and DNA - D.B. Beres and Anna
     Prokos
They Came from DNA - Billy Aronson & Danny O'Leary (Illustrator)
Gregor Mendel: Father of Genetics - Roger Klare
Gregor Mendel: *The Friar Who Grew Peas* – Cheryl Bardoe

Grades 7+
The Cartoon Guide to Genetics - Larry Gonick & Mark Wheelis
The Human Genome Project: Cracking the Code Within Us - Elizabeth L. Marshall
New Genetics: The Study of Lifelines - Jerry S. Kidd & Renee A. Kidd
Genetic Engineering: Progress or Peril? - Linda Tagliaferro
Diabetes (Genes and Disease) - Toney Allman
Genetics and Genetic Engineering - Lisa Yount
Genetics and Evolution: The Molecules of Inheritance - Jill Bailey
DNA Analysis (Forensics: The Science of Crime-Solving) - William Hunter
Guilty By a Hair!: Real-life DNA Matches! - Anna Prokos (**CAUTION:** Contains
     information on cases involving violent crime, such as murder.)
The Making of the Fittest: DNA and the Ultimate Forensic Record of Evolution – Sean B.
     Carroll
DNA: *The Secret of Life* – James D. Watson & Andrew Berry
The Double Helix - James D. Watson
Understanding DNA: The Molecule & How It Works - Chris R. Calladine, Horace Drew,
     Ben Luisi, and Andrew Travers (actually a college textbook)

Scientific Journal Articles (included on Teacher CD!)
Han, B. And D. L. Denlinger. 2009. Mendelian inheritance of pupal diapause in the flesh
     fly, *Sarcophaga bullata*. *Journal of Heredity* 100(2):251-255.
Mendel, Gregor. 1866. Versuche über Plflanzenhybriden (Experiments in Plant
     Hybridization). *Verhandlungen des naturforschenden Vereines in Brünn, Bd. IV*
     für das Jahr 1865, Abhandlungen, 3–47. (An updated English translation of the
     original is provided!)
Pennisi, E. 1996. Studly sheep by non-Mendelian means. *Science*, New Series
     272(5265):1099-1100.

Watson, J.D. and F.H.C. Crick. 1953. Molecular structure of nucleic acids – A structure for deoxyribose nucleic acid. *Nature* 171(4356):737-738.

LINKS

The Gene Scene - The Genetics homepage on the American Museum of Natural History's OLogy site for kids. Lots of great introductory information on genetics, DNA, heredity, and other related topics. Includes several crafts, as well as an directions for extracting a clump of goopy DNA from an onion, using only simple household materials!
>   http://www.amnh.org/ology/index.php?channel=genetics#channel

Biology4Kids.com: Cell Structure - Great site with information on cell structure and function, including discussions of DNA, chromosomes, etc. (Particularly check out the sections on the nucleus, chromosomes, centrioles, and ribosomes!)
>   http://www.biology4kids.com/files/cell_main.html

Understanding Genetics: Human Health and the Genome - A great website from The Tech Museum of Innovation and Stanford School of Medicine; also check out the nifty "Ask a Geneticist" section!
>   http://www.thetech.org/genetics/asklist.php

The Structure of the DNA Molecule - This "Access Excellence" website from the National Health Museum contains a brief (but informative) history of Mendel's experiments, as well as Watson and Crick's discovery of the shape of the DNA molecule, as well as a glossary, nice graphics, and a few classroom activities.
>   http://www.accessexcellence.org/AE/AEC/CC/DNA_structure.php

Patterns of Inheritance - Good website providing basic genetics terminology, information on the genetics of eye color in humans, a "genetic puzzle" to determine a family's genotypes based on phenotypes, and details on formation of gametes during meiosis.
>   http://www.windows.ucar.edu/tour/link=/earth/Life/genetics_inheritance.html

Basic Principles of Genetics - A good overview site by the Behavioral Sciences Department at Palomar College, including great information on basic Mendelian genetics, the probability of inheritance (includes a good intro to the use of Punnett squares), and exceptions to simple inheritance. Also includes practice quizzes, flashcards, and a fun crossword puzzle, as well as a glossary and links.
>   http://anthro.palomar.edu/mendel/Default.htm

Genome: The Secret of How Life Works - A visually appealing and informative site from Pfizer, which includes "informative and interactive content for anyone with a genome." Yes, that means you. Includes games, quizzes, and even lesson plans at both the elementary and high school levels, as well as a nice compilation of links. The entire website is also available in Spanish.
>   http://genome.pfizer.com/index.cfm

Learn.Genetics - Beautiful and HUGE website from the University of Utah containing information on genetic technology, virtual labs, resources and lesson plans for teachers,

information on genetic disorders, epigenetics, and even such interesting topics as molecular genealogy and the genetics of addiction.
http://learn.genetics.utah.edu/

Genetics Education Center - A great comprehensive compilation of links from the University of Kansas Medical Center, including multiple links on the Human Genome Project, genetic education resources, activities, museums with genetics exhibits, and more!
http://www.kumc.edu/gec/

Patterns of Inheritance - Good information with details on mechanisms of inheritance, including dominance and recessiveness, codominance, and multiple alleles, in the context of budgerigars (parakeets) and other parrots, from BirdHobbyist.com. May be of particular interest to those with pet birds at home!
http://www.birdhobbyist.com/parrotcolour/patterns01.html

Feline Genome Project: Coat Colors & Fur Types - Have cats at home? Ever wondered what color kittens they could have? This site, from the College of Veterinary Medicine at UC Davis, is the place to find out.
http://www.vetmed.ucdavis.edu/phr/lyonsden/color.htm

Dog Coat Color Genetics - Since we mentioned pet genetics, we couldn't leave out man's best friend! This site from the University of Saskatchewan gives "a brief review of the genes controlling dog coat colors and patterns, as well as coat type".
http://homepage.usask.ca/~schmutz/dogcolors.html

MendelWeb - A great resource for teachers and students interested in "the origins of classical genetics, introductory data analysis, elementary plant science, and the history and literature of science." Also includes PDF versions of Mendel's original paper (in German), as well as an updated, revised, English translation (which is also included on the teacher CD), and lots of other good genetics links!
http://www.mendelweb.org/

Mendelian and Non- A good description of terminology used regarding Mendelian and non-Mendelian patterns of inheritance by Ken Parejko at the University of Wisconsin Stout.
http://www.uwstout.edu/faculty/parejkok/Intro/mendelian_and_non.htm

Mendelian Inheritance Patterns in Humans – A past project conducted by several participating schools. Though the links on this page no longer work, it may serve as a good source of inspiration for exploring the genetics of several traits that students can easily observe in themselves and their families.
http://scout.wisc.edu/Projects/PastProjects/NH/96-03/96-03-06/0033.html

List of Mendelian traits in humans – Wikipedia article listing several traits in humans that have Mendelian patterns of inheritance, as well as several that were previously thought to be Mendelian, but probably have a more complex genetic basis.
http://en.wikipedia.org/wiki/List_of_Mendelian_traits_in_humans

Online Mendelian Inheritance in Man - A comprehensive database of human genes and phenotypes from the National Center for Biotechnology Information (NCBI). Pretty heavy-duty, so probably most appropriate for advanced high school classes.
http://www.ncbi.nlm.nih.gov/omim

Myths of Human Genetics - Website exploring commonly held misconceptions about human traits previously thought be inherited in a simple Mendelian fashion.
http://udel.edu/~mcdonald/mythintro.html