Mendel Used the Scientific Approach to Identify Two Laws of Inheritance

Mendel's Experimental, Quantitative Approach

- **Character**: a heritable feature that varies among individuals
- **Trait**: each variant for a character
- Mendel used peas to test traits b/c of short generation time, many varieties, large number of offspring, and easy to control
- **True Breeding**: after years of self-pollination, same variety parent plant is produced
- **Hybridization**: mating, or crossing, of two true-breeding varieties
- **P generation**: true-breeding parents
- **F1 generation**: first filial generation; produces **F2 generation**

The Law of Segregation

- If blending model was correct, F1 of white-flowered and purple-flowered plants would have pale purple flowers
- Heritable trait for white flowers did not disappear, but stay somewhat hidden
- Purple flowers are dominant, white flowers are recessive
- Mendel's Model
    - Alternative versions of genes account for variations in inherited characters
        - **Alleles**: alternate versions of genes
        - Each genes is a sequence of nucleotides at a specific place, or locus, along particular chromosome
        - DNA at locus can vary in information
    - For each character, an organism inherits two copies of a gene,one from each parent
        - A genetic locus is represented 2x in diploid cell, 1x on each homolog
        - If the two alleles at a locus differ, then one, the **dominant allele**, determines the organism's appearance; the other, the **recessive allele**, has no noticeable effect on the organism's appearance
    - The two alleles for a heritable character segregate during gamete formation and end up in different gametes; The Law of Segregation
        - An egg or sperm only gets one of the two alleles present in the somatic cells of the organism making the gamete
        - Segregation corresponds to the distribution of the two members of a pair of homologous chromosomes to different gametes in meiosis
    - **Punnett Square**: illustrates combinations of alleles
- Useful Genetic Vocabulary
    - **Homozygous**: organism that has a pair of identical alleles for a character
        - PP or pp "Breed true"
    - **Heterozygous**: organism that has two different alleles for a gene
        - Produce gametes with different alleles
    - **Phenotype**: organism's appearance, physiological, or observable traits
    - **Genotype**: organism's genetic makeup
    - PP and Pp have same phenotype but different genotype

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The Law of Segregation *cont.*

- Test Cross
    - Test cross with recessive homozygous to determine mystery organism's genotype
    - Reveals organism's genotype

The Law of Independent Assortment

- **Monohybrids**: heterozygous for the particular character being followed in the cross
- Mendel identified second law by following two characters at the same time
- **Dihybrids**: individual heterozygous for 2 characters being followed in cross (YyRr)
- Are alleles packaged together and passed on? Dependent assortment hypothesis
- Independent assortment hypothesis-alleles segregate independently of each other
- **Law of Independent Assortment**: each pair of alleles segregates independently of each other pair of alleles during gamete formation
    - Applies to genes on different chromosomes/not homologous

<u>The Laws of Probability Govern Mendelian Inheritance</u>

The Multiplication and Addition Rules Applied to Monohybrid Crosses

- **Multiplication Rule**: to determine the probability of two or more independent events will occur together, we multiply the probability of one event by the probability of the other event
- **Addition Rule**:the probability that any one or two mutually exclusive events will occur is calculated by adding their individual probabilities

Solving Complex Genetics Problems with the Rules of Probability

- A dihybrid or other multicharacter cross is equivalent to two or more independent monohybrid crosses occurring simultaneously
- Knowing probabilities from Punnett Square, we can use multiplication rule to determine probability of each genotype
- Rules of probability give the chance of various outcomes

<u>Inheritance Patterns are Often More Complex than Predicted by Simple Mendelian Genetics</u>

- Relationship between phenotype & genotype is rarely as straightforward as peas

Extending Mendelian Genetics for a Single Gene

- Inheritance of characters deviates from simple Mendelian patterns when alleles are not completely dominant or recessive, when particular gene has 2+ alleles, or when a single gene produces multiple phenotypes
- Degrees of Dominance
    - Alleles can show different degrees of dominance and recessiveness in relation to each other
    - **Complete Dominance**: phenotypes of the heterozygote and dominant homozygote are indistinguishable
    - **Incomplete Dominance**: neither allele is dominant; blending hypothesis
    - **Codominance**: the two alleles can affect the phenotype in separate, distinguishable ways

<u>Inheritance Patterns are Often More Complex than Predicted by Simple Mendelian Genetics</u>

Extending Mendelian Genetics for a Single Gene

- Degrees of Dominance *cont.*
    - The Relationship Between Dominance and Phenotype
        - Dominant because it shows in phenotype, not because it subdues recessive
        - The observed dominant/recessive relationship of alleles depends on the level at which we examine phenotype
            - **Tay-Sachs Disease**: inherited disorder in humans;
    - Frequency of Dominant Alleles

            - Dominant gene might not show often as it is may not be found so frequently
- Multiple Alleles
    - Blood type determined by 3 alleles: I^A, I^B, i ⇒ A, B, AB, O
- Pleiotropy
    - Most genes have multiple phenotypes
    - Responsible for multiple symptoms associated with diseases
    - Single gene can affect multiple characteristics

Extending Mendelian Genetics for Two or More Genes

- Epistasis
    - Phenotypic expression of a gene at one locus alters that of a gene at a second locus
    - BB/Bb- Black fur, bb- brown fur, EE/Ee- black/brown fur, ee- yellow fur
    - 9:3:3:1
- Polygenic Inheritance
    - Quantitative characters: characters that vary in the population in gradations along a continuum
        - Indicates polygenic inheritance
            - Additive effect of two or more genes on a single phenotypic character
            - Converse of pleiotropy
    - Skin variation from AABBCC (dark) to aabbcc (light)
        - Can be affected by environmental factors

Nature and Nurture: The Environmental Impact on Phenotype

- Phenotype depends on genotype and environment
- Norm of reaction: range of phenotypic possibilities due to environmental influences

Integrating a Mendelian View on Heredity and Variation

- Phenotype- special characteristics or organism in entirety- physical appearance, anatomy, physiology, and behavior
- Genotype- individual alleles or entire genetic makeup
- Mendel's laws explain heritable variations based on genes

<u>Many Human Traits Follow Mendelian Patterns of Inheritance</u>

Pedigree Analysis

- Pedigree: family tree describing the traits of generations
- Helps to calculate probability of future child generating genotype/phenotype

Recessively Inherited Disorders

- The Behavior of Recessive Alleles
    - Aa produces enough proteins to not show disorder, only shown in aa
    - Carriers: Heterozygous; Aa; may transmit recessive alleles
- Cystic Fibrosis
    - Most common lethal genetic disease in US
    - Normal allele codes for membrane protein that functions in transport of chloride ions between certain cells and the extracellular fluid
    - Defective in children who inherit 2 recessive alleles
    - High concentration of extracellular chloride causes mucus buildup
- Sickle-Cell Disease
    - Most common disorder amongst Africans