Chapter I. Introduction to Mendelian Genetics

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Outlines of the Chapter I

- Introduction
- Concept of gene
- Concepts of genotype and phenotype
- Concepts of dominance and recessiveness of traits
- Mendel's laws

Objectives of this chapter

1. **Understand** the Fundamental Concepts of Genetics ;
2. **Learn** Mendel’s Laws of Inheritance;
3. **Analyze** Mendel’s Experimental Results and **apply** them in resolving problems.

1. Introduction

Genetics is the study of heredity. Johann Gregor Mendel (1822–1884) set the framework for genetics long before chromosomes or genes had been identified, at a time when meiosis was not well understood. Mendel selected a simple biological system and conducted methodical, quantitative analyses using large sample sizes. Because of Mendel's work, the fundamental principles of heredity were revealed. We now know that genes, carried on chromosomes, are the basic functional units of heredity with the capability to be replicated, expressed, or mutated. Today, the postulates put forth by Mendel form the basis of classical, or Mendelian, genetics. Not all genes are transmitted from parents to offspring according to Mendelian genetics, but Mendel's experiments serve as an excellent starting point for thinking about inheritance.

2. Concept of Gene

2.1.Definition: A gene is a unit of hereditary information; it is a segment of DNA that occupies a fixed position (locus) on a chromosome. A gene exerts its effects by directing the synthesis of proteins.

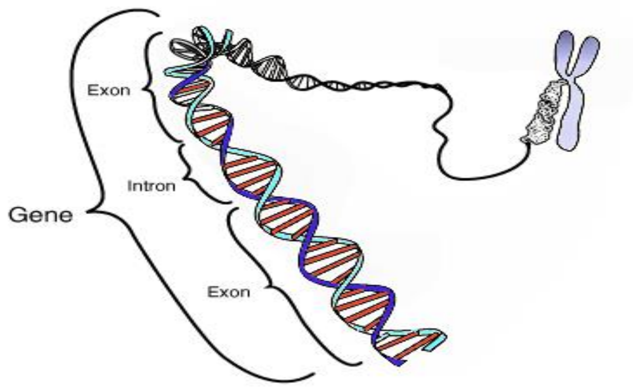


Fig.1: Structure of a gene.

2.2.Related definitions

- ✓ **Alleles:** one alternative form of a given gene pair. E.g. white and purple color of pea flowers.
- ✓ **Chromosomes**

Genes are organized and packaged within the cell in structures called "chromosomes." A chromosome is a strand of packaged DNA found in the nucleus of cells and particular cellular organelles that contain noncoding and coding sequences (chloroplasts and mitochondria).

- ✓ **Genetic Code**

Each gene is composed of a series of letters adenine (A), thymine (T), guanine (G) and cytosine (C), which correspond to the different bases in the DNA, the sequence of which forms the genetic code. The genetic code is universal among most organisms and consists of codons, which are groups of three nucleotides that correspond to specific amino acids or signals in the protein synthesis process.

- ✓ **DNA (deoxyribonucleic acid)**

DNA is a molecule found in the nucleus of every cell that holds the template for how **living organisms are built and all the proteins we need to make to be healthy.** It is made up of chemical building blocks and the order, or sequence, of these determines what **biological instructions** are contained in a strand of DNA.

- ✓ **RNA**

RNA stands for ribonucleic acid. It is an important molecule with long chains of nucleotides. The main function of RNA is to carry information from the genes to where **proteins** are assembled on ribosomes in the cytoplasm. This is done by messenger RNA (mRNA).

- ✓ **Proteins**

Proteins are large, complex molecules that play many critical roles in the body. They do most of the work in cells and are required for the structure, function, and regulation of the body's tissues and organs.

Proteins are made up of hundreds or thousands of smaller units called amino acids, which are attached to one another in long chains. There are 20 different types of amino acids that can be combined to make a protein. The sequence of amino acids determines each protein's unique 3-dimensional structure and its specific function.

3. Concepts of genotype and phenotype

3.1.Genotype: it is the genetic makeup or constitution of an individual with reference to the character under consideration. It is typically represented by symbols (e.g., TT, Tt) to denote different alleles. The genotype encompasses the set of genetic potentialities of a given cell or organism, specifically in relation to the trait being examined.

3.2.Phenotype: Phenotype is the physical expression of the genotype. It is the external appearance of an individual for a particular gene. Phenotype can also be affected by the environment (e.g., height influenced by both genotype and nutrition).

Example: consider two alleles of flower color: purple and white. In this example, purple is a dominant trait and the genotypes BB (homozygous dominant) and Bb (heterozygous) code for purple. The genotype bb (homozygous recessive) does not produce the protein that makes the purple color, so this genotype codes for a white flower. The flower colors are the phenotype.

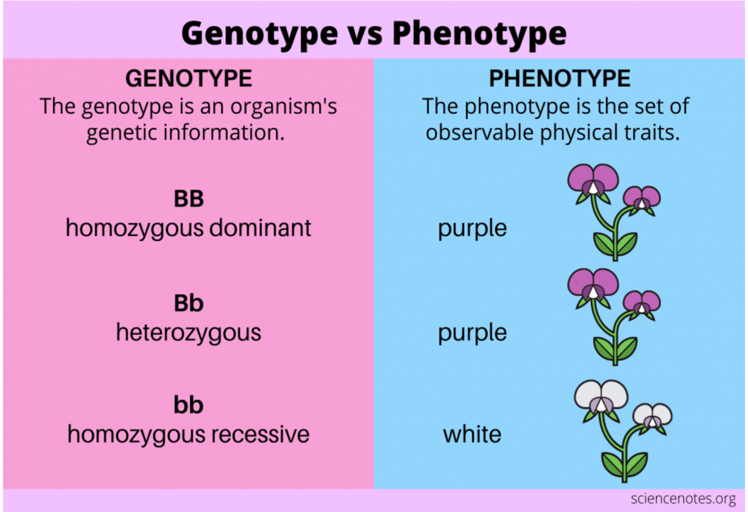


Fig.2: Difference between genotype and phenotype.

4. Concepts of dominance and recessiveness

The terms dominant and recessive refer to the genotypic interaction of alleles in producing the phenotype of the heterozygote.

- ✓ **Dominant**: is a relationship between alleles of a gene, in which one allele masks the expression (phenotype) of another allele at the same locus.
- ✓ **Excessive:** means that the recessive allele can to be covered up by a dominant trait.
- ✓ **Heterozygous (heterozygote)** : an individual in which the 2 alleles of a particular gene are non-identical
- ✓ **Homozygous (homozygote)** : an individual in which the 2 alleles of a particular gene are identical
- ✓ **Gamete**: a sex cell having haploid set of chromosomes and produced by meiotic cell division of a diploid.
- ✓ **Filial**: a generation obtained by a given cross (F1= generation given by the 1st cross, F2= generation given by the 2nd cross ….)

Important points:

- Dominant alleles are expressed exclusively in a heterozygote, while recessive traits are expressed only if the organism is homozygous for the recessive allele.
- A single allele may be dominant over one allele, but recessive to another.
- Not all traits are controlled by simple dominance as a form of inheritance; more complex forms of inheritance have been found to exist.

5. Mendel's laws

Mendel developed the principles of **segregation**, **independent assortment**, and **dominance** based on his studies of seven traits in peas (figure 3), including flower color, pea color, and pea shape. The pea traits that Mendel focused on were determined by single genes, each of which had 2 alternative alleles, 1 fully dominant and 1 fully recessive.

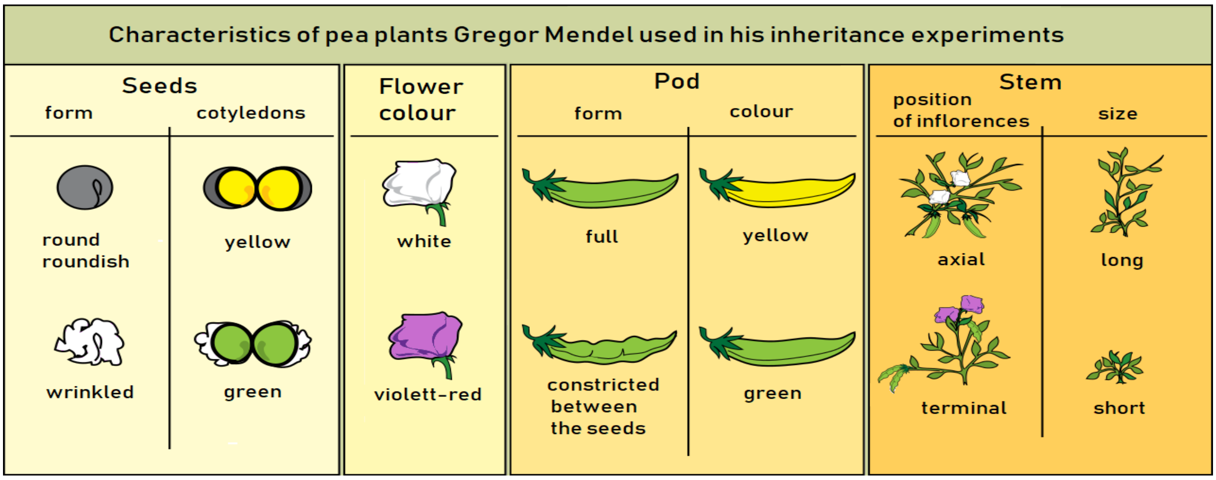
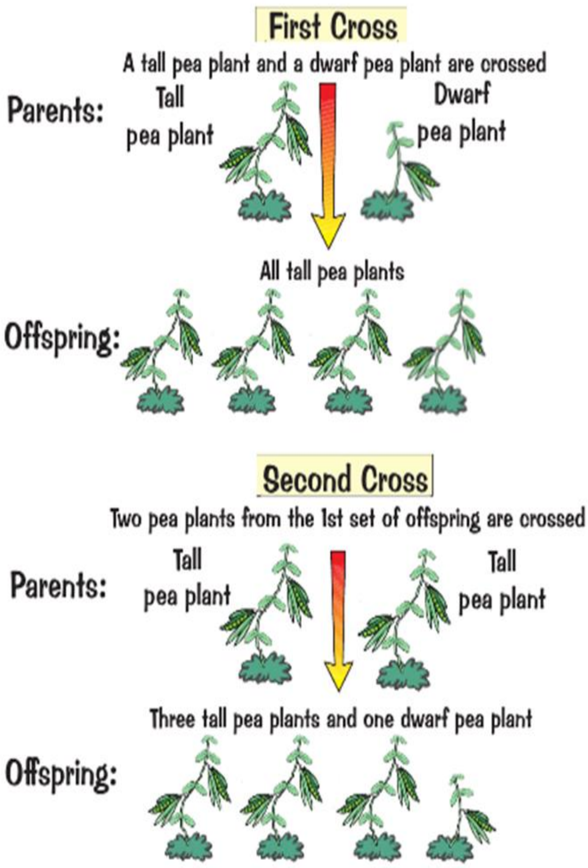


Fig.3. the seven traits that Mendel studied in his experiments.

5.1.Mendel’s experiments :

Plants used in first-generation crosses were called **P**, or parental generation, plants. Mendel collected the seeds that resulted from each cross and grew them the following season. These offspring were called the **F1**, or the first **filial** (filial = offspring, daughter or son), generation.



Once Mendel examined the characteristics in the F1 generation of plants, he allowed them to self-fertilize. He then collected and grew the seeds from the F1 plants to produce the F2, or second filial, generation. Mendel’s experiments extended beyond the F2 generation to the F3 and F4 generations, and so on, but it was the ratio of characteristics in the P−F1−F2 generations that were the most intriguing and became the basis for Mendel’s principles.

5.2.Mendel's first law "Law of segregation "

"This principle states that the two coexisting alleles of an individual for each trait segregate during gamete formation so that each gamete gets only one of two alleles. Alleles unite again at random fertilization of gametes".

- **Experimental basis**:

In Mendel's experiments with flower color, "**R**" represents the dominant allele for purple flowers, and "**r**" the recessive allele for white flowers. In the F1 generation, crossing a homozygous purple (**RR**) and a homozygous white (**rr**) plant produces heterozygous (**Rr**) offspring, **all** displaying purple flowers, but their genotype contains both **R** and **r** genes, and these alternative genes do not blend or contaminate each other. Mendel inferred that, when a heterozygote forms its sex cells, the allelic genes **segregate** and pass to different gametes.

Now, if the gametes unite at random, then the F2 generation should contain about **1/4 white-flowered** and **3/4 purple-flowered** plants. The white-flowered plants, which must be **recessive homozygotes**, with the genotype **rr**. About **1/3** of the plants exhibiting the dominant trait of purple flowers must be **dominant homozygotes**, **RR**, and **2/3 heterozygotes**, **Rr**. The prediction is tested by obtaining a third generation, F3, from the purple-flowered plants; though phenotypically all purple-flowered, 2/3 of this group of plants reveal the presence of the recessive gene allele, r, in their genotype by producing about 1/4 white-flowered plants in the F3 generation.

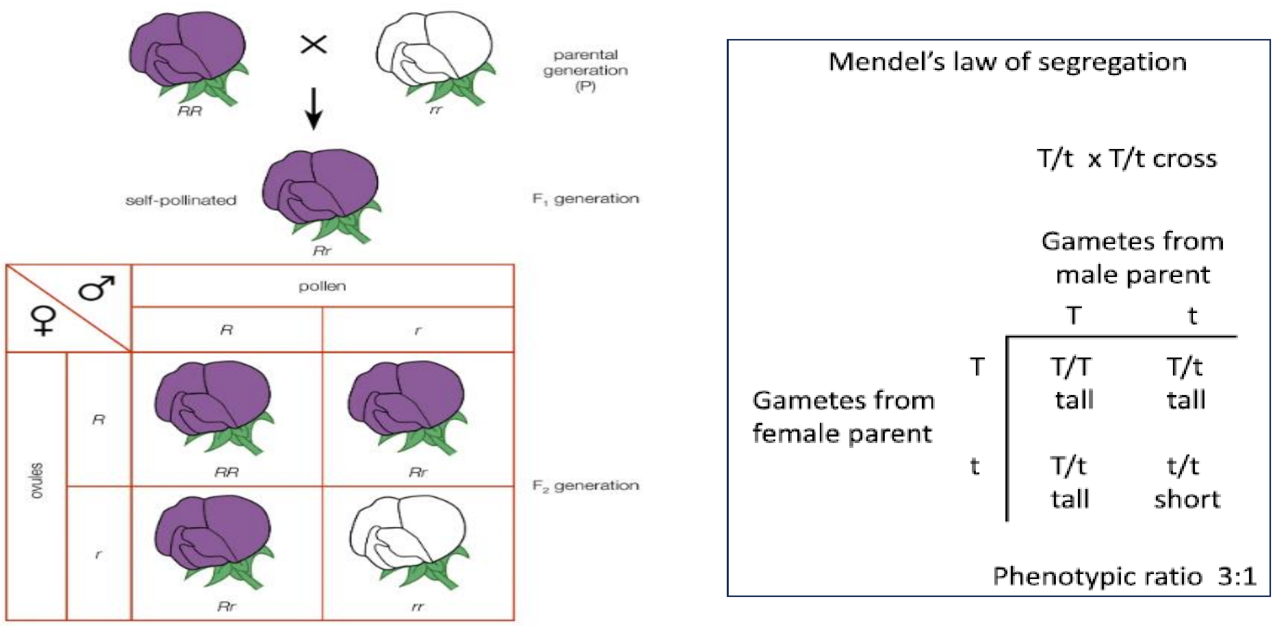


Fig.5. Phenotypic Ratios According to Mendel's Law of Segregation.

5.3.Law of independent assortment

“The law states that alleles of two or more different genes separate (segregate) independently from the other into gametes that reproduce sexually. Thus, the allele received from one gene does not influence the allele received from the other. What it means: different genes are inherited separately. For example, the gene which codes for flowers color is inherited separately from the gene which codes for grain shape.

- **Experimental basis**:

Mendel crossed two pure-breeding pea plants in his experiment: one with yellow, round seeds and one with green, wrinkled seeds (yyrr). By the law of segregation, the gametes made by the round, yellow plant are all RY, and the gametes made by the wrinkled, green plant all are ry. Thus, all the F1 offspring are RrYy that are phenotypically identical, producing yellow and round seeds.

In the F2 generation, Mendel self-pollinated all F1 plants. He found four categories of pea seeds: yellow and round, yellow and wrinkled, green and round, and green and wrinkled. They were found to be phenotypically in the ratio of 9:3:3:1.

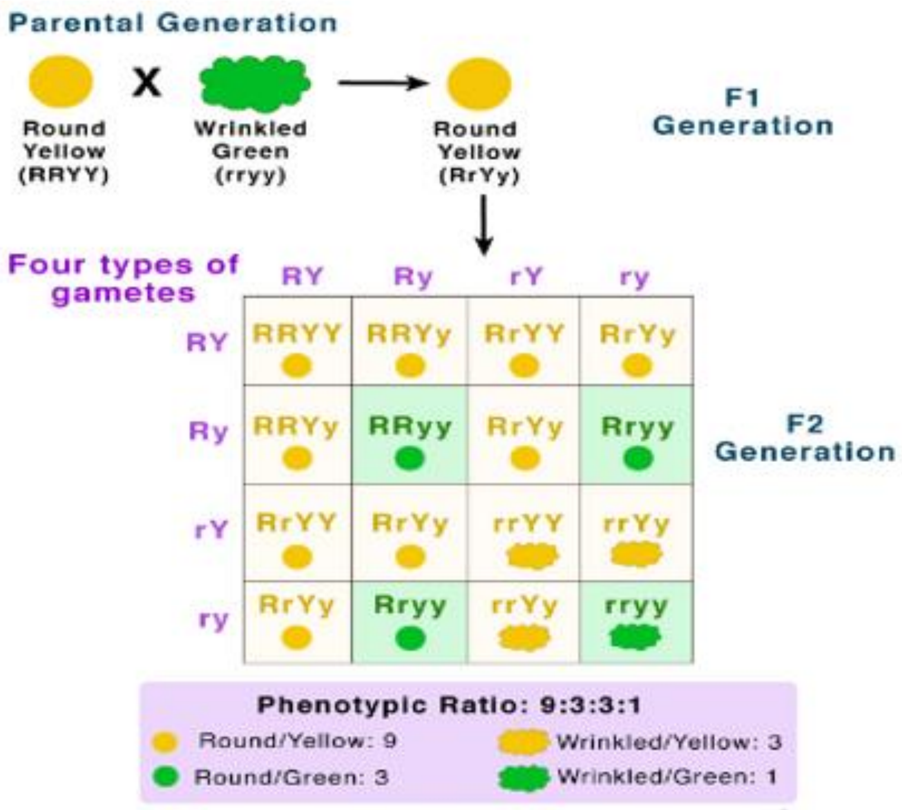


Fig.5. Phenotypic Ratios According to Mendel’s Law of independent assortment.

5.4.Law of dominance

The principle of dominance states that in a heterozygote, only the dominant allele will be expressed. The recessive allele will remain “latent” but will be transmitted to offspring by the same manner in which the dominant allele is transmitted. The recessive trait will only be expressed by offspring that have two copies of this allele. Individuals with a dominant trait could have either two dominant versions of the trait or one dominant and one recessive version of the trait. Individuals with a recessive trait have two recessive alleles.