Genetics
Standards
• Use Mendel’s laws of segregation & independent assortment to analyze patterns of inheritance.
• Discuss observed inheritance patterns caused by various modes of inheritance, including dominant, recessive, codominant, sex-linked, polygenic, and multiple alleles.

Mendel and His Pea Plants
• Gregor Mendel was born in 1822 did well in school and became a monk.
• He went to the University of Vienna, where he studied science and math.
• Mendel is best known for his experiments with the pea plant *Pisum sativum*.

Blending Theory of Inheritance
• During Mendel’s time, the blending theory of inheritance was popular.
• This is the theory that offspring have a blend, or mix, of the characteristics of their parents.
• Mendel noticed plants in his own garden that weren’t a blend of the parents.
• For example, a tall plant and a short plant had offspring that were either tall or short but not medium in height.

Why Study Pea Plants?
• Pea plants are a good choice because they are fast growing and easy to raise.
• They also have several visible characteristics that may vary.
• These characteristics, include seed form and color, flower color, pod form and color, placement of pods and flowers on stems, and stem length.
• Each characteristic has two common values.
Mendel’s First Set of Experiments
- Mendel first experimented with just one characteristic of a pea plant at a time.
- He began with flower color.
- Mendel cross-pollinated purple- and white-flowered parent plants.
- The parent plants in the experiments are referred to as the P (for parent) generation.

F1 and F2 Generations
- The offspring of the P generation are called the F1 (for filial, or “offspring”) generation.
- All of the plants in the F1 generation had purple flowers. None of them had white flowers.
- Mendel wondered what had happened to the white-flower characteristic.
- He assumed some type of inherited factor produces white flowers and some other inherited factor produces purple flowers.
- Did the white-flower factor just disappear in the F1 generation? If so, then the offspring of the F1 generation—called the F2 generation—should all have purple flowers like their parents.
- To test this prediction, Mendel allowed the F1 generation plants to self-pollinate.
- He was surprised by the results. Some of the F2 generation plants had white flowers.
- He studied hundreds of F2 generation plants, and for every three purple-flowered plants, there was an average of one white-flowered plant.

Law of Segregation
- Mendel did the same experiment for all seven characteristics.
- In each case, one value of the characteristic disappeared in the F1 plants and then showed up again in the F2 plants.
- And in each case, 75 percent of F2 plants had one value of the characteristic and 25 percent had the other value.
- Based on these observations, Mendel formulated his first law of inheritance. This law is called the law of segregation.
- **The law of segregation** states that there are two factors controlling a given characteristic, one of which dominates the other, and these factors separate and go to different gametes when a parent reproduces.

**Mendel’s Second Set of Experiments**
- Mendel wondered whether different characteristics are inherited together.
- Or do these two characteristics show up in different combinations in offspring?
- To answer these questions, Mendel next investigated two characteristics at a time.

**F1 and F2 Generations**
- Mendel observed that plants in the F1 generation were all alike.
- When the F1 generation plants self-pollinated, however, their offspring—the F2 generation—showed all possible combinations of the two characteristics.
- These combinations of characteristics were not present in the F1 or P generations.

![Diagram showing F1 and F2 generations with genetic combinations]

**Law of Independent Assortment**
- Mendel repeated this experiment with other combinations of characteristics.
- Each time, the results were the same.
- The results of Mendel’s second set of experiments led to his second law.
- **The law of independent assortment** states that factors controlling different characteristics are inherited independently of each other.

![Diagram illustrating independent assortment]

**Genetics of Inheritance**
- Today, we known that characteristics of organisms are controlled by genes on chromosomes.
- The position of a gene on a chromosome is called its locus.
- In sexually reproducing organisms, each individual has two copies of the same gene, as there are two versions of the same chromosome (homologous chromosomes).
- One copy comes from each parent.
- The gene for a characteristic may have different versions, but the different versions are always at the same locus.
- The different versions are called alleles.
- Different alleles account for much of the variation in the characteristics of organisms.

**Genotype and Phenotype**

- When gametes unite during fertilization, the resulting zygote inherits two alleles for each gene. One allele comes from each parent.
- The alleles an individual inherits make up the individual's genotype.
- The two alleles may be the same or different.
- The expression of an organism's genotype produces its phenotype.
- The phenotype refers to the organism's characteristics. Different genotypes may produce the same phenotype.
- In a Bb heterozygote, only the B allele is expressed, so the b allele doesn't influence the phenotype.
- In general, when only one of two alleles is expressed in the phenotype, the expressed allele is called the dominant allele.
- The allele that isn't expressed is called the recessive allele.

<table>
<thead>
<tr>
<th>Alleles</th>
<th>Genotypes</th>
<th>Phenotypes</th>
</tr>
</thead>
<tbody>
<tr>
<td>B (purple)</td>
<td>Bb (heterozygote)</td>
<td>purple flowers</td>
</tr>
<tr>
<td>b (white)</td>
<td>bb (homozygote)</td>
<td>white flowers</td>
</tr>
</tbody>
</table>

**Probability**
• Probability is the likelihood, or chance, that a certain event will occur. The easiest way to understand probability is with coin tosses.
• When you toss a coin, the chance of a head turning up is 50 percent.
• This is because a coin has only two sides, so there is an equal chance of a head or tail turning up on any given toss.
• If you toss a coin twice, you might expect to get one head and one tail.
• But each time you toss the coin, the chance of a head is still 50 percent.
• Therefore, it’s quite likely that you will get two or even several heads (or tails) in a row.
• What if you tossed a coin ten times? You would probably get more or less than the expected five heads.

**Probability and Inheritance**

How is gamete formation like tossing a coin?

• Assume that an organism is heterozygous for a trait, so it has the genotype Bb.
• During meiosis the alleles segregate and go to different gametes.
• Based on the rules of probability, any given gamete of this parent has a 50 percent chance of having the B allele and a 50 percent chance of having the b allele.

**Punnett Squares**

• A Punnett square is a chart that allows you to easily determine the expected percentage of different genotypes and phenotypes in the offspring of two parents.
• The different possible combinations of alleles in their offspring are determined by filling in the cells of the Punnett square with the correct letters (alleles).
Punnett Square for Two Characteristics

- When you consider more than one characteristic at a time, using a Punnett square is more complicated.
- This is because many more combinations of alleles are possible.
- For example, with two genes each having two alleles, an individual has four alleles, and these four alleles can occur in 16 different combinations.

Non-Mendelian Inheritance

- The inheritance of characteristics is not always as simple as it is for the characteristics that Mendel studied in pea plants.
- For each characteristic Mendel investigated one allele was completely dominant to the other. However, the two alleles may have a different relationship than the simple dominant-recessive relationship.

Codominance

- Codominance occurs when both alleles are expressed equally in the phenotype of the heterozygote.
- A red and white flower has codominant alleles for red petals and white petals.

Incomplete Dominance

- Incomplete dominance occurs when the phenotype of the offspring is somewhere in between the phenotypes of both parents; a completely dominant allele does not occur.
Multiple Alleles

Many genes have multiple (more than two) alleles.

- An example is ABO blood type in humans.
  - There are three common alleles for the gene that controls this characteristic.
  - The alleles $I^A$ and $I^B$ are dominant over $i$.
  - A person who is homozygous recessive $ii$ has type O blood.
  - Homozygous dominant $I^A I^A$ or heterozygous dominant $I^A i$ have type A blood, and homozygous dominant $I^B I^B$ or heterozygous dominant $I^B i$ have type B blood.
  - $I^A I^B$ people have type AB blood, because the A and B alleles are codominant.
  - Type A and type B parents can have a type AB child.
  - Type A and type B parents can also have a child with Type O blood, if they are both heterozygous ($I^B i$, $I^A i$).

Polygenic Characteristics

Polygenic characteristics are controlled by more than one gene, and each gene may have two or more alleles.

- The genes may be on the same chromosome or on nonhomologous chromosomes.
- If the genes are located close together on the same chromosome, they are likely to be inherited together.
- However, it is possible that they will be separated by crossing-over during meiosis, in which case they may be inherited independently of one another.
- If the genes are on nonhomologous chromosomes, they may be recombined in various ways because of independent assortment.

Effects of Environment on Phenotype

Genes play an important role in determining an organism’s characteristics. However, for many characteristics, the individual’s phenotype is influenced by other factors as well.
Environmental factors, such as sunlight and food availability, can affect how genes are expressed in the phenotype of individuals. Here are just two examples:

- Genes play an important part in determining our adult height. However, factors such as poor nutrition can prevent us from achieving our full genetic potential.
- Genes are a major determinant of human skin color. However, exposure to ultraviolet radiation can increase the amount of pigment in the skin and make it appear darker.

**Chromosomes and Genes**

Each species has a characteristic number of chromosomes.

- Chromosomes are coiled structures made of DNA and proteins called histones.
- Chromosomes are the form of the genetic material of a cell during cell division.

**The human genome**

- The human genome has 23 pairs of chromosomes located in the nucleus of somatic cells.
- Each chromosome is composed of genes and other DNA wound around histones (proteins) into a tightly coiled molecule.

**Autosomes**

Of the 23 pairs of human chromosomes, 22 pairs are autosomes.

- Autosomes are chromosomes that contain genes for characteristics that are unrelated to sex. These chromosomes are the same in males and females.
- The great majority of human genes are located on autosomes.

**Sex Chromosomes**

- The remaining pair of human chromosomes consists of the sex chromosomes, X and Y.
• Females have two X chromosomes, and males have one X and one Y chromosome.
• In females, one of the X chromosomes in each cell is inactivated and known as a Barr body. This ensures that females, like males, have only one functioning copy of the X chromosome in each cell.

Human Genes
• Humans have an estimated 20,000 to 22,000 genes.
• Far simpler species have almost as many genes as humans.
• However, human cells use splicing and other processes to make multiple proteins from the instructions encoded in a single gene.
• Of the 3 billion base pairs in the human genome, only about 25 percent make up genes and their regulatory elements.
• The functions of many of the other base pairs are still unclear.

Sex-Linked Genes
Genes located on the sex chromosomes are called sex-linked genes.
• Most sex-linked genes are on the X chromosome, because the Y chromosome has relatively few genes.
• Strictly speaking, genes on the X chromosome are X-linked genes, but the term sex-linked is often used to refer to them.

Mendelian Inheritance in Humans
• Characteristics that are encoded in DNA are called genetic traits.
• Different types of human traits are inherited in different ways.
• Some human traits have simple inheritance patterns like the traits that Gregor Mendel studied in pea plants.
• Other human traits have more complex inheritance patterns.

Autosomal Traits
• Autosomal traits are controlled by genes on one of the 22 human autosomes.
• Consider earlobe attachment.
  o A single autosomal gene with two alleles determines whether you have attached earlobes or free-hanging earlobes.
  o The allele for free-hanging earlobes (F) is dominant to the allele for attached earlobes (f).

**Sex-Linked Traits**
• Traits controlled by genes on the sex chromosomes are called sex-linked traits, or X-linked traits in the case of the X chromosome.
• Single-gene X-linked traits have a different pattern of inheritance than single-gene autosomal traits.
• Because males have just one X chromosome, they have only one allele for any X-linked trait.
• Therefore, a recessive X-linked allele is always expressed in males.
• Because females have two X chromosomes, they have two alleles for any X-linked trait.
• Therefore, they must inherit two copies of the recessive allele to express the recessive trait.
• This explains why X-linked recessive traits are less common in females than males.

**Genetic Terminology**
• **Trait** - any characteristic that can be passed from parent to offspring
- **Heredity** - passing of traits from parent to offspring
- **Genetics** - study of heredity
- **Monohybrid cross** - cross involving a single trait e.g. flower color
- **Dihybrid cross** - cross involving two traits e.g. flower color & plant height
- **Alleles** - two forms of a gene (dominant & recessive)
- **Dominant** - stronger of two genes expressed in the hybrid; represented by a capital letter (R)
- **Recessive** - gene that shows up less often in a cross; represented by a lowercase letter (r)
- **Genotype** - gene combination for a trait (e.g. RR, Rr, rr)
- **Phenotype** - the physical feature resulting from a genotype (e.g. red, white)
- **Homozygous genotype** - gene combination involving 2 dominant or 2 recessive genes (e.g. RR or rr); also called pure
- **Heterozygous genotype** - gene combination of one dominant & one recessive allele (e.g. Rr); also called hybrid