HUMAN GENETICS

Topic Covered

Mendel's Principles

Objectives

Overview of Mendelian inheritance and Mendelian disease, Definitions of important terms in genetics with support lesson (Examples of Genotype & Phenotype) besides a discussion about the Dominate and recessive traits and the Hybrid.

Content

The course is given in English based on Explaining and Discussing in details for all course lectures

Teaching and learning methods

The course includes theoretical parts support with PowerPoint lectures

Assessments

Weekly quizzes , hand writing reports in each course , first and second course exam, mid exam and final exam

Source book

Human Genetics Book Tenth Edition by **Lewis** **McGraw-Hill International Edition**

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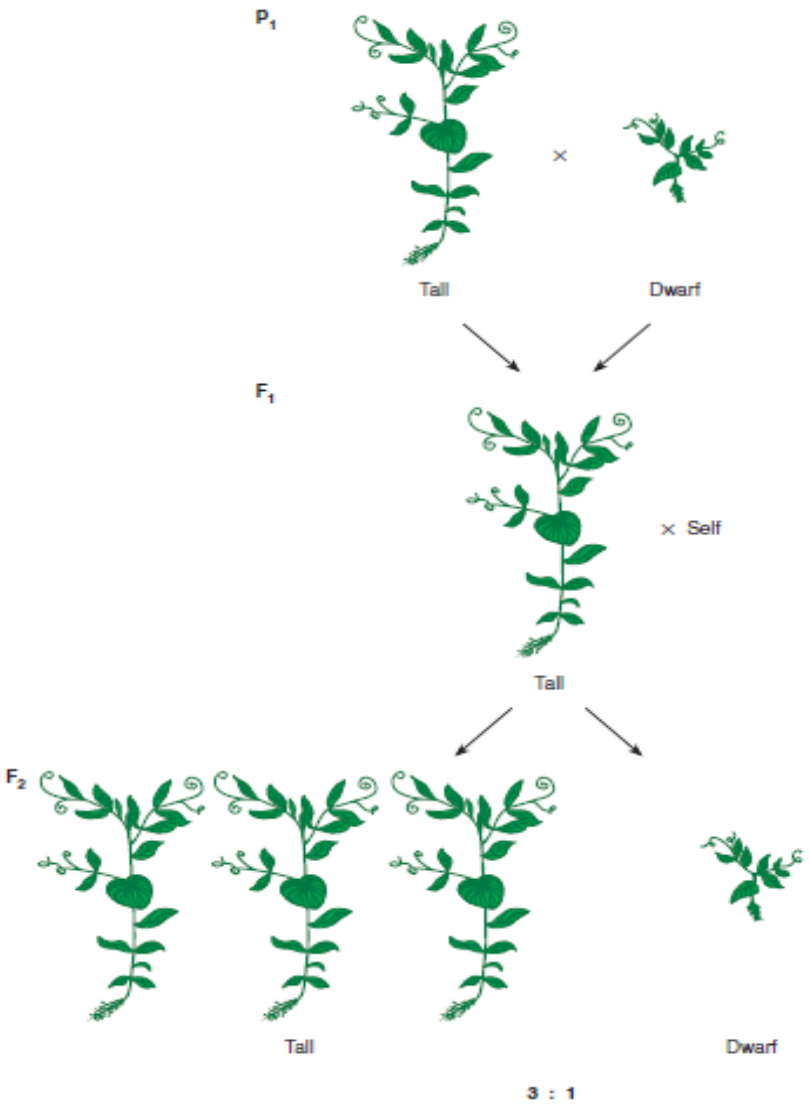
Mendel's Principles

Genetics is concerned with the transmission, expression, and evolution of genes, the molecules that control the function, development, and ultimate appearance of individuals.

In this part, we will look at the rules of transmission that govern genes and affect their passage from one generation to the next. Gregor Johann Mendel discovered these rules of inheritance.



What are the following terms?

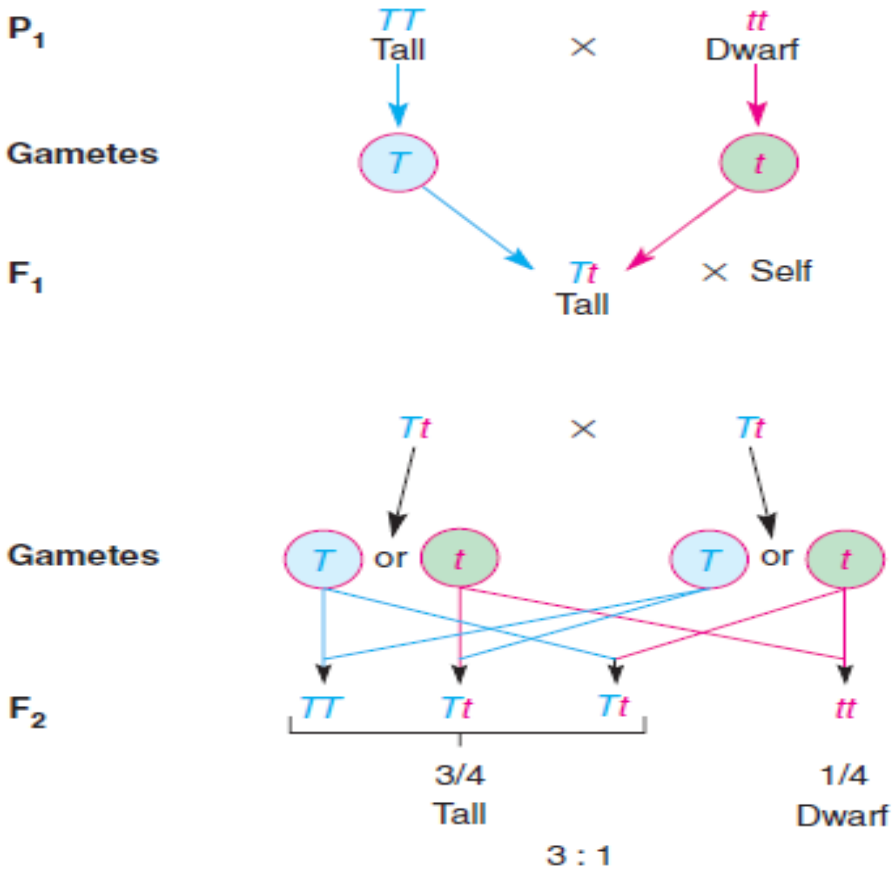
1. Self-Fertilization:
2. Cross-Fertilization:
3. Filial Generation:
4. Hybrids:
5. Dominant Trait:
6. Recessive Trait:
7. Gene:
8. Allele:
9. Gamete:
10. Zygote:
11. Homozygote:
12. Heterozygote:

1) Rule of Segregation:

Mendel's first principle, the **rule of segregation.** The rule of segregation can be summarized as follows: A gamete receives only one allele from the pair of alleles an organism possesses; fertilization (the union of two gametes) reestablishes the double number.

Mendel used capital letters to denote alleles that control dominant traits and lowercase letters for alleles that control recessive traits. Following this notation, *T* refers to the allele controlling tallness and *t* refers to the allele controlling shortness (Dwarf stature).

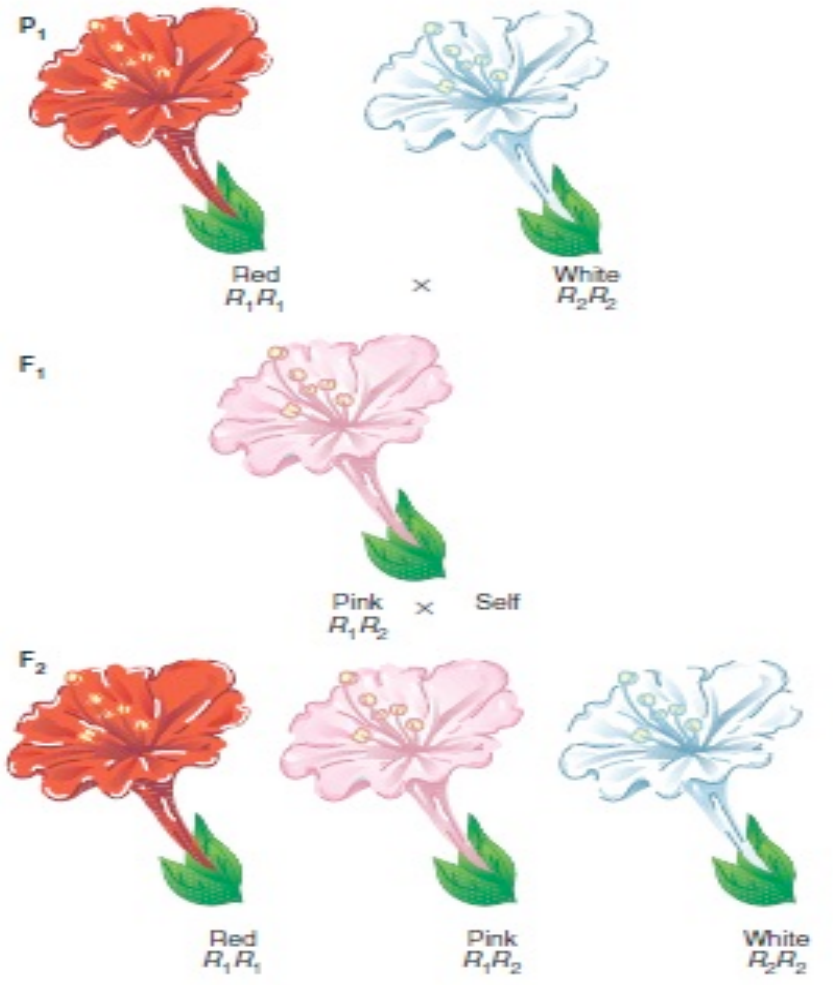
The **genotype** of an organism is the gene combination it possesses, the genotype of the parental tall plant is *TT;* that of the F1 tall plant is *Tt.* **Phenotype** refers to the observable attributes of an organism. Plants with either of the two genotypes *TT* or *Tt* are phenotypically tall. Genotypes come in two general classes: **homozygotes,** in which both alleles are the same, as in *TT* or *tt,* and **heterozygotes,** in which the two alleles are different, as in *Tt.*



DOMINANCE IS NOT UNIVERSAL

If dominance were universal, the heterozygote would always have the same phenotype as the dominant homozygote, and we would always see the 3:1 ratio when heterozygotes are crossed. If, however, the heterozygote were distinctly different from both homozygotes, we would see a 1:2:1 ratio of phenotypes when heterozygotes are crossed. In **partial dominance (**or **incomplete dominance),** the phenotype of the heterozygote falls between those of the two homozygotes.

An example occurs in flower petal color in some plants by cross a plant that has red flower petals with another that has white flower petals; the offspring will have pink flower petals. If these pink-flowered F1 plants are crossed, the F2 plants appear in a ratio of 1:2:1, having red, pink, or white flower petals, respectively.


P1
Red
R1R1
×
White
R2R2
F1
Pink
R1R2
×
Self
F2
Red
R1R1
Pink
R1R2
White
R2R2

The pink-flowered plants are heterozygotes that have a petal color intermediate between the red and white colors of the homozygotes. In this case, one allele (*R*1) specifies red pigment color, and another allele specifies white color (*R*2). Flowers in heterozygotes (*R*1*R*2) have about half the red pigment and white pigment.

The other category in which the heterozygote is not related to Mendel's Law occurs when the heterozygous phenotype is not on a scale somewhere between the two homozygotes, but actually expresses both phenotypes simultaneously or called **codominance.**

For example, people with blood type AB are heterozygotes who express both the *A* and *B* alleles for blood type).

MULTIPLE ALLELES

A given gene can have more than two alleles. The classic example of multiple human alleles is in the ABO blood group. There are four blood-type phenotypes produced by three alleles.

Blood Type Corresponding to Antigens on Red Blood Cells	Antibodies in Serum	Genotype	Reaction of Red Cells to Anti-A Antibodies	Reaction of Red Cells to Anti-B Antibodies
O	Anti-A and anti-B	*ii*	–	–
A	Anti-B	I^AI^A or I^Ai	+	–
B	Anti-A	I^BI^B or I^Bi	–	+
AB	None	I^AI^B	+	+

The I^A and I^B alleles are responsible for the production of the A and B antigens found on the surface of the erythrocytes (red blood cells).

The ABO system is unusual because antibodies can be present (e.g., anti-B antibodies can exist in a type A person) without prior exposure to the antigen.

Thus, people with a particular ABO antigen on their red cells will have in their serum the antibody against the other antigen: type A persons have A antigen on their red cells and anti-B antibody in their serum; type B persons have B antigen on their red cells and anti-A antibody in their serum; type O persons do not have either antigen but have both antibodies in their serum; and type AB persons have both A and B antigens and form neither anti-A or anti-B antibodies in their serum.

Since both *I*A and *I*B are dominant to the *i* allele, this system not only shows multiple allelism, it also demonstrates both codominance and simple dominance.

2) Rule of Independent Assortment:

The Law of Independent Assortment, also known as "Inheritance Law", states that separate genes for separate traits are passed independently of one another from

parents to offspring. More precisely, the law states that alleles for one gene can segregate independently of alleles for other genes. While Mendel's experiments with mixing one trait always resulted in a 3:1 ratio between dominant and recessive phenotypes, his experiments with mixing two traits (dihybrid cross) showed 9:3:3:1 ratios. Mendel concluded that different traits are inherited independently of each other, so that there is no relation, for example: the dominant phenotypic class, with round, yellow seeds, represents four genotypes: *RRYY, RRYy, RrYY,* and *RrYy.*