Inheritance

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<u>Mendel and the Black Box</u>

- The concept of inheritance is ancient but historically poorly understood
  - Kids often look like their parents
  - How these traits were transmitted from generation to generation was, for a long time, a mystery

- Genetics is the study of inheritance
- The field of genetics began with the work of Gregor Mendel
    - He had no knowledge of chromosomes, meiosis, or DNA

- Mendel used the scientific method to study inheritance
  - *Pisum sativum*, the common garden pea was his experimental organism
- Mendel began with some original pea plants
  - Parents
- Mendel performed numerous breeding experiments, generating offspring
  - Children

- Mendel observed multiple generations of these plants, and applied mathematical analysis to his observations
- Mendel was able to make some powerful inferences (conclusions)
  - He was the first to perceive a set of principles that govern inheritance

<u>Mendel's inferences</u>

  - The basic units of inheritance are material elements
  - These elements come in pairs
  - These elements retain their character through many generations
  - Pairs of these elements separate during the formation of gametes
- Our understanding of each of these points has increased since Mendel's time

<u>Experimental Subject: *Pisum sativum*</u>

- Flowers are reproductive structures
  - In peas, they have both male and female parts
- Pollination is a means of sexual reproduction
  - Pea plants self-pollinate
    - Sexual reproduction with itself
    - Normal mode of pollination in peas
  - Pea plants can be made to cross-pollinate
    - This allows sexual reproduction between two different individuals

- A pea plant starts as a single pea in a pod
- Each pea within a pod contains one embryo
- Each embryo is a separate individual
  - Each was produced by a separate fertilization event from one sperm and one egg
  - Each embryo can have unique traits
  - Thus, peas in a pod are not really all alike

- Mendel used pea plants that varied for seven different characters
  - e.g., Flower color
- Each character had two contrasting traits
  - e.g., Purple or white flowers
- For each character, two traits exist
  - One trait was termed "dominant"
    - e.g., Purple flowers are dominant
  - The other trait was termed "recessive"
    - e.g., White flowers are recessive

- Dominant traits are normally visible, but recessive traits can sometimes remain hidden
  - These terms will be discussed more later

- Phenotype: A characteristic or feature of an individual
  - A phenotype is typically an adjective
  - e.g. "Purple" or "white" flowers, etc.
- Genotype: The genetic makeup of an individual (this influences a specific phenotype)
  - A genotype is typically an abbreviated notation of paired upper- and/or lower-case letters
  - e.g. "AA", "Aa", or "aa"

The Experiment

- The parental generation is referred to as the "P generation"
- The offspring of the P generation are termed the "F_1 generation"

- "First filial generation"
- The offspring of the F_1 generation are termed the "F_2 generation"
  - "Second filial generation"

- Mendel began his experiments with "true-breeding" plants
  - e.g., All purple-flowered plants self-pollinate to produce only purple-flowered plants
  - These true-breeding plants were used for the P generation

- True-breeding P generation
  - "purple flowers" x "white flowers"
- F_1 generation is produced and the seeds grown
  - <u>All</u> of the flowers were purple
    - No white flowers were present
    - Flowers were not blended "light purple"
  - Purple is dominant, white is recessive
    - The white color "hid" behind purple

- F_1 is then inbred - crossed with itself
  - Self-pollination of the F_1 produced the F_2 generation
- F_2 generation seeds grew and they produced flowers

- The F_2 generation had purple <u>and</u> white flowers
  - ¾ purple flowers = 705
  - ¼ white flowers = 224
    - *3:1* ratio *purple:white* (dominant:recessive)
  - The other traits Mendel studied also displayed this same 3:1 ratio

<u>What did Mendel learn?</u>
- No "blending" of characteristics
- Heredity is due to the transmission of discrete elements
  - White flowers were absent from the F_1
  - White flowers reappeared in the F_2
  - Mendel inferred that the F_1 individuals retained a white-flower element
    - Elements must be discrete units
- Traits are caused by pairs of elements
  - F_1 individuals must possess a purple-flower element
  - F_1 individuals also possess a white-flower element

The Experiment Illustrated

- Mendel's pairs of elements represent pairs of genes
  - Paired genes exist on pair of homologous chromosomes
- Alternative forms of genes are termed "alleles"
  - e.g., A purple allele (A) and white allele (a) exist for the flower color gene

- Crossed a P1 with purple flowers and a P2 with white flowers
  - Each is homozygous dominant (AA) or homozygous recessive (aa)
- Produced F1 with all purple flowers
  - All are heterozygous (Aa)
  - Possible combinations are all the same
  - This can be demonstrated with a Punnett square

- F1 self-pollinated to produce F2
  - A mix of purple and white flowered offspring result
  - All genotype combinations can be produced
  - Phenotype ratio is 3:1 purple to white
  - Ratios of different genotypes and phenotypes are best illustrated using a Punnett square

Mendel's Law of Segregation

- Individuals possess two alleles for each gene
- These alleles separate during gamete production
  - (Since alleles reside on chromosomes, alleles separate when homologous chromosomes separate during meiosis)

Dihybrid Crosses

- Mendel performed single-cross experiments with single characters
  - e.g., Purple vs. white flowers
  - "Monohybrid cross"
- Mendel also performed double-cross experiments with two characters
  - "Dihybrid cross"
  - e.g., Purple tall x White dwarf

- P: purple tall (AABB) x white dwarf (aabb)
- F_1 generation: All purple tall (*AaBb)*
- F_2 generation: Four different phenotypes

- FOIL method to determine gametes that can be produced by F_1 with genotype AaBb

- F_2 generation phenotypes
  - 9/16 purple tall
  - 3/16 white tall
  - 3/16 purple dwarf
  - 1/16 white dwarf
- 9:3:3:1 phenotypic ratio for F_2
  - Same ratio obtained with other dihybrid pairings

- F_2 generation
  - 9/16 purple tall
  - 3/16 white tall
  - 3/16 purple dwarf
  - 1/16 white dwarf
- 3:1 phenotypic ratios for specific characters
  - purple:white and tall:dwarf

- 3:1 phenotypic ratios
  - purple:white
  - tall:dwarf
- These 3:1 ratios are superimposed upon each other to produce the 9:3:3:1 ratio
  - The transmission of one trait did not affect the transmission of the other trait

Mendel's Law of Independent Assortment

- During gamete formation, gene pairs assort independent of one another
  - The transmission of one character does not influence the transmission of another character
  - (The independent assortment of gene pairs is a result of the independent assortment of chromosomes during meiosis)

Reception of Mendel's Ideas

- Though Mendel's work was widely available, nobody grasped its significance for over three decades
- Mendel's work was rediscovered in 1900
  - 16 years after his death
- The field of genetics is founded upon Mendel's work

Non-Mendelian Genetics

- Some patterns of inheritance are more complex than those studied by Mendel

- Mendel's traits
  - Governed by one gene with two alleles
  - Only two phenotypes exist
- Many other traits
  - More than two alleles for many genes
  - Governed by multiple genes

Multiple Alleles

- Human blood types
  - A, B, AB, and O
  - Determined by types of glycoproteins on the surface of red blood cells
  - Type of glycoprotein is genetically determined
    - Single gene on chromosome 9

- Each individual has two alleles of this gene
  - Identical or non-identical
- Multiple alleles of this gene exist in the population (three in this case)
  - "I^A" allele → "A" molecule
  - "I^B" allele → "B" molecule
  - "i" allele →"O" = inactive (no molecule encoded)

- Six combinations of these alleles produce four blood types

Blood type	Genotype	Molecules present
– Type A	I^AI^A or I^Ai	"A" molecule present
– Type B	I^BI^B or I^Bi	"B" molecule present
– Type AB	I^AI^B	"A" and "B" present
– Type O	ii	neither is present

- The "A" and "B" alleles both display normal dominant/recessive relationships with the "O" allele
- Neither "A" nor "B" is dominant over the other
  - "A" and "B" are "codominant"

Polygenic Inheritance

- Some traits are governed by multiple genes
  - "Polygenic traits"
  - Several genes contribute to a character
  - Many examples of polygenic inheritance

    - Human height, skin color, etc.

- Polygenic traits show continuous variation
  - Not "either-or" variation
- Frequencies of variants display a bell curve
  - Most individuals fall near an average value
  - Bell curve is also called "normal distribution"

Plieotropy and Epistasis

- Plieotropy - One gene can affect multiple traits
  - Sickle Cell Anemia
  - Marfan's Syndrome
  - Cystic Fibrosis
- Epistasis - Two unrelated genes can interact
  - Lab coat color
  - Rooster comb

Genes and Environment

- Genes code for and influence traits, but…
- Environmental factors can also influence traits
  - External factors can affect the phenotypic expression of a trait
    - e.g., Nutritional status influences health and development of infants and children
    - e.g., Soil pH influences the color of hydrangea flowers