Father of Genetics

A. Gregor Mendel

Mendel was an Austrian monk.

1. Mendel formulated two fundamental laws of heredity in the early 1860's.
2. He had previously studied science and mathematics at the University of Vienna.
3. At time of his research, he was a substitute science teacher at a local technical high school.

B. Blending Concept of Inheritance

1. **This theory stated that offspring would possess traits intermediate between those of different parents**.
2. Red and white flowers produce pink; a later return to red or white progeny was considered instability in genetic material.
3. Charles Darwin wanted to develop a theory of evolution based on hereditary principles; blending theory was of no help.
   a. The blending theory did not account for variation and could not explain species diversity.
   b. The particulate theory of inheritance proposed by Mendel can account for presence of differences among member of a population generation after generation.
   c. Mendel's work was unrecognized until 1900; Darwin was never able to use it to support his theory of evolution.

C. Mendel's Experimental Procedure

1. Mendel did a statistical study (he had a mathematical background).
2. He prepared his experiments carefully and conducted preliminary studies.
   a. He chose the garden pea because peas were easy to cultivate, had a short generation time, and could be cross-pollinated.
   b. From many varieties, Mendel chose 22 true-breading varieties (pure) for his experiments.
   c. **True-breeding** varieties had all offspring like the parents and like each other.
   d. Mendel studied 7 simple traits (e.g., seed shape and color, flower color, etc.).
3. Mendel traced inheritance of individual traits and kept careful records of numbers.
4. He used his understanding of mathematical principles of probability to interpret results.
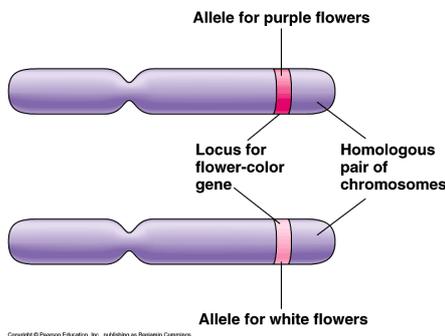
The Monohybrid Cross

A. Cross-pollination Monohybrid Crosses

1. A **hybrid** is the product of parent organisms that are true-breeding for different forms of one trait.

2. A **monohybrid** cross is between two parent organisms true-breeding for two distinct forms of a trait.
3. Mendel tracked each trait through two generations.
   a. **P generation** is the parental generation in a breeding experiment.
   b. **F₁ generation** is the first-generation offspring in a breeding experiment.
   c. **F₂ generation** is the second-generation offspring in a breeding experiment.
   d. He also performed reciprocal crosses of pollen on stigmas (e.g. tall-with-short and short-with tall).

B. Mendel's Results

1. His results were contrary to those predicted by a blending theory of inheritance.
2. He found that the F₁ plants resembled only one of the parents.
3. Characteristic of other parent reappeared in about 1/4 of F₂ plants; 3/4 of offspring resembled the F₁ plants.
4. Mendel saw that these 3:1 results were possible if
   a. F₁ hybrids contained two factors for each trait, one *dominant* and one *recessive*;
   b. factors separated when gametes were formed; a gamete carried on copy of each factor;
   c. Random fusion of all possible gametes occurred upon fertilization.
5. Results of his experiments led Mendel to develop his first law of inheritance:
   a. **Mendel's law of segregation:** Each organism contains two factors for each trait; factors segregate in formation of gametes; each gamete contains one factor for each trait.
   b. Reshuffling of factors explains variations and why offspring differ from their parents.

C. As Viewed By Modern Genetics

Allele for purple flowers

Locus for flower-color gene

Homologous pair of chromosomes

Allele for white flowers

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1. Each trait in a pea plant is controlled by two **alleles**, alternate forms of a gene that occur at the same **gene locus** on homologous chromosomes.
   a. A **dominant allele** masks or hides expression of a recessive allele; it is represented by an uppercase letter.
   b. A **recessive allele** is an allele that exerts its effect only in the **homozygous** state; its expression is masked by a dominant allele; it is represented by a lowercase letter.
2. **Gene locus** is specific location of a particular gene on homologous chromosomes.
3. In Mendel's cross, the parents were true-breeding; each parent had two identical alleles for a trait -- they were **homozygous**, indicating they possess two identical alleles for a trait.
   a. **Homozygous dominant** genotypes possess two dominant alleles for a trait.
   b. **Homozygous recessive** genotypes possess two recessive alleles for a trait.
4. After cross-pollination, all individuals of the F₁ generation had one of each type of allele.
   a. **Heterozygous** genotypes possess one of each allele for a particular trait.
   b. The allele not expressed in a heterozygote is a recessive allele.

D. Genotype Versus Phenotype

1. Two organisms with different allele combinations can have same outward appearance (e.g., *TT* and *Tt* pea plants are both tall; therefore, it is necessary to distinguish between alleles and appearance of organism).

2. **Genotype** refers to the alleles an individual receives at fertilization.
3. **Phenotype** refers to the physical appearance of the individual.

E. Monohybrid Genetics Problems

1. First determine which characteristic is dominant; then code the alleles involved by assigning letters to represent the trait.
2. Determine genotype and gametes for both parents; an individual has two alleles for each trait; each gamete has only on allele for each trait.
3. Each gamete has a 50% chance of having either allele.

F. Laws of Probability

1. **Probability** is the likely outcome a given event will occur from random chance.
   a. With each coin flip there is a 50% chance of heads and 50% chance of tails.
   b. Chance of inheriting one of two alleles from a parent is also 50%.
2. **Multiplicative law of probability** states that the chance of two or more independent events occurring together is the product of the probability of the events occurring separately.
   a. Chance of inheriting a specific allele from one parent and a specific allele from another is 1/2 x 1/2 or 1/4.
   b. Possible combinations for the alleles Ee of heterozygous parents are the following:

   EE = 1/2 x 1/2 = 1/4 eE = 1/2 x 1/2 = 1/4 Ee= 1/2 x 1/2 = 1/4 ee = 1/2 x 1/2 = 1/4

3. Additive law of probability calculates probability of an event that occurs in two or more independent ways; it is sum of individual probabilities of each way an event can occur; in the above example where unattached earlobes are dominant (EE, Ee, and eE), the chance for unattached earlobes is 1/4 + 1/4 + 1/4 = 3/4.

G. The Punnett Square

1. Provides a simple method to calculate probable results of a genetic cross.
2. In a Punnett square, all possible types of sperm alleles are lined up vertical, all possible egg alleles are lined up horizontally; every possible combination is placed in squares.
3. The larger the sample size examined, the more likely the outcome will reflect predicted ratios; a large number of offspring must be counted to observe the expected results; only in that way can all possible genetic types of sperm fertilize all possible types of eggs.
4. We cannot testcross humans in order to count many offspring; in humans, the phenotypic ratio is used to estimate the probability of any child having a particular characteristic.
5. Punnett square uses laws of probability; **it does not dictate what the next child will inherit.**
6. "Chance has no memory": if two heterozygous parents have first child with attached earlobes (likely in 1/4th of children), second child born still has 1/4 chance of having attached earlobes.

H. One-Trait Testcross

1. Mendel performed **testcrosses** by crossing his F_1 plants with homozygous recessive plants.
2. Results indicated the recessive factor with present in the F_1 plants; they were heterozygous.

3. A **testcross** is between an individual with **dominant phenotype and individual with recessive phenotype** to see if the individual with dominant phenotype is homozygous or heterozygous.

Multiple Factor Crosses

A. Dihybrid Crosses

1. A **dihybrid cross** is an experimental cross between two parent organisms that are true-breeding for different forms of two traits; produces offspring *heterozygous* for both traits.
2. Mendel observed that the F_1 individuals were dominant in both traits.

B. Plants to Self-Pollinate
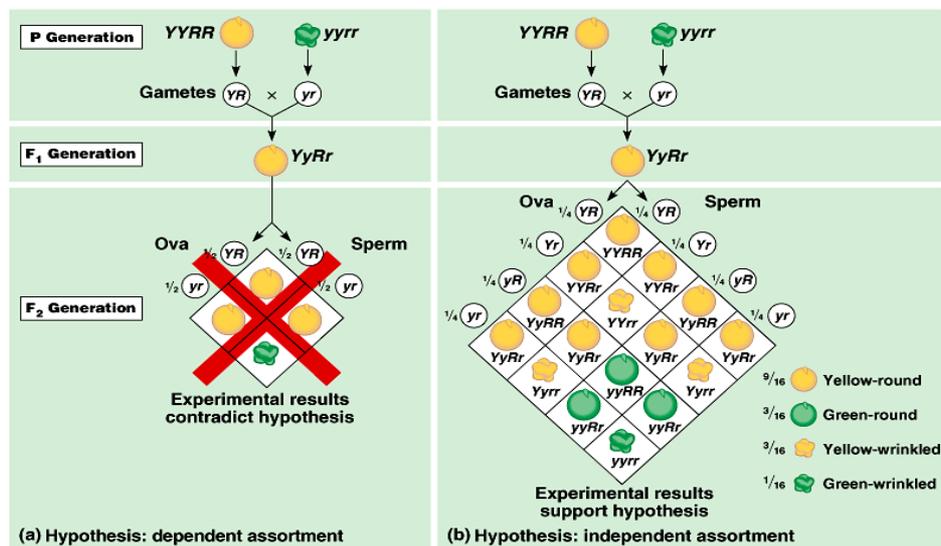
1. Mendel observed four phenotypes among F_2 offspring; he deduced second law of heredity.
2. **Mendel's law of independent assortment** states members of one pair of factors assort independently of members of another pair; all combinations of factors occur in gametes.
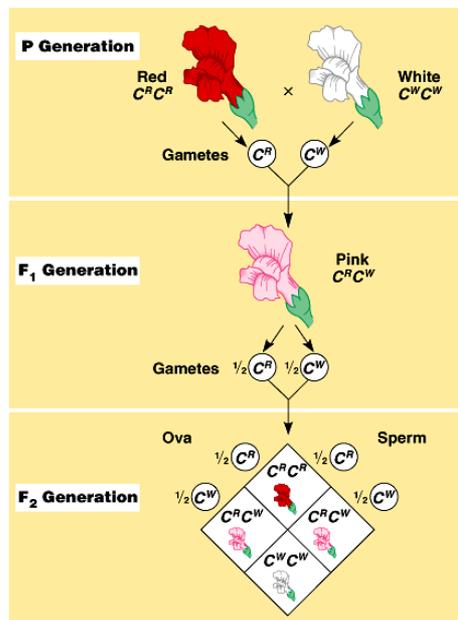
C. Dihybrid Genetics Problems

1. Laws of probability indicate a 9:3:3:1 phenotypic ratio of F_2 offspring resulting in the following:
   a. 9/16 of the offspring are dominant for both traits;
   b. 3/16 of the offspring are dominant for one trait and recessive for the other trait;
   c. 3/16 of the offspring are dominant and recessive opposite of the previous proportions; and
   d. 1/16 of the offspring are recessive for both traits.

2. The Punnett Square for Dihybrid Crosses
   a. A larger Punnett square is used to calculate probable results of a dihybrid cross.
   b. A phenotypic ratio of 9:3:3:1 is expected when heterozygotes for two traits are crossed and simple dominance is present for both genes.
   c. Meiosis explains these results of independent assortment.

D. Two-Trait Test Cross

1. A **dihybrid test cross** tests if individuals showing two dominant characteristics are homozygous for both or for one trait only, or is heterozygous for both.
2. If an organism heterozygous for two traits is crossed with another recessive for both traits, expected phenotypic ratio is 1:1:1:1.
3. In dihybrid genetics problems, the individual has four alleles, two for each trait.

Extensions of Mendelian Genetics

P Generation — Red $C^R C^R$ × White $C^W C^W$ — Gametes C^R C^W

F$_1$ Generation — Pink $C^R C^W$ — Gametes ½ C^R ½ C^W

F$_2$ Generation — Ova ½ C^R ½ C^W Sperm ½ C^R ½ C^W — $C^R C^R$, $C^R C^W$, $C^R C^W$, $C^W C^W$

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A. **Incomplete Dominance and Codominance**

1. **Incomplete dominance:** offspring show traits intermediate between two parental phenotypes.
   a. Red and white-flowered four o'clocks produce pink-flowered offspring.
   b. Incomplete dominance has a biochemical basis; level of gene-directed protein production may be between that of the two homozygotes.
   c. One allele of a heterozygous pair only partially dominates expression of its partner.
   d. This does not support a blending theory; parental phenotypes reappear in F$_2$ generation.
2. **Codominance** is a pattern of inheritance in which both alleles of a gene are expressed.
   a. A person with AB blood has both A and B antigens on their red blood cells.
   b. With codominance, both alleles produce and effective product.

B. Genes That Interact

1. More than one pair of genes may interact to produce the phenotype.
2. **Epistasis:** absence of expected phenotype as a result of masking expression of one gene pair by the expression of another gene pair.
   a. The homozygous recessive condition masks the effect of a dominant allele at another locus.
   b. Crossing sweet pea plants produces purple; F$_2$ generation has a 9:7 rather than 9:3:3:1 dihybrid ratio; explained by homozygous recessive blocking production of a metabolic enzyme.
   c. Albino animals inherit allelic pair (*aa*) preventing production of melanin, expression of eye, hair, color.

C. Pleiotropy

1. **Pleitropy**: a single gene exerts an effect on many aspects of an individual's phenotype.
   a. Marfan syndrome: a mutant gene is unable to code for production of a normal protein, fibrillin.
   b. Results in the inability to produce normal connective tissue.

    c. Individuals with Marfan syndrome tend to be tall and thin with long legs, arms, and fingers; are nearsighted; and the wall of their aorta is weak.

    d. From his lanky frame and other symptoms, Abraham Lincoln may have had Marfan syndrome.

    e. Sickle cell anemia causes crystallization of hemoglobin in red blood cells.

    f. Results in scicle shape of cell causing blood clotting which effects a number of organ systems.

D. Multiple Alleles

1. There may be more than two alleles for one locus, but each individual inherits only two alleles.
2. A multiple allele system is peppered moths has three possible alleles for wing color in order of dominance: *M>M'>m*; therefore, there are three possible phenotypes.
3. The ABO system of human blood type involves three alleles (A, B, and O).
4. As a result, there are four possible phenotypes or blood types: A, B, AB, and O.

E. Polygenic Inheritance
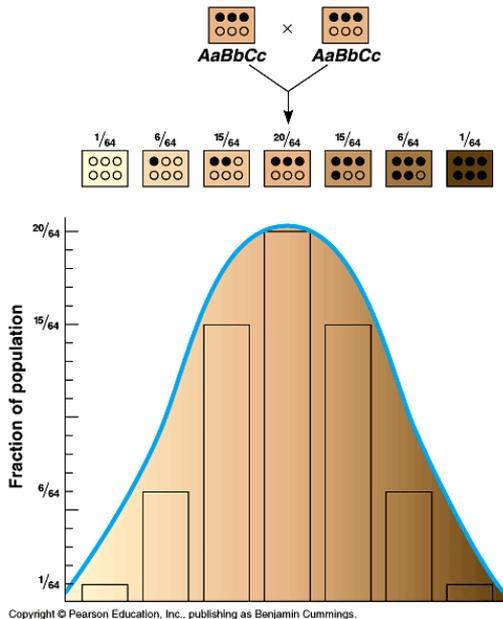
1. **Polygenic inheritance** occurs when a trait is controlled by several allelic pairs at different loci.
2. Allelic pairs at different loci on a chromosome or on different chromosomes all control one trait.
3. Gene alleles can be contributing or non-contributing.
4. Contributing alleles have an addictive effect, resulting in quantitative variations.
5. Examples include seed color in wheat and skin color and height in humans.
6. Polygenic traits are subject to environmental effects that cause intermediate phenotypes; so they produce continuous variations whose frequency distribution forms a normal (bell-shaped) curve.

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F. Environment and the Phenotype

1. Both genotype and environment affect phenotype; relative importance of both influences vary.
2. Aquatic environment (above and below water level) influences the phenotype of water buttercup, *Ranunculus peltatus*.
3. Temperature can affect the phenotypes of some plants (e.g., primroses) and animals (e.g., Siamese cats, Himalaya rabbits).

Images: Campbell, Neil and Reece, Jane. *Biology* (6th ed.) San Francisco: Benjamin Cummings

Modified Notes: Mader, Sylvia. *Biology* (7th ed) New York: McGraw Hill Publishing