

Lecture Outline: Genes, Alleles, and Inheritance

I. Introduction to Genetics and Gregor Mendel

A. Genetics and DNA

1. Chapter primarily focuses on **Mendelian genetics**

B. Gregor Mendel

1. Known as the "**father of genetics**"
2. Monk who worked in the 1800s, but his careful work was unappreciated until the 1900s
3. Conceptually figured out how inheritance works before DNA was even known
4. Exceptional for his time due to his **careful, hardworking, and statistical approach**
 - a. Biologists in the 1800s were generally observational, not experimental or focused on statistics
 - b. Mendel performed experiments with **hundreds to thousands of replicates** to ensure statistical significance
 - c. Conducted **seven major sets of experiments**, each requiring its own plot of land
 - d. His work required years of preparation, such as ensuring plants were true breeding

II. Mendel's Experimental Technique with Pea Plants

A. Pea Plants as a Subject

1. Were a **good choice** because they were easy to grow and experimentally work with

B. Controlling Fertilization

1. **Complete flowers** have both male (stamens producing pollen/male gametes) and female (carpels producing ova/eggs) parts

2. Pea plants can naturally self-fertilize
3. Mendel **prevented self-fertilization** by carefully cutting off the stamens
4. He then **cross-pollinated** by brushing pollen from a chosen male parent onto the carpel of a chosen female parent
5. This technique allowed him to precisely control which plants reproduced with each other

C. Observing Offspring

1. After successful fertilization, the flower develops into a ****fruit**** (which contains the seeds)
2. The seeds are the ****offspring**** of the cross
3. Mendel had to plant these seeds (peas) and wait until the next season for them to grow into plants to observe their characteristics
4. This process was done repeatedly for years, requiring immense patience and careful note-taking

III. Key Genetic Definitions

A. Characteristic vs. Trait

1. **Characteristic**: Any descriptive attribute of an organism (e.g., flower color, eye color, speed of an enzyme)
2. **Trait**: A **specific value** for a given characteristic (e.g., purple or white for flower color; brown or hazel for eye color)
3. Mendel focused on observing and taking notes on only **one characteristic per experiment**

B. Allele, Gene, and Locus

1. **Chromosome**: An enormously long double helix of DNA that contains many different genes
2. **Gene**: A tiny segment of a chromosome (a sequence of nucleotides) that codes indirectly for a protein
 1. Proteins are what give an organism its traits
 2. A gene corresponds to a **characteristic**
3. **Allele**: Different possible versions of a gene

1. An allele corresponds to a **trait**
4. **Locus (plural: loci)**: The specific physical position that any given gene occupies on a chromosome
5. In a **diploid cell**, there are two homologous chromosomes, meaning two loci for each gene, and therefore two alleles for each characteristic

C. Phenotype vs. Genotype

1. **Phenotype**: The observable **trait** or a verbal description of an individual for a given characteristic (e.g., "white flowered," "purple flowered," "fast running")
2. **Genotype**: The **symbolic representation** of the actual set of alleles that an individual has in its cells for a specific gene
 1. Alleles are usually represented by letters: an **uppercase letter for the dominant allele** and a **lowercase letter for the recessive allele** (e.g., P for purple, p for white)
 2. Since diploid organisms have two alleles for each gene, a genotype is represented by two letters (e.g., PP, Pp, pp)

IV. Mendelian Inheritance

A. Mendelian Assumptions/Requirements

1. For a characteristic to be governed by Mendelian genetics, there must be **exactly two alleles** in the population
 1. This corresponds to **exactly two traits** for that characteristic
2. One of those two traits (and its corresponding allele) must be **completely dominant** over the other
 1. A single dominant allele is sufficient to fully express the dominant phenotype

B. Genotype Categories in Diploid Individuals (Mendelian Case)

1. **Homozygous**: Individuals that have two of the same kind of allele
 1. **Homozygous dominant**: Has two dominant alleles (e.g., PP); expresses the dominant trait
 2. **Homozygous recessive**: Has two recessive alleles (e.g., pp); this is the **only way** to express the recessive trait

3. Homozygous individuals are also referred to as **true breeding** because they can only pass on one type of allele for that gene
2. **Heterozygous**: Individuals that have one dominant and one recessive allele (e.g., Pp)
 1. Because of **complete dominance**, heterozygous individuals always express the **dominant phenotype**
 2. The term "heterozygous dominant" or "heterozygous recessive" is not used
 3. A heterozygous individual for a recessive disorder is called a **carrier** because they can pass on the recessive allele without expressing the disorder themselves

C. Monohybrid Cross

1. A cross (reproduction) between two parents that differ in their traits for **only one characteristic** being studied
2. **P (Parental) Generation Cross (True breeding dominant x True breeding recessive)**
 - a. Example: True breeding purple-flowered pea plant (PP) crossed with a true breeding white-flowered pea plant (pp)
 - b. Produces **100% heterozygous (Pp) F1 offspring**
 - c. All F1 offspring exhibit the **dominant phenotype** (e.g., 100% purple flowers)
 1. This was counter-intuitive to Mendel's contemporaries, who expected a mixture or blend of parental traits
3. **F1 Generation Cross (Heterozygous F1 x Heterozygous F1)**
 - a. Example: Pp x Pp (F1 individuals are crossed with each other to produce the F2 generation)
 - b. Results in a **re-emergence of the recessive trait** in the F2 generation
 1. A ****recessive trait**** "recedes" or hides in the F1 generation and then "reappears" in the F2
 2. A ****dominant trait**** "dominates" or is expressed when at least one dominant allele is present

- c. The F2 generation exhibits a characteristic **phenotypic ratio** of approximately **3:1** (dominant phenotype : recessive phenotype)
- d. The F2 generation exhibits a characteristic **genotypic ratio** of approximately **1:2:1** (homozygous dominant : heterozygous : homozygous recessive)
- e. Mendel observed this famous 3:1 ratio in the F2 generation across all seven of his monohybrid experiments

D. Punnett Square (Represents Fertilization)

- 1. A shorthand device used to easily list and predict the possible offspring genotypes and phenotypes from a genetic cross
- 2. The sides and top of the square list the **haploid gametes** (containing a single allele) that each parent can produce via meiosis
- 3. The inner boxes of the Punnett square represent the specific **zygotes** (products of fertilization) that the two parents are able to produce
- 4. It exhaustively lists all possible combinations of gametes, allowing prediction of genotypic and phenotypic ratios in the offspring

V. Dihybrid Cross and Independent Assortment

A. Dihybrid Cross Definition

- 1. A cross in which **two different characteristics** are simultaneously studied and tracked
- 2. The parents typically differ in their traits for both of these characteristics

B. P Generation Cross (Double homozygous dominant x Double homozygous recessive)

- 1. Example: A pea plant homozygous dominant for yellow seeds (YY) and round texture (RR) crossed with a plant homozygous recessive for green seeds (yy) and wrinkled texture (rr) - Genotype: YYRR x yyrr
- 2. Because both parents are homozygous for both genes, each parent produces only **one type of gamete** (e.g., YR from the first parent, yr from the second parent)
- 3. All F1 offspring are **double heterozygous** (e.g., YyRr)

4. All F1 individuals exhibit **both dominant phenotypes** (e.g., yellow and smooth seeds)

C. **F1 Generation Cross (Double heterozygous F1 x Double heterozygous F1)**

1. Each F1 parent (e.g., YyRr) can produce **four different types of gametes** (YR, Yr, yR, yr)
 1. This is due to **independent assortment** during meiosis
2. **Independent Assortment:** The way one homologous chromosome pair (tetrad) lines up during metaphase of meiosis is **independent** of how other homologous pairs line up
 1. This process leads to a significant increase in genetic diversity in the gametes
3. The Punnett square for a dihybrid cross is a 4x4 grid, resulting in 16 possible zygote combinations
4. The F2 generation typically exhibits a characteristic **phenotypic ratio** of approximately **9:3:3:1**
 1. **9**: Individuals showing both dominant traits (e.g., yellow and smooth - parental type)
 2. **3**: Individuals showing one dominant and one recessive trait (e.g., yellow and wrinkled - recombinant type)
 3. **3**: Individuals showing the other dominant and recessive trait (e.g., green and smooth - recombinant type)
 4. **1**: Individuals showing both recessive traits (e.g., green and wrinkled - parental type)
5. Mendel's observation of this 9:3:3:1 ratio provided **experimental evidence for independent assortment**

D. **Probability in Genetics**

1. The production of sperm or eggs via meiosis from a heterozygous individual is analogous to a **coin flip**, with a 0.5 (1/2) probability for either the dominant or recessive allele to be included in a gamete
2. The probability of a particular zygote formation (union of specific gametes) is calculated by **multiplying the individual probabilities** of

those gametes (e.g., $0.5 \times 0.5 = 0.25$ for a specific homozygous zygote)

3. The sum of all possible probabilities for offspring genotypes/phenotypes must always add up to 1 (100%)

VI. Non-Mendelian Genetics (Violations of Mendelian Assumptions)

A. Incomplete Dominance

1. **Violation:** The dominant allele does ****not completely dominate**** over the recessive allele
2. **Characteristics:** There are two alleles in the population, but they result in ****three distinct phenotypes/traits****
3. **Phenotype Expression:**
 - a. **Homozygous dominant:** Shows full expression of the dominant trait (e.g., red flower)
 - b. **Heterozygous:** Exhibits an ****intermediate phenotype**** (e.g., pink flower) that is a blend or partial expression of the dominant trait
 - c. **Homozygous recessive:** Shows the recessive phenotype (e.g., white flower)
4. **Molecular Basis:** In heterozygous individuals, having only one dominant allele means producing half the amount of the corresponding protein compared to a homozygous dominant individual, which may not be sufficient for full phenotypic expression
5. **Phenotypic Ratio (from a heterozygous cross):** The F₂ generation exhibits a **1:2:1 ratio** (dominant : intermediate : recessive)

B. Multiple Alleles

1. **Violation:** The characteristic of interest has ****more than two alleles**** present in the population (e.g., three or more)
2. **Example:** The ****ABO blood group system in humans****
 1. There are three alleles in the human population for this characteristic: I^A , I^B , and i
 2. I^A and I^B alleles code for A and B antigens, respectively, which are expressed on the surface of red blood cells

3. The i allele does not code for any antigens
4. These multiple alleles lead to four possible blood phenotypes:
Type A, Type B, Type AB, and Type O

C. Codominance

1. **Violation:** Two different alleles are ****both fully and simultaneously expressed**** in a heterozygous individual
2. **Example:** The ****ABO blood group system**** (specifically the I^A and I^B alleles)
 1. If an individual inherits both the I^A and I^B alleles (genotype $I^A I^B$), they will have ****Type AB blood****
 2. In Type AB blood, both A antigens and B antigens are fully present on the red blood cells at the same time, demonstrating simultaneous full expression of both traits
3. **Blood Transfusion Rules (Related to Antigens and Antibodies):**
 - a. Antigens are particles on red blood cells, while antibodies are proteins in the blood plasma that recognize and attack foreign antigens
 - b. **Type AB individuals:** Have both A and B antigens, produce no antibodies. They are ****universal recipients of red blood cells**** (can receive from A, B, AB, or O) and ****universal donors of plasma**** (their plasma contains no antibodies to attack recipient cells)
 - c. **Type O individuals:** Have no A or B antigens, produce both anti-A and anti-B antibodies. They are ****universal donors of red blood cells**** (their cells won't be attacked by recipient antibodies) and ****universal recipients of plasma**** (their plasma can receive any type as it has antibodies against A and B cells)

D. Epistasis

1. **Violation:** Involves ****gene interaction**** where the product of one gene affects the expression of another gene
2. **Characteristics:** Two or more different genes are involved in determining a single characteristic
3. **Example:** ****Coat color in rats****

1. Two genes are involved: the **B gene** (which codes for pigment color) and the **C gene** (which codes for pigment production/ expression)
2. The B gene has alleles for black (dominant, B) and brown (recessive, b) pigment
3. The C gene determines whether any pigment is produced and deposited into the hairs
4. If a rat has at least one dominant C allele (CC or Cc), the color determined by the B gene will be expressed (black or brown)
5. However, if a rat is homozygous recessive for the C gene (cc), it will be an **albino (white)**, regardless of the alleles present at the B gene locus, because the pigment cannot be produced or deposited
6. Thus, the C gene's product directly controls whether the B gene's product is expressed

E. Pleiotropy

1. **Violation:** **One gene simultaneously affects more than one characteristic**
2. **Example:** **Sickle cell disease in humans**
 1. A single gene (the hemoglobin gene) and its mutation can affect multiple distinct characteristics:
 2. (1) The presence or absence of **sickle cell disease** (a severe condition if homozygous recessive for the mutant allele)
 3. (2) Providing **increased resistance to malaria** (an advantage if heterozygous for the mutant allele)
3. **Heterozygote Advantage:** Individuals who are heterozygous for the sickle cell allele have enough normal hemoglobin to avoid the severe symptoms of sickle cell disease, but they also gain protection against malaria. This advantage helps keep the mutant allele in the human population, especially in malaria-prone regions.

F. Polygenic Inheritance

1. **Violation:** **One characteristic is affected by multiple different**

genes, with each gene contributing a small, additive effect to the phenotype

2. **Characteristics:** Results in a ****continuum of traits**** (a smooth range of variation) between two extremes, rather than discrete, distinct categories
3. **Example:** ****Human skin color****
 1. Human skin color varies widely from very light to very dark, representing a continuous spectrum
 2. This is because it is controlled by multiple genes (not just one or two), each contributing "dark" or "light" alleles
 3. The overall skin color phenotype depends on the cumulative contribution of all these involved genes

VII. Other Genetic Concepts

A. Nature vs. Nurture

1. ****Phenotype**** is determined by both a ****genetic component (nature)**** and an ****environmental component (nurture)****
2. Genetically identical individuals (e.g., identical twins, certain plants) can exhibit significant phenotypic differences if they are raised or live in different environments (e.g., diet, lifestyle, exposure to elements)
3. This illustrates that genes provide the blueprint, but environmental factors influence how that blueprint is expressed and built

B. Pedigree Analysis

1. A ****pedigree**** is essentially a genetic ****family tree**** used to analyze inheritance patterns in humans
2. It allows geneticists to infer genotypes of individuals, especially for recessive traits (e.g., if a child expresses a recessive trait but both parents show the dominant trait, the parents must both be heterozygous carriers)
3. While historically important for human genetic studies, direct DNA testing is now often used to determine genotypes more directly

C. Genetic Testing (Fetal Analysis)

1. Genetic tests can be performed during pregnancy to check the

genetics of a fetus or embryo for potential genetic disorders, especially if parents are known carriers

2. Two major types of fetal genetic testing mentioned:

1. **Amniocentesis**: A sample of the amniotic fluid (which contains fetal cells) is taken and analyzed
2. **Chorionic Villus Sampling (CVS)**: A sample of tissue is taken from the placenta (an organ that contains fetal DNA) for genetic analysis