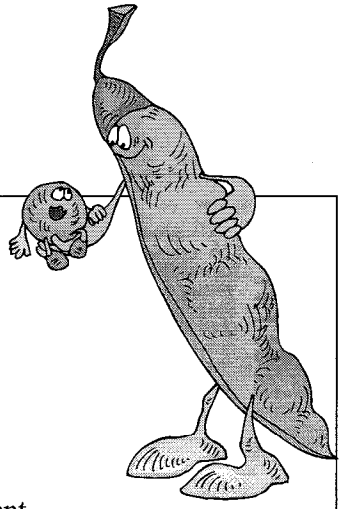
Basic Genetics

Section 10.1
Introduction to Mendelian Genetics



Pre-View 10.1

- **Gregor Mendel** – an Austrian monk whose study of garden peas earned him the title Father of Genetics
- **Genetics** – the study of heredity
- **Heredity** – the passing of traits from one generation to the next
- **Gene** – a section of DNA that determines a specific trait, such as eye color
- **Alleles** – different forms of the same gene; for example, blue and brown are different alleles for eye color
- **Dominant** – a trait that is expressed over another trait
- **Recessive** – a trait that can be hidden by another trait
- **Genotype** – the combination of alleles for a particular trait (homozygous or heterozygous)
- **Homozygous** (or **pure**) – having two of the same alleles for a trait
- **Heterozygous** (or **hybrid**) – having two different alleles for a trait
- **Phenotype** – the physical characteristics of an organism that show how genes are expressed

In the 1800s, an Austrian monk named **Gregor Mendel** studied garden peas. He studied a LOT of garden peas — thousands of them. (Just think about it. He was living in a monastery with no TV, no radio, no telephone, no computer, no internet, no video games, and no friends to hang out with at the mall, so garden peas were pretty interesting!) He started writing his observations, and he noticed that, over time, certain patterns appeared in the plants. For many traits, the peas would have two contrasting forms. Flowers would be purple or white, plant height would be tall or short, the seeds would be wrinkled or smooth, etc. He also noticed that some of the plants were true-breeding for certain traits — that is, they always produced offspring that had traits identical to the parent plants. Then he began experimenting with the plants. Through his experiments, he was able to discover some of the basic concepts of genetics and heredity. Because of his work, Mendel is known as the Father of Genetics.

Genetics is the study of heredity, and **heredity** is the passing on of traits from one generation to the next. To study genetics, Mendel started with true-breeding parent plants. We'll label them *P* for parental. The parental plants had contrasting forms of a trait. For example, one parental plant would have white flowers, and the other parental plant would have purple flowers. When he crossed these plants, the offspring (we'll call them F1) were identical to each other and to one of the parent plants. In the case of one parent having white flowers and the other parent having purple flowers, the offspring all had purple flowers. From this, Mendel reached several conclusions, and these conclusions later became known as Mendel's Laws. It's pretty amazing that Mendel was able to come up with these laws a long time before people knew anything about DNA, genes, and chromosomes!

Genetics uses many specific terms, such as the vocabulary given in the Pre-View above. As you see each bolded word in the following text, be sure you understand what each term means. Once you understand the vocabulary, genetics is not difficult, and you may find it quite interesting.

Genes and Alleles

Do you remember reviewing the process of meiosis in Section 9.2? The process of meiosis forms gametes (sperm and egg). Remember that gametes have half the number of chromosomes as the rest of the body's cells. When an egg is fertilized by a sperm, the resulting offspring gets half of its genetic information from its mother and half from its father.

Each chromosome is made up of one long strand of DNA, and sections of the DNA contain the **genes** that determine specific traits. You get one set of genes from your mother and one set of genes from your father. Let's say your mother has blue eyes, but your father has brown eyes. Your mother may give you a gene for blue eyes, and your father may give you a gene for brown eyes. You get two different forms of the same gene. These different forms are called **alleles**.

Mendel's Law of Dominance

Mendel concluded that biological inheritance is determined by what he called "factors" from the parents. We now refer to Mendel's factors as *genes*. Mendel noticed that one trait appeared more often than the other trait, and the gene for that trait was **dominant** over the other gene. The gene that was not dominant was called **recessive**. The dominant gene hid the recessive gene. Mendel's observation became known as the **Law of Dominance.** In the case of eye color, the gene for brown eyes is dominant over the gene for blue eyes. For an individual who has one allele for blue eyes and one allele for brown eyes, he will have brown eyes.

Mendel found that when he crossed pure purple-flowered plants with pure white-flowered plants, all of the offspring would have purple flowers. When he crossed pure tall plants with pure short plants, all of the offspring were tall. Pure yellow-seeded plants and pure green-seeded plants produced yellow-seeded offspring. The traits for purple flowers, tall plants, and yellow seeds in pea plants are all dominant. If an organism has one dominant allele, the dominant trait will always show. A recessive trait shows only when no dominant allele is present.

We use capital letters for dominant alleles and lowercase letters for recessive alleles. Since they are forms of the same gene, the same letter is used. For example, *W* represents the allele for purple flowers, and *w* represents the allele for white flowers. Different letters are normally used for different traits.

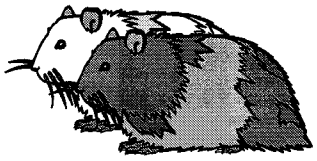
Genotype and Phenotype

The **genotype** of an organism is the combination of alleles it has for a particular trait. A pair of letters (examples: **Bb, dd,** or **EE**) is used for each trait in a genotype because one gene — one letter — comes from momma organism, and one comes from daddy organism, so each baby organism has two genes (two letters) for each trait. For most traits there are three possible genotypes: BB, Bb, or bb.

- If there are <u>two capital</u> letters or <u>two lowercase</u> letters in the genotype (like BB or bb), then the trait is called **homozygous** or **pure**. The alleles for the trait are the same.
- If there is one capital letter and one lowercase letter (like Bb), then the genotype is called **heterozygous** or sometimes **hybrid**. The alleles for the trait are different.

The **phenotype** is the physical characteristics of an organism — what it looks like. The phenotype is the expression of the genes for a particular trait. For example, different phenotypes may be black hair or blonde hair, freckles or no freckles, white flowers or purple flowers, right handed or left handed, etc.

Example: In guinea pigs, brown fur is dominant, and white fur is recessive. The allele for brown fur is shown by B, and the allele for white fur is shown as b. What are the possible genotypes of guinea pigs regarding fur color? What are the hair color phenotypes for guinea pigs?



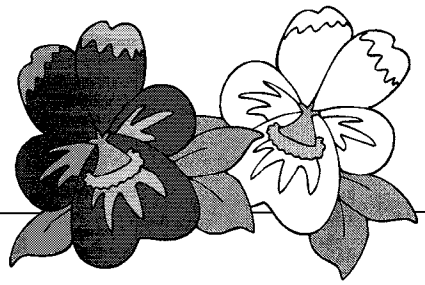
GENOTYPE	DEFINITION	PHENOTYPE
BB	homozygous dominant (or pure dominant)	brown fur
Bb	heterozygous (or hybrid)	brown fur
bb	homozygous recessive (or pure recessive)	white fur

From the chart, you can see that there are three different combinations of genotypes. A guinea pig can be homozygous dominant, BB; homozygous recessive, bb; or heterozygous, Bb.

You can also see that there are only two phenotypes, either brown fur or white fur. Two of the genotypes, BB and Bb, give the same phenotype, brown fur.

Basic Genetics

Section 10.2
Monohybrid Crosses



Pre-View 10.2

- **Law of Segregation** – a natural law explaining that alternative forms of a gene separate during the formation of gametes (sex cells); they do not mix to form a new trait
- **Monohybrid cross** – crossing one trait from two parent organisms
- **Punnett square** – a diagram that shows all possible gene combinations for a cross

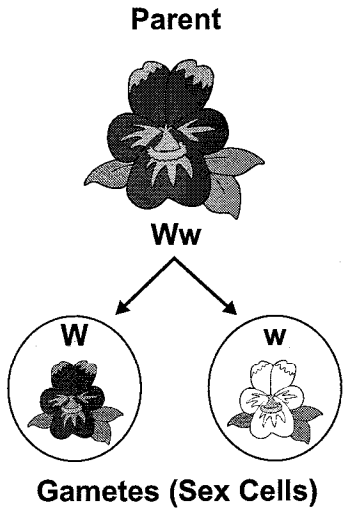
Law of Segregation

By now, you should understand that organisms get half their genetic information from their mother and the other half from their father. If an individual receives the dominant gene for a trait from its mother and a recessive gene for that same trait from its father, it will display the dominant trait.

Let's go back to Mendel's work. He observed that inherited traits such as flower color come from different forms of the same gene. These different gene forms are called alleles. Mendel recognized that each organism inherits two genes for each trait, one from each parent. These genes can be the same alleles, or they can be different. Remember, the Law of Dominance says that a dominant gene will express itself over a recessive gene, and a recessive trait will only be expressed if the individual has two recessive alleles.

Mendel also recognized that each allele for a trait segregates (or separates) during gamete production so that each gamete carries only one of the genes. These two alleles do not change or mix with one another. Mendel's observation of gene segregation is known as the **Law of Segregation**. In other words, when an individual forms sex cells, sperm or egg, the different forms of a gene will separate. It might pass on either a dominant gene or a recessive gene, but it will not pass on a combination of both for that one trait.

Example 1: In pea plants, a heterozygous plant for flower color has the alleles Ww. It has a dominant allele for purple flowers and a recessive allele for white flowers. The phenotype for this plant is purple flowers. According to the Law of Segregation, what genes would be present in the gametes for this plant?

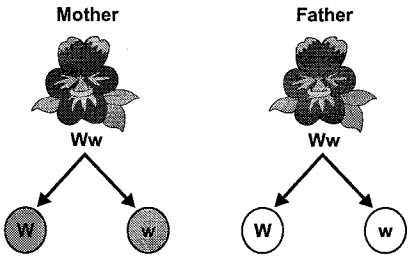


During gamete formation, the alleles separate. This heterozygous plant will form gametes that have either the gene for purple flowers or the gene for white flowers. The same gamete, however, will NOT contain both genes. When these genes separate, they are not affected by the other gene. In other words, the purple gene doesn't mix with the white gene to form a gene for light purple flowers or for flowers that are white with purple stripes.

Monohybrid Crosses

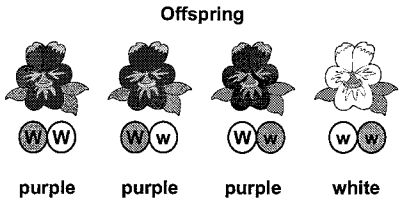
When the inheritance patterns of only one trait, such as flower color, are studied, the cross between two parent organisms is called a **monohybrid cross.** In a monohybrid cross, one allele from a female egg is joined with one allele from a male sperm.

Example 2: What if a heterozygous pea plant for flower color was crossed with another heterozygous plant? What would be the possible genotypes of the offspring? What would be the possible phenotypes for the offspring?



The gametes for the parent plants could be combined four different ways: WW, Ww, Ww, and ww. Notice that in this case, two of those combinations are the same, Ww.

For a monohybrid cross of heterozygous traits, the possible genotypes are homozygous dominant (WW), heterozygous (Ww), or homozygous recessive (ww). Statistically, one out of four offspring will be homozygous dominant (WW), one out of four will be homozygous recessive (ww), and two out of four (one half) will be heterozygous (Ww).



The possible phenotypes for this example are purple flowers or white flowers. Statistically, three out of four offspring will have purple flowers.

Probability and Predicting Inheritance

Because alleles for a trait separate independently of each other during meiosis, we can use the laws of probability to predict genetic outcomes. Probability is the chance that a certain event will occur. Here are three things you should know about probability:

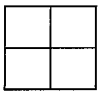
1. Probability predicts what is most likely to occur, but what actually occurs may be different. For example, let's say humans have an equal chance of having a girl or a boy. A couple may have 4 daughters and no sons.
2. Each outcome is not influenced by the outcomes before it. For example, let's assume humans have a 50/50 chance of having a female child. A couple has two daughters. The chance that a third child will also be a daughter is still 50%. The previous offspring do not influence the probability of future offspring.
3. The actual results will be closer to the predicted results for a large number of outcomes. If in fact humans have a 50/50 chance of having a girl or a boy, the greater the number of children a couple has, the more likely it is that they will have an equal number of sons and daughters.

Using a Punnett Square

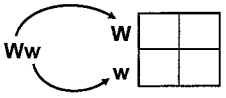
How do scientists make genetic predictions? One way is by using a **Punnett square.** A Punnett square is a diagram that shows all possible gene combinations from a genetic cross. Dominant alleles are represented by capital letters, and lowercase letters stand for recessive alleles. A simple Punnett square looks something like a window pane. Letters that represent the parents' alleles are listed at the top and left sides of the grid. The squares inside the grid are used to show all possible allele combinations of the offspring.

Let's use Mendel's pea plants to see how a Punnett square works. One of Mendel's crosses was between a purple-flowered pea plant that was heterozygous (Ww) with a white-flowered pea plant that was homozygous recessive (ww). The cross between these two parents is **Ww × ww.** Follow the steps below to fill in a Punnett square for this cross and to determine the probability of possible outcomes for the offspring.

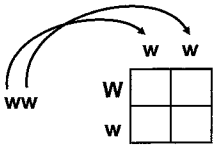
Step 1: Draw a Punnett square with four compartments.



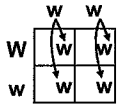
Step 2: Take the genotype for one parent and put one letter outside the Punnett square to the left of each row.



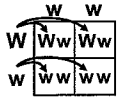
Step 3: Take the genotype for the other parent and put one letter outside the Punnett square above each column.



Step 4: Now you can fill in the grid. Take the letter over each column and copy it into the two squares below it.



Step 5: Take the letter at the beginning of each row and copy it into the two squares to the right.



In this cross, notice that 50% (one half) of the offspring are predicted to be heterozygous for flower color, and the other 50% are predicted to be homozygous recessive. Likewise, we could predict that 50% of the offspring would have purple flowers and that the other 50% would have white flowers.

Math Review

Probability can be given as a ratio, a fraction, or a percentage. In genetics, ratios are commonly given as the number of offspring that have a particular gene or trait versus the number of offspring that have the other form of the gene or trait. For example, if a Punnett square predicts that three out of four offspring will have purple flowers, the ratio is commonly written 3:1 (read three to one), and this notation shows that statistically, three offspring will have purple flowers and one offspring will have white flowers. Fractions show the predicted number of offspring having a particular gene or trait over the total number of possible outcomes. For a monohybrid cross, there are four possible outcomes. Percentages can be calculated from the fractions. For a monohybrid cross, let's review what each of these math elements looks like. The chart below gives the equivalence between ratios, fractions, and percentages.

Description	Ratio	Fraction	Percent
one to three or *one out of four*	1:3	1/4	25%
two to two or *two out of four*	2:2	1/2	50%
three to one or *three out of four*	3:1	3/4	75%
four out of four			100%

Suppose that you wanted to know if a tall pea plant was homozygous dominant (TT) or heterozygous (Tt). How could you find out? The easiest way to find out would be to cross it with a short pea plant (tt). If you complete a Punnett square for a cross between a short plant and a heterozygous tall plant, it would predict that half of the offspring would be short (figure 10-1). If you complete a Punnett square for a cross between a short plant and a homozygous dominant tall plant, it would predict that all of the offspring would be tall (figure 10-2).

Tt × tt

	T	t
t	Tt	tt
t	Tt	tt

50% tall offspring
50% short offspring

Fig. 10-1

TT × tt

	T	T
t	Tt	Tt
t	Tt	Tt

100% tall offspring

Fig. 10-2

Example 3: A heterozygous tall pea plant is crossed with a homozygous short pea plant. A Punnett square predicts that the offspring of this cross will be 50% tall and 50% short. In a classroom experiment, students tested this prediction with four seeds obtained from this cross. The first three offspring from this cross produced all tall plants. How is this possible? What is the probability that the fourth offspring will be tall?

To answer these questions, remember the review of probability.

1. Even though a Punnett square predicts that half the offspring will be tall and half will be short, this may not actually occur.

2. The probability that each seed will produce a tall plant is 50% (or 1/2). Therefore, the probability that the fourth seed will be tall is also 1/2. The probability of the fourth seed is not affected by the three previous seeds.

3. The more seeds produced and tested using this cross, the more likely it will be to have equal numbers of tall and short plants.

Example 4: Two heterozygous tall pea plants are crossed. They produce 16 seeds. How many of those seeds will likely result in short plants?

This question can be answered by following several steps.

Step 1: Fill in a Punnett square.

Step 2: Use the Punnett square to determine the predicted fraction or percentage of short plants. In this case, 1 out of 4 (or 25%) of the plants is expected to be short.

Step 3: Multiply the fraction or percent of short plants to the total number of seeds. One-fourth (or 25%) of 16 is 4, so 4 out of the 16 seeds would likely result in short plants.

Tt × Tt

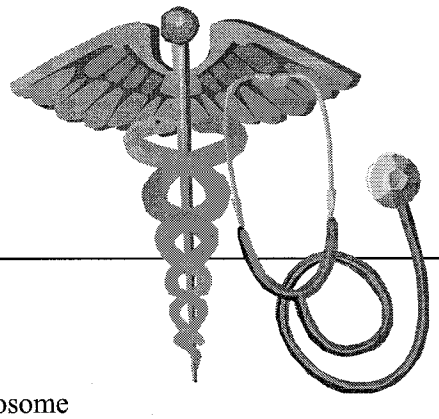
	T	t
T	TT	Tt
t	Tt	tt

1/4 (25%) short

1/4 × 16 = 4

Basic Genetics

Section 10.3
Human Autosomal Genetic Diseases



Pre-View 10.3

- **Autosomes** – all chromosomes except the sex chromosomes (X and Y)
- **Autosomal disease** – genetic disease that is carried by a gene on an autosome
- **Carrier** – a person who has one recessive gene for a recessive genetic disease; will not display symptoms of the disease but can pass on the gene to offspring
- **Cystic fibrosis** – a genetic disease that causes the body to produce unusually thick, sticky mucus; decreases life-expectancy
- **Phenylketonuria (PKU)** – a genetic disease where a person is born without the enzyme to break down and use phenylalanine, an essential amino acid
- **Tay-Sachs** – a fatal genetic disease that causes fatty material to build up in the nerves and brain

OK, so maybe you're thinking, "Who cares about stupid pea plants anyway? This doesn't have anything to do with what's going on now!" Ah, but it does. These same principles apply to humans, and a Punnett square can be used to determine the probability that a child will be born with a genetic disease.

Now that you know something about genes and how they are inherited, let's take a look at some genetic diseases. Genetic diseases are caused by abnormalities in the genes or chromosomes. They are present in all body cells, and they are present for the person's entire life. There are three main types of genetic diseases:

1. autosomal gene diseases
2. sex-linked gene diseases
3. missing or extra chromosome diseases

Most of the over 4,000 known genetic diseases are rare and may affect only one person out of millions. Others are more common and may be carried by 1 out of every 20 people. In this sub-section, let's look at autosomal gene diseases. We'll look at other types later.

Autosomal Disorders

Autosomes are all of the chromosomes except the X and Y (sex) chromosomes. **Autosomal diseases** are genetic diseases carried on autosomes. Most autosomal diseases are recessive. When a disease is recessive, a person must inherit a copy of the gene from both parents to "inherit" the disease and display symptoms. If a person has just one defective gene, he or she is called a **carrier**. Carriers will not have symptoms of the disease, but they can pass on the defective gene to their children.

Cystic Fibrosis

One autosomal recessive disorder is **cystic fibrosis**. In the United States, about 5% of the human population carries the gene for cystic fibrosis. The defective gene is found on chromosome 7, and it causes production of a protein that makes a person's body produce unusually sticky, thick mucus. This mucus can lead to severe lung infections, and it prevents enzymes from the pancreas from helping the body to break down food and absorb nutrients. If a person has cystic fibrosis, he or she is likely to have a persistent cough, frequent lung infections, poor growth and weight loss even when he or she eats well, and shortness of breath. Right now, there is no known cure for cystic fibrosis although medical treatments can help patients live longer and better lives than they did in the past.

Phenylketonuria

Another autosomal recessive disorder is PKU, which is short for **phenylketonuria**. A person who has PKU is missing an enzyme called phenylalanine hydroxylase. This enzyme breaks down and allows the body to use phenylalanine, one of the eight essential amino acids found in protein. Without the enzyme, phenylalanine builds up in the person's body. The build-up affects the central nervous system and causes brain damage.

PKU is a treatable disease that is easily diagnosed by a blood test. In fact, it is so simple to diagnose that all 50 states routinely test every newborn baby for PKU soon after they are born. Treatment is simple. If the blood test on an infant is positive for PKU, then the infant is placed on a special diet that avoids all proteins containing phenylalanine. By the time the child is five or six, a regular diet can usually be resumed. If the special diet is not followed during early childhood, then usually the infant will develop severe mental retardation by age one, with tremors, seizures, and hyperactivity.

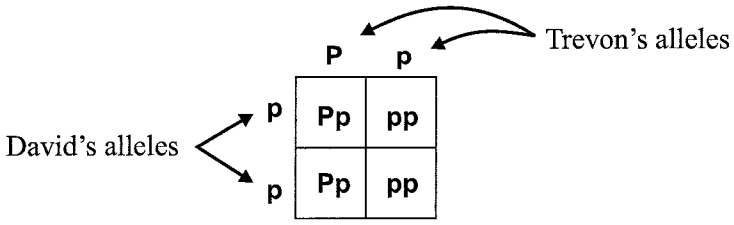
Tay-Sachs

Tay-Sachs disease is another recessive genetic disorder. It affects people of Eastern European Jewish descent more often than any other ethnic group although others can be affected. Babies that have Tay-Sachs seem normal for the first few months of their lives. After a few months, a fatty material starts to build up in their nerve cells and brain, and they become deaf, blind, and unable to swallow. Eventually they become paralyzed and may need a feeding tube. These children seldom live past the age of four. At this time there is no cure for Tay-Sachs.

Now let's look at a real world example of how a Punnet square can predict inheritance of a disease.

Example 1: A young couple has decided that they want to start their family. David is 28 years old and has been promised a promotion in his company that will gave him a better position and more benefits, as well as a larger salary. He has phenylketonuria, or PKU. His mother and one sister also have PKU. His wife Trevon is 25 years old and has an aunt with PKU. She also had two grandparents with phenylketonuria although neither of her parents had it. David and Trevon want to know what their chances are of having a child with PKU. What are the possible genotypes and phenotypes of their future children? What is the probability that they will have children with PKU?

Since David has PKU, we know that his genotype is pp. Trevon could be either PP or Pp since she doesn't have PKU, but there is a history of it in her family. We're going to assume that she is heterozygous — Pp.



If Trevon is heterozygous for PKU (which means she is a carrier), the Punnett square above shows the possible genotypes for their children. The possible genotypes are heterozygous, Pp, or recessive, pp.

The possible phenotypes for their children would be either that they have PKU or that they do not have PKU. A child who inherits a genotype of Pp will not have the disease but will be a carrier of the disease. A child who inherits a genotype of pp will have the disease.

Statistically, one half (or 50%) of their children will be carriers, and one half will have PKU.

Example 2: A married couple has their first child, and the child has cystic fibrosis. What do you know about the genotypes of each parent? What are the chances that they will have another child with the disease?

Since cystic fibrosis is an autosomal recessive disease, each parent must be a carrier of the disease. In order for the child to get the disease, he had to get one defective gene from the mother and another defective gene from the father. So, both parents have the same genotype. They are heterozygous for the disease and have one "good" gene and one "bad" gene.

To find the chances that a second child will be affected by the disease, fill in a Punnett square.

	C	c
C	CC	Cc
c	Cc	cc

From the Punnett square, you can see that there is a 25% (or 1 in 4) chance that additional children will have the disease. There is a 50% chance that the next child will be a carrier of the disease and a 25% chance that another child will not carry the defective gene at all.