

GENETICS

Basic Concepts

A. Organismal Reproduction

1. One of the most important requisites of all life, from the earliest life forms to present-day organisms, is **reproduction**.
2. Characteristics or **traits** of organisms must be passed on during reproduction.

B. Cellular Reproduction

1. Life as we know it is based on the **cell**, the basic unit of life.
2. Cell theory states all organisms are made up of cells and come from cells.

C. DNA

1. DNA (deoxyribonucleic acid) is the **molecule of inheritance** in ALL cellular forms of life.

D. Chromosomes

1. Eukaryotic cells possess nuclear DNA, with structural and enzymatic proteins, forming **chromatin**, which is visible as **chromosomes** during parts of the cell cycle.
2. Prokaryotic cells possess simpler DNA.
3. Sexually reproducing organisms typically have pairs of **homologous chromosomes** (look-alike chromosomes).

E. RNA

1. RNA (ribonucleic acid) is found in several forms, most of which are used in protein synthesis.
2. RNA is the **molecule of inheritance** in some viruses, which are not cell-based life forms.

F. Genes

1. Functional unit of inheritance and basis for most traits.
2. Located at **loci**, or specific positions, on DNA; to be preserved and transmitted.
3. Control biological processes through production of proteins and RNA.

4. **Genotype** refers to the **genetic composition** of the organism.
5. **Phenotype** refers to the **observable inherited traits** (e.g., physical, behavioral, physiological characteristics) based on the inherited genotype.

Genes Form Basis of Inheritance



G. Ploidy

1. Homologous chromosome pairs have the same **loci**, thus **genes**.
2. When both chromosomes are present, for each gene there are two representatives; this is represented by the symbol **2n** or **diploid** condition.
3. When only half of each homologous chromosome pair is present, such as in gametes, this is represented by the symbol **n** or **haploid**.

H. Alleles

1. Alternate forms of the same gene that could occupy the same **locus** (e.g., brown versus blue eye color).

2. Homologous chromosomes possess two representatives of each gene (i.e., **2n**).
3. **Homozygous** refers to the diploid condition where **both alleles of the genotype are identical** (e.g., AA, aa).
4. **Heterozygous** refers to the diploid condition where **both alleles of the genotype are different** (i.e., Aa).
5. **Dominant** alleles form a phenotypic expression regardless of the other allele on the matched chromosome of the homolog (e.g., "AA" or "Aa" genotypes will both express the phenotype designated by the "A" allele).
6. **Recessive** alleles fail to form a phenotypic expression unless the other allele on the matched chromosome is also recessive (e.g., "aa" genotype is the only way for the phenotype designated by the "a" allele to be expressed, assuming no other gene pairs influence inheritance [see epistasis discussion in Gene Action Categories, page 2]).
7. Additional types of allelic interactions will be discussed in subsequent sections.
8. **Determining gamete types:** Assuming there are no mutations, alleles present in gametes are determined by the diploid genotypes of parents.
 - a. For **homozygous** genotypes, **haploid** gametes will be identical for the given trait (i.e., AA individual would produce "A" gametes only; AAbb individual would produce "Ab" gametes only).
 - b. For **heterozygous** genotypes, **haploid** gametes will be **different** for the given trait (i.e., Aa individual would produce "A" & "a" gametes; AaBb individual would produce "AB, Ab, aB, ab" gametes—assuming two traits are unlinked [see Independent Assortment & Dihybrid Crosses, page 2]).

Mendelian Genetics

A. Gregor Mendel (1822–1884)

1. An Austrian monk who, through his love and interest in nature, developed the basic ideas of genetics long before chromosomes and genes (i.e., molecular biology) were discovered.
 - a. He developed his ideas by studying plants; in particular, his most famous work involved crosses with **pea plant varieties**.
2. His results and interpretations contrasted with a prevailing (at that time) theme of inheritance called "**blending**"—the concept that **inherited traits mixed to create a composite characteristic** in offspring.

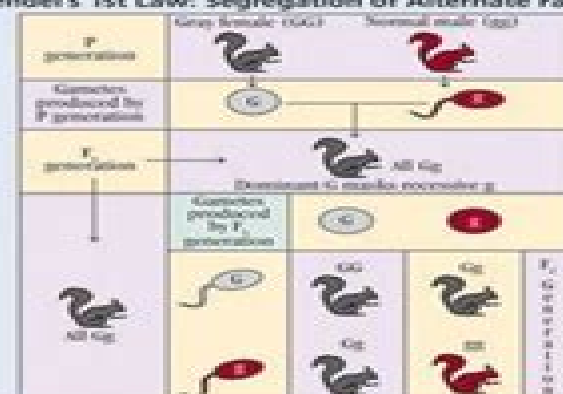
B. Mendel's Genetics Laws

1. Segregation of Alternate Factors & Monohybrid Crosses

- a. Specifically, Mendel discovered that with certain traits, there were individual plants which, if only crossed with other plants just like them, would almost always produce the exact same phenotype.

- i. These individuals were called **true-breeding**.
 - ii. We now call this condition **homozygous**.
- a. He also found that some individuals with similar appearance, when crossed, would not have all offspring of the same kind.
 - i. We now call this condition **heterozygous**.
 - c. Mendel decided to systematically do **single-trait** crosses to determine the causes for the previously stated observations.
 - d. Specifically, a **parental generation (P)** initiated these experimental crosses by using two true-breeding pea plants for opposite phenotypes (e.g., purple versus white flowers).
 - e. **Offspring** from this cross (**F₁**) all showed only one of the traits (e.g., purple flowers), and this trait was called the **dominant trait**.
 - f. Traits from the P generation "**did not blend**" in these F₁ individuals.
 - g. F₁ individuals, the **hybrids**, were cross-pollinated—the **monohybrid cross**—to produce F₂ individuals.
 - h. 1/4 of the F₂ individuals expressed the dominant trait, while 3/4 expressed the trait of the other P parent (e.g., white)—this latter trait was the **recessive form**.
 - i. The expected **phenotypic ratio** of the F₂ individuals in **monohybrid** crosses would be **3:1**.
 - j. The expected **genotypic ratio** of the F₂ individuals in **monohybrid** crosses would be **2:1:1**.
 - k. The diagram at right, called a **Punnett square**, summarizes results of a single-trait cross similar to those done by Mendel on pea plants and other organisms.

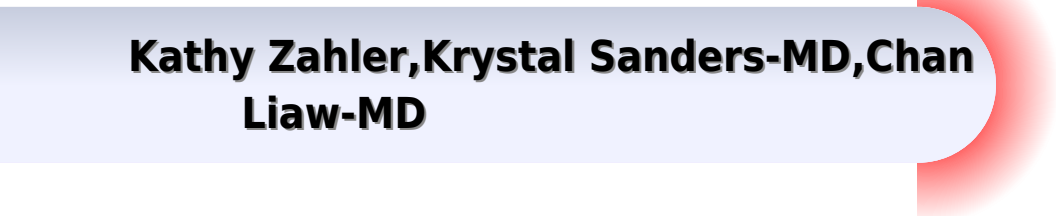
Mendel's 1st Law: Segregation of Alternate Factors



1. Mendel concluded there had to be some physical entities or "**factors**" passed on by each parent of a cross.
 - i. We now know these to be **genes**.
 - ii. He also concluded that these factors came in **pairs**, which then became **separated** (in the production of gametes, which occurs during meiosis) and **recombined** during fertilization.
 - iii. The two P generation individuals had the factors in **alternate forms** called **alleles** (e.g., purple versus white flowers).

Basic Genetics Student Guide

**Kathy Zahler, Krystal Sanders-MD, Chan
Liaw-MD**



Basic Genetics Student Guide:

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Table of Contents Basic Genetics Student Guide

1. Understanding the eBook Basic Genetics Student Guide
 - The Rise of Digital Reading Basic Genetics Student Guide
 - Advantages of eBooks Over Traditional Books
2. Identifying Basic Genetics Student Guide
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Basic Genetics Student Guide
 - User-Friendly Interface
4. Exploring eBook Recommendations from Basic Genetics Student Guide
 - Personalized Recommendations
 - Basic Genetics Student Guide User Reviews and Ratings
 - Basic Genetics Student Guide and Bestseller Lists
5. Accessing Basic Genetics Student Guide Free and Paid eBooks
 - Basic Genetics Student Guide Public Domain eBooks
 - Basic Genetics Student Guide eBook Subscription Services
 - Basic Genetics Student Guide Budget-Friendly Options
6. Navigating Basic Genetics Student Guide eBook Formats

- ePub, PDF, MOBI, and More
- Basic Genetics Student Guide Compatibility with Devices
- Basic Genetics Student Guide Enhanced eBook Features
- 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Basic Genetics Student Guide
 - Highlighting and Note-Taking Basic Genetics Student Guide
 - Interactive Elements Basic Genetics Student Guide
- 8. Staying Engaged with Basic Genetics Student Guide
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Basic Genetics Student Guide
- 9. Balancing eBooks and Physical Books Basic Genetics Student Guide
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Basic Genetics Student Guide
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine Basic Genetics Student Guide
 - Setting Reading Goals Basic Genetics Student Guide
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Basic Genetics Student Guide
 - Fact-Checking eBook Content of Basic Genetics Student Guide
 - Distinguishing Credible Sources
- 13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
- 14. Embracing eBook Trends
 - Integration of Multimedia Elements
 - Interactive and Gamified eBooks

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