

Introduction to Genetics

History of genetic:

- The history of genetics (Modern genetics) began with the work of the Augustinian friar Gregor Johann Mendel. His work on pea plants, published in 1866, described what came to be known as Mendelian Inheritance.
- In 1902, Walter Sutton and Theodore Boveri, using dyes synthesized by the German organic chemistry industry, observed that "colored bodies" in cells behaved in ways parallel to the hypothetical agents of heredity proposed by Mendel. These bodies were called chromosomes.
- In 1905, Nettie Stevens observed in *Tenebrio* beetles that all pairs of homologous chromosomes are the same size, except for one pair which determines sex (X & Y).
- In 1944, Oswald Avery identified DNA as the genetic material. Pieces of DNA can transfer genes into bacteria cells, and transform them genetically.
- In 1953, Rosalind Franklin and Maurice Wilkins showed that DNA is a double helix.
- In 1970, Temin and Baltimore discovered reverse transcriptase in retroviruses; an enzyme later used to clone genes based on the RNA encoding the product.
- In 1987, Kary Mullis invented the polymerase chain reaction (PCR), using a thermostable enzyme from a thermophilic bacterium discovered by Thomas Brock at a geyser in Yellowstone.
- The first bacterial genome sequence, *Haemophilus influenzae*, was completely determined in 1995.
- In 1996, Ian Wilmut cloned the lamb Dolly from adult mammary gland tissue.
- In 2000, Completion of the human genome.

Genetic terminology:

Genetic: is a branch of biology concerned with the study of genes, genetic variation, and heredity in organisms.

Genome is the complete set of instructions for making an organism. It contains the master blueprint for all cellular structures and activities for the lifetime of the cell or organism

Heredity (also called inheritance or biological inheritance), is the passing on of traits from parents to their offspring; either through asexual reproduction or sexual reproduction, the offspring cells or organisms acquire the genetic information of their parents.

Offspring is another name for the children of a male and female parent.

Traits are specific characteristics that vary from one individual to another such as ear lobe attachment, brow line, or chin type.

Genes are small sections of DNA that code for a specific trait, their coded information determines the nature and development of organisms the code is the order of the bases (letters). The human genome is estimated to comprise more than 25,000 genes.

Genotype is the genetic makeup of an organism, a description of the genes it contains e. g: AA or Aa or aa.

Phenotype is the characteristics that can be observed in an organism, for example red hair or brown hair. Phenotype is determined by interaction of genes and environment

Allele is a different form of a particular gene, positioned in the same relative place (locus) on a homologous chromosome. Each allele is represented by a letter.

Chromosomes: bundles of DNA that make up your genome, where genes are located. The 3 billion bp in the human genome are organized into 24 distinct physically separate chromosomes. All genes are arranged linearly along the chromosomes. The nucleus of most human cells contains two sets of chromosomes, 1 set given by each parent. Each set has 23 single chromosomes; 22 autosomes and a sex chromosome (X or Y).

Dominant and Recessive Genes: Two alleles are inherited for each gene in simple single trait crosses:

- **Pure dominant** - an organism with two dominant genes
- **Pure recessive** - an organism with two recessive genes

Homozygous: having two identical alleles for a trait. Also called **Purebred**.

Heterozygous: have two different alleles for a trait. The organism has one dominant and one recessive gene. The dominant gene shows, the recessive trait is hidden. Also called hybrid.

Monohybrid crosses: which are mating between individuals that differ in only on the trait, or the tracking of the inheritance of a single trait.

Dihybrid cross: cross between parents that are heterozygous for 2 traits.

True breeding is a term used to describe organisms that produce offspring identical to them if allowed to self-pollinate. True-breeding means there is no new genetic material being introduced into the offspring.

Self-pollination: the pollen and egg are derived from the same plant

Cross pollination: the transfer of pollen from the anther of a flower to the stigma of a flower on a different plant. Cross pollination produces offspring that are different from their parents and each other.

Blending theory of heredity:

- Pre-Mendelian theory of heredity proposing that hereditary material from each parent mixes in the offspring; once blended like two liquids in a solution, the hereditary material is inseparable in the offspring
- Individuals of a population should reach a uniform appearance after many generations
- Once traits blended, they cannot be separated out to appear again in later generations.

Particulate theory of heredity

states that discrete "particles" (now referred to as genes) are passed from parents to offspring. the character of the offspring is not a smooth blend of essences from the parents. Mendel also knew that particles may not be present in every generation, but they remain and keep their ability to be expressed in later generations. For example, perhaps one of your parents has blue eyes and the other brown eyes. You yourself have brown eyes but you keep the gene for blue eyes, which can be expressed in your offspring if you mate with someone who also carries the gene for blue eyes.

Introduction to Gregor mendel:

- known as the father of Genetics
- Augustinian monk
- Studied at University of Vienna from 1851-1853
- Mendel used a quantitative approach to his experimentation.
- Discovered principles of genetics.
- Explained some basic laws of genetics.

Mendel chose pea plant as his experimental organisms:

Mendel study of genetics grew out of his interest in ornamental flowers. Prior to his work with pea plants, many plant breeders had conducted experiment aimed at obtaining flowers with new varieties of colors. When two distinct individuals with different characteristic are mated (or crossed) to each other, this called a hybridization experimented and offspring are referred to as Hybrid.

The parental plants are called the parental (**P**) **generation**. Their hybrid offspring represent the next generation called the **first filial (F1) generation**. Mendel allowed the F1 generation to self-fertilize to produce the next generation called **the second filial (F2) generation**

Mendel's work is an elegant model of experimental design and analysis!

He provided a remarkable insight into the methodology necessary for good experimental biology. The essential components of his research are:

He chose the garden pea (*Pisum sativum*) due its ideal characteristics:

A: easy to grow-interbreed















B: self-fertilizing

C: easily crossbred in designed experiments

D: reproduces well and mature in a single season.

1. He worked with seven-unit characters. These are visible features that were each represented by two contrasting traits Figure 1.
2. He established TRUE-BREEDING varieties in which the seeds were available from local seed merchant.

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Trait	Characteristics		F ₂ Results*	
	*Dominant	*Recessive	Dominant	Recessive
Stem length	Tall 	Short 	787	277
Pod shape	Inflated 	Constricted 	882	299
Seed shape	Round 	Wrinkled 	5,474	1,850
Seed color	Yellow 	Green 	6,022	2,001
Flower position	Axial 	Terminal 	651	207
Flower color	Purple 	White 	705	224
Pod color	Green 	Yellow 	428	152

*All of these produce approximately a 3:1 ratio. For example, $\frac{787}{277} = \frac{3}{1}$.

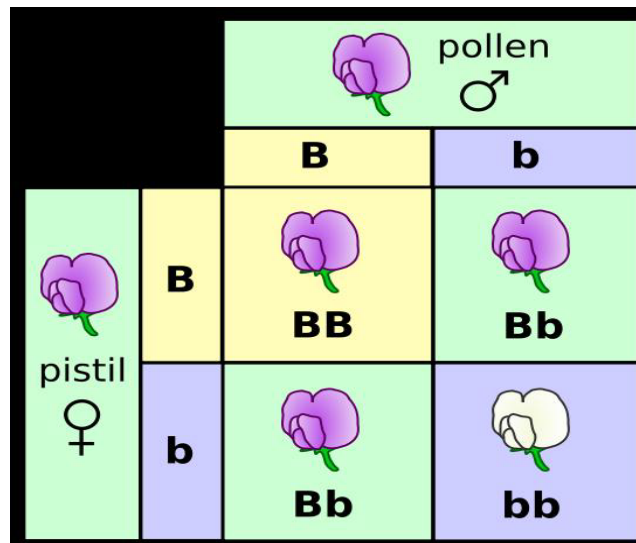
Factors that contributed to Mendel's success in an area where other failed:

1. Choice of suitable organism.
2. Restricted his experiments to one or few pairs of contrasting traits
3. Kept accurate quantitative records.

Mendel laws of inheritance

Mendel postulated three laws, which are now called after his name as Mendel's laws of heredity. These are:

A. Law of Dominance: When two homozygous individuals with one or more sets of contrasting characters are crossed, the characters that appear in the F1 hybrids are dominant characters and those do not appear in F1 are recessive characters.



B. Mendel's law of segregation: Allele pairs segregate during gamete formation (meiosis) and the paired condition is restored by the random fusion of gametes at fertilization.

1. Each trait has two genes, one from the mother and one from the father.
2. Traits can be either dominant or recessive.
3. A dominant trait only needs one gene in order to be expressed.
4. A recessive trait needs two genes in order to be expressed
5. Egg and sperm are sex cells called gametes.
6. Segregation is the separation of alleles during gamete formation.

- C. Law of Independent Assortment:** The genes for different traits are inherited independently of each other. (when two or more characteristics are inherited, individual hereditary factors assort independently during gamete production, giving different traits an equal opportunity of occurring together).

Mendel Did A Dihybrid cross:

Mendel performed a second series of cross that are called dihybrid crosses because the offspring are dihybrid, they are product of two different strains that differ in regard to two traits. For example, he crossed tall plant having green pods with short plants having yellow pods. The F1 plants showed both dominant characteristics. as before. Mendel then allowed the F1 plants to self pollinate. Two possible results could occur in F2 generations:

1. If the dominant factor (TG) always segregations into the gametes together. And the recessive factors (tg). Then there would be two phenotypes among the F2 plants, tall plants with green pods and short plants with yellow pods.
2. If the four factors segregate into the gametes independently. Then there would be four phenotypes among the F2 plants, tall plants with green pods, tall plants with yellow pods, Short plants with green pods, and short plants with yellow pods.

Mendel's Hypothesis:

- 1: Alternative forms of genes are responsible for variations in inherited characters (alternative forms are now called alleles)
- 2: For each character, an organism inherits two alleles, one from each parent
- 3: If the two alleles differ, one is fully expressed (dominant allele) whereas, the other is completely masked (recessive allele)
- 4: The two alleles from each character segregate during gamete production.