HERITABLE VARIATION AND PATTERNS OF INHERITANCE

- **Heredity** is the transmission of traits from one generation to the next.
- **Genetics** is the scientific study of heredity.
- Gregor Mendel
  - worked in the 1860s,
  - was the first person to analyze patterns of inheritance, and
  - deduced the fundamental principles of genetics.

In an Abbey Garden

- Mendel studied garden peas because they
  - were easy to grow,
  - came in many readily distinguishable varieties,
  - are easily manipulated, and
  - can self-fertilize.

Monohybrid Crosses

- A **monohybrid cross** is a cross between purebred parent plants that differ in only one character.

- Mendel developed four hypotheses from the monohybrid cross, listed here using modern terminology (including "gene" instead of "heritable factor").
  1. The alternative versions of genes are called **alleles**.
  2. For each inherited character, an organism inherits two alleles, one from each parent.
     - An organism is **homozygous** for that gene if both alleles are identical.
     - An organism is **heterozygous** for that gene if the alleles are different.
  3. If two alleles of an inherited pair differ,
     - then one determines the organism's appearance and is called the **dominant allele** and
     - the other has no noticeable effect on the organism's appearance and is called the **recessive allele**.
  4. Gametes carry only one allele for each inherited character.
     - The two alleles for a character segregate (separate) from each other during the production of gametes.
     - This statement is called the **law of segregation**.
- Do Mendel's hypotheses account for the 3:1 ratio he observed in the F_2 generation?
- A **Punnett square** highlights
  - the four possible combinations of gametes and
  - the four possible offspring in the F_2 generation.
- Geneticists distinguish between an organism's physical appearance and its genetic makeup.
  - An organism's physical appearance is its **phenotype**.
  - An organism's genetic makeup is its **genotype**.

Genetic Alleles and Homologous Chromosomes

- A gene **locus** is a specific location of a gene along a chromosome.
- Homologous chromosomes have alleles (alternate versions) of a gene at the same locus.

Mendel's Law of Independent Assortment

- A **dihybrid cross** is the mating of parental varieties differing in two characters.
- What would result from a dihybrid cross? Two hypotheses are possible:
  1. dependent assortment or
  2. independent assortment.

- Mendel's dihybrid cross supported the hypothesis that each pair of alleles segregates independently of the other pairs during gamete formation.
- Thus, the inheritance of one character has no effect on the inheritance of another.
- This is called Mendel's **law of independent assortment**.
- Independent assortment is also seen in two hereditary characters in Labrador retrievers.

Using a Testcross to Determine an Unknown Genotype

- A **testcross** is a mating between
  - an individual of dominant phenotype (but unknown genotype) and
  - a homozygous recessive individual.

The Rules of Probability

- Mendel's strong background in mathematics helped him understand patterns of inheritance.
- The **rule of multiplication** states that the probability of a compound event is the product of the separate probabilities of the independent events.

Family Pedigrees

- Mendel's principles apply to the inheritance of many human traits.
- Dominant traits are not necessarily
  - normal or
  - more common.
- **Wild-type traits** are
  - those seen most often in nature and
  - not necessarily specified by dominant alleles.
- A family **pedigree**
  - shows the history of a trait in a family and
  - allows geneticists to analyze human traits.

Human Disorders Controlled by a Single Gene

- Many human traits
  - show simple inheritance patterns and
  - are controlled by single genes on autosomes.

Recessive Disorders

- Most human genetic disorders are recessive.
- Individuals who have the recessive allele but appear normal are **carriers** of the disorder.
- **Cystic fibrosis** is
  - the most common lethal genetic disease in the United States and
  - caused by a recessive allele carried by about one in 31 Americans.

- Prolonged geographic isolation of certain populations can lead to **inbreeding**, the mating of close relatives.
- Inbreeding increases the chance of offspring that are homozygous for a harmful recessive trait.

Dominant Disorders

- Some human genetic disorders are dominant.
  - **Achondroplasia** is a form of dwarfism.
    - The homozygous dominant genotype causes death of the embryo.
    - Thus, only heterozygotes have this disorder.
  - **Huntington's disease**, which leads to degeneration of the nervous system, does not usually begin until middle age.

Genetic Testing

- Today many tests can detect the presence of disease-causing alleles.
- Most genetic tests are performed during pregnancy.
  - Amniocentesis collects cells from amniotic fluid.
  - Chorionic villus sampling removes cells from placental tissue.
- Genetic counseling helps patients understand the results and implications of genetic testing.

VARIATIONS ON MENDEL'S LAWS

- Some patterns of genetic inheritance are not explained by Mendel's laws.

Incomplete Dominance in Plants and People

- In **incomplete dominance**, F_1 hybrids have an appearance between the phenotypes of the two parents.
- **Hypercholesterolemia**
  - is a human trait that is an example of incomplete dominance and
  - is characterized by dangerously high levels of cholesterol in the blood.
  - heterozygotes have blood cholesterol levels about twice normal, and
  - homozygotes have about five times the normal amount of blood cholesterol and may have heart attacks as early as age 2.

ABO Blood Groups: An Example of Multiple Alleles and Codominance

- The **ABO blood groups** in humans are an example of multiple alleles.
- The immune system produces blood proteins called antibodies that bind specifically to foreign carbohydrates.
- If a donor's blood cells have a carbohydrate (A or B) that is foreign to the recipient, the blood cells may clump together, potentially killing the recipient.
- The clumping reaction is the basis of a blood-typing lab test.
- The human blood type alleles I^A and I^B are **codominant**, meaning that both alleles are expressed in heterozygous individuals who have type AB blood.

Pleiotropy and Sickle-Cell Disease

- **Pleiotropy** is when one gene influences several characters.
- **Sickle-cell disease**
  - exhibits pleiotropy,
  - results in abnormal hemoglobin proteins, and
  - causes disk-shaped red blood cells to deform into a sickle shape with jagged edges.

Polygenic Inheritance

- **Polygenic inheritance** is the additive effects of two or more genes on a single phenotype.

The Role of Environment

- Many human characters result from a combination of
  - heredity and
  - environment.
- Only genetic influences are inherited.

THE CHROMOSOMAL BASIS OF INHERITANCE

- The **chromosome theory of inheritance** states that
  - genes are located at specific positions (loci) on chromosomes and
  - the behavior of chromosomes during meiosis and fertilization accounts for inheritance patterns.

- It is *chromosomes* that
  - undergo segregation and independent assortment during meiosis and
  - account for Mendel's laws.

Linked Genes

- **Linked genes**
  - are located close together on a chromosome and
  - tend to be inherited together.
- Thomas Hunt Morgan
  - used the fruit fly *Drosophila melanogaster* and
  - determined that some genes were linked based on the inheritance patterns of their traits.

Linkage Maps

- Early studies of crossing over were performed using the fruit fly *Drosophila melanogaster*.
- Alfred H. Sturtevant, a student of Morgan,
  - developed a method for mapping the relative gene locations,
  - which resulted in the creation of **linkage maps**.

SEX CHROMOSOMES AND SEX-LINKED GENES

- Sex chromosomes influence the inheritance of certain traits. For example, humans that have a pair of sex chromosomes designated
  - X and Y are male or
  - X and X are female.

Sex Determination in Humans

- Nearly all mammals have a pair of sex chromosomes designated X and Y.
  - Males have an X and Y.
  - Females have XX.

Sex-Linked Genes

- Any gene located on a sex chromosome is called a **sex-linked gene**.
  - Most sex-linked genes are found on the X chromosome.
  - **Red-green colorblindness** is
    - a common human sex-linked disorder and
    - caused by a malfunction of light-sensitive cells in the eyes.
- **Hemophilia**
  - is a sex-linked recessive blood-clotting trait that may result in excessive bleeding and death after relatively minor cuts and bruises and
  - has plagued the royal families of Europe.