Observing Patterns in Inherited Traits

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Mendel and the Black Box

- 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
  - Austrian monk and naturalist
  - Studied inheritance in the mid-1800s
    - His work was performed in a monastery in what is now the Czech Republic
    - He had no knowledge of chromosomes, meiosis, or DNA

- Mendel used the scientific method to study inheritance in garden peas
- Mendel made observations of multiple generations of the plants
- Mendel 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

- Mendel's inferences
  - 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

Experimental Subject: *Pisum sativum*

- 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

How to cross-pollinate a pea plant

- Cross-pollination of a pea plant requires that the male portions of a flower are removed
  - This individual is "mom", since only the female parts remain, with eggs
- A flower with male parts can be used to supply pollen for desired crossings
  - This individual is "dad", since it would supply sperm
- The pea pod produced by the mom contain peas
  - These are the offspring from this mating

- Each pea within a pod contains an embryo
- Each embryo is a separate individual
- The peas in a pod are genetically different from each other

- Mendel used pea plants that varied for seven different characters
  - e.g., Flower color
- Each character had two contrasting traits
  - One trait was considered dominant
    - e.g., Purple flowers
  - The other trait was considered recessive
    - e.g., White flowers

- A characteristic or feature of an individual is called a "**phenotype**"
  - A phenotype is typically an adjective
  - e.g. "Purple" or "white" flowers, etc.
- The genetic makeup of an individual that influences a phenotype is called a "**genotype**"
  - A genotype is typically an abbreviated notation of paired upper- and/or lower-case letters
  - e.g. "AA", "Aa", or "aa"

The Experiment

- Alternative forms of genes are termed "**alleles**"
  - Pairs of genes, or alleles, exist on pair of homologous chromosomes
    - e.g., Purple (A) and white alleles (a) exist for the flower color gene

- Alleles are paired in two basic ways
  - Homozygous (Identical pairs = "AA" or "aa")
  - Heterozygous (Non-identical pairs = "Aa")

- Mendel used a parent generation (P) to produce two generations of offspring
- Direct offspring of the P generation are called the "F_1 generation"
- Subsequent offspring from interbreeding the F_1 generation are called the "F_2 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
- Mendel's F_1 and F_2 generations developed according to the laws of inheritance

- The F_1 generation was all purple
- The F_2 generation had purple and white flowers in a 3:1 ratio
- The other traits Mendel studied also displayed this same 3:1 ratio

What did Mendel learn?

- No "blending" of characteristics
  - No "light purple" flowers
- Heredity is due to the transmission of discrete elements, or genes (as we know them today)
  - 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 allele
    - Genes must be discrete units
- Traits are caused by pairs of genes
  - F_1 individuals must possess a purple-flower allele
  - F_1 individuals also possess a white-flower allele

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
  - 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
- 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)

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^O	"A" molecule present
Type B	I^BI^B or I^BI^O	"B" molecule present
Type AB	I^AI^B	"A" and "B" present
Type O	I^OI^O	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

- One gene can affect multiple traits
  - Sickle Cell Anemia
  - Marfan's Syndrome
  - Cystic Fibrosis

Genes and Environment

- Genes influence traits
- Environmental factors also influence traits
  - External factors that 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