وزارة التعليم العالي و البحث العلمي
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المرحلة الأولى

المحاضرة الثالثة

٢٠١٩ - ٢٠٢٠

Mendelian Inheritance Concepts:

- **Inheritance** is controlled **by information stored** in discrete factors called **genes.**
- **Genes** are transmitted from generation to generation **on vehicles** called **chromosomes.**
- **Chromosomes,** which **exist in pairs** in **diploid** organisms, provide the basis of biparental inheritance.
- **During gamete formation,** chromosomes are distributed according to postulates first described by **Gregor Mendel,** based on his nineteenth-century research with the garden pea.
- **Mendelian postulates** prescribe that **homologous chromosomes segregate** from one another and **assort independently** with other segregating homologus during gamete formation.
- **Genetic ratios,** expressed as probabilities, are subject to chance deviation and may be evaluated statistically.

Mendel's first law: Mendel's theory of segregation

Mendel had an idea that in every generation, a plant inherits two factors (now called genes) of information for a trait, one from each parent. To test this idea, he performed what we now call a monohybrid cross. He tracked many individual's traits through two generations. For instance, he crossed *true- breeding purple –flowered plants and true-breeding white- flowered plants. All the plants grown were of purple flowers (they are heterozygous for the studied trait which is what monohybrid means). When Mendel allowed these plant to self-fertilize, some plant grown were of white flowers.

Mendel hypothesized that each plant had inherited two units of information about flower color, one from each parent and the unit for purple had to be dominant because it had masked unit for white in F1 plants.

Germ cells of pea plants are diploid which pairs of homologous chromosomes. If we assume that one plant is homozygous dominant (AA) and the other is homozygous recessive (aa) for flower color, then after meiosis a sperm (pollen grain) or egg carries one allele for flower color. Thus, when a sperm fertilizes an egg, only one outcome is possible: A+a = Aa

*True-breeding plant is a plant when self fertilized produces offspring with the same traits. The alleles of such plant are homozygous.

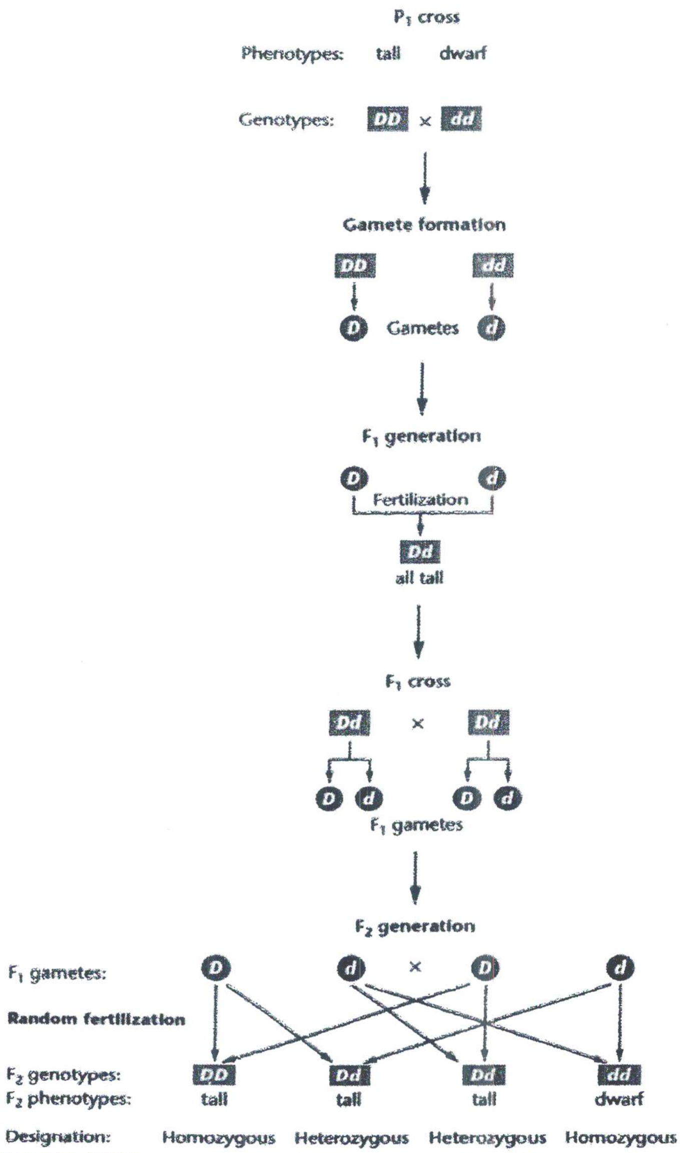
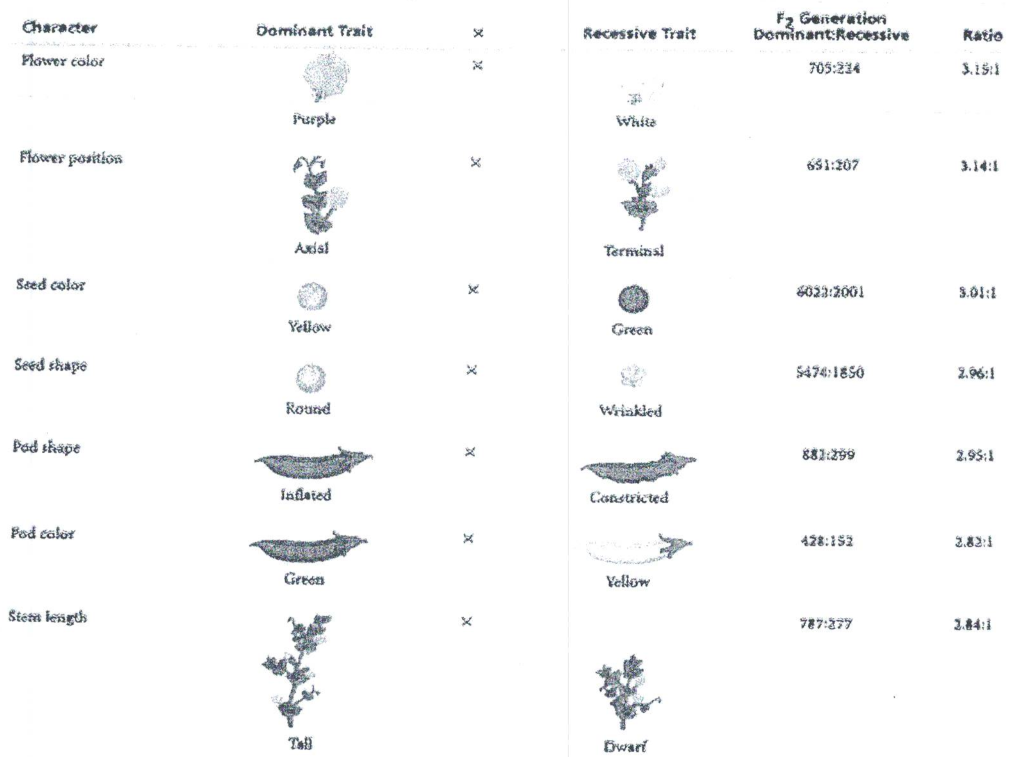


FIGURE 1.2 The monohybrid cross between tall (*D*) and dwarf (*d*) pea plants. Individuals are shown in rectangles, and gametes are shown in circles.

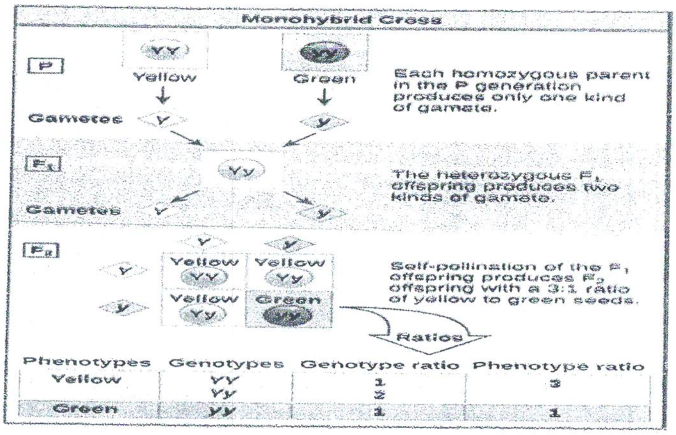
Mendel crossed hundreds of plants and tracked thousands offspring and counted and recorded the number of plants showing dominance or recessiveness for many traits before he concluded his hypothesis. An interesting ratio emerged on average (three of every four F2 plants had the dominant phenotype and one had the recessive phenotype. F2 plants resulted from the self-fertilization of F1 plants. To visualize what happened when the heterozygous F1 plants undergo self-fertilization, it was depend on punnett square which provides a simple method for tracking the kinds of gametes produced as well as all the possible combinations that might occur at fertilization.



Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant:Recessive	Ratio
Flower color	Purple	×	White	705:224	3.15:1
Flower position	Axial	×	Terminal	651:207	3.14:1
Seed color	Yellow	×	Green	6022:2001	3.01:1
Seed shape	Round	×	Wrinkled	5474:1850	2.96:1
Pod shape	Inflated	×	Constricted	882:299	2.95:1
Pod color	Green	×	Yellow	428:152	2.82:1
Stem length	Tall	×	Dwarf	787:277	2.84:1

As the **punnett square** shows, each **hybrid** produces two kinds of gametes (**Y and y**) in a ratio of **1:1**, thus half of pollens and half of the eggs carry (Y) and other half carry (y). At fertilization ¼ **of the progeny will be (YY), ¼ (Yy), ¼ (yY) and ¼ (yy)**. The source of gametes of alleles for Mendel's traits (whether from egg and pollen) had no influence on the effect of alleles. So, (Yy) and (yY) are the same. This mean that ½ **of the progeny are (Yy) yellow hybrids, ¼ (YY) true breeding yellows** and ¼ **(yy) true- breeding greens.** Also the punnett square shows how the segregation of alleles during gamete formation and random union of egg and pollen at fertilization can produces 3:1 ratio of yellow to green seeds that Mendel observed in F2 generation.

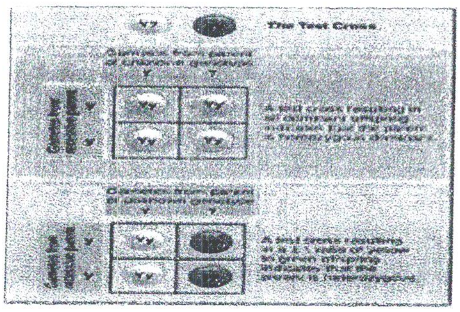
This ratio made Mendel suggests that fertilization is a chance event with a number of possible outcomes and probabilities. So by understanding the rules of probability, he performed a large number of crosses and predicted the outcomes of the crosses.



Test Cross

An individual shows dominance for a specific trait has phenotype of dominant trait but its genotype is unknown because it might be a homozygous dominant (YY) or a heterozygous dominant (Yy). To determine whether the individual is a homozygote or a heterozygote, Mendel tested his F1 offspring by crossing them with a known homozygous recessive individual.

- If they were all homozygous dominant, then all F2 offspring would show the dominant form of the trait.
- If they were all heterozygous, F2 offspring would show as many dominant individuals as recessive ones. Half of the plants showed yellow seeds (Yy) and the other half had green seeds (yy).

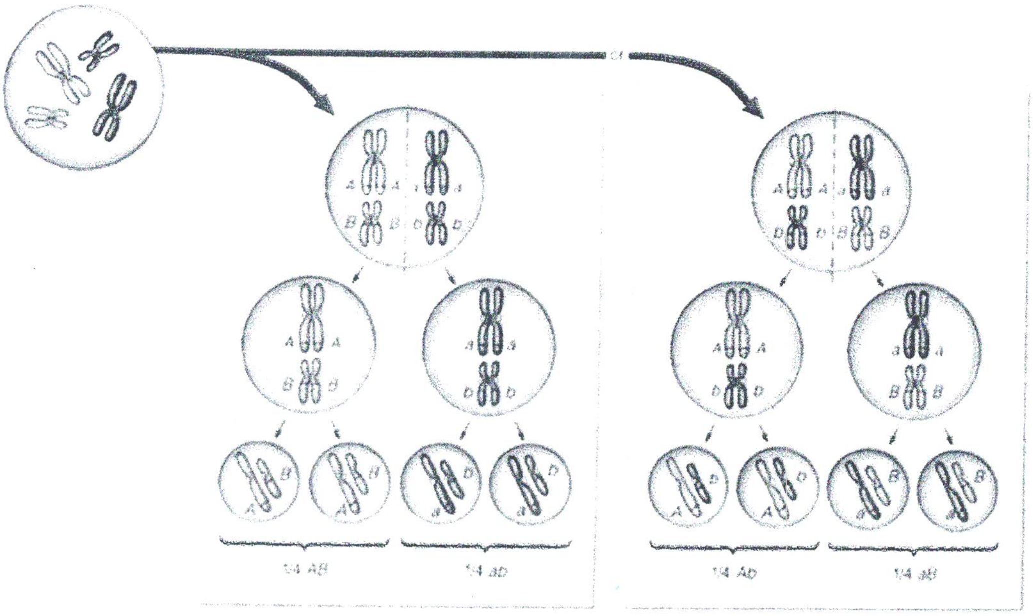


The result of Mendel's monohybrid crosses and test crosses became the basis of the theory of segregation.

The theory of segregation states that diploid cells have pairs of genes on pairs of homologous chromosomes. During meioses, the two genes of each pair are segregate from each other. As a result, they end up in different gametes.

Law of independent assortment (the second law)

Having determined from monohybrid crosses that genes are inherited according to the law of segregation, Mendel attempted to explain how two pairs of genes might be assorted into gametes. He selected true breeding plants that differed in two traits, for example, flower color and hight or seed color and shape. He mate true-breeding plants grown from yellow round seeds (YYRR) with true-breeding plants grown from green wrinkled seeds (yyrr) and as Mendel predicted, F1generation from this cross was all with yellow round seeds (YyRr) showing only the two dominant phenotypes: yellow and round. Now, when F1 plants mature and reproduce by self-fertilization, how will the two pairs of genes be assorted into gametes? The answer partly depends on the location of the chromosomes that carry the gene pairs. The following diagram shows the chances of allele's combinations in the gametes according to the possible alignments of the homologous chromosomes at metaphase I & II of meiosis. This diagram shows that after meiosis and gamete formation, four combinations of alleles are possible in the sperms (pollen grains) or eggs: ¼ AB, ¼ Ab, ¼ aB and ¼ ab. So during fertilization **4** different kinds of eggs can combine with any of **4** different kinds of pollen producing a total of **16** possible zygotes. Once again the punnett square is a appropriate way to visualize the process. When Mendel examined the F2 generation, he noticed that new recombination types had been arised like yellow wrinkle and green round seeds.



The example above shows that the results were as follow: **9** yellow round, **3** green round, **3** yellow wrinkled and **1** green wrinkled. However if we look at just pea color or shape, we can see that each trait is inherited in the **3:1** ratio predicated by Mendel's law of segregation. There are **12** yellows for every **4** greens and **12** rounds for every **4** wrinkles. In other words, the ratio of each dominant trait (yellow or round) to its antagonistic recessive trait (green or wrinkled) is **12:4** or **3:1** which means that the inheritance of each gene is not affected by the inheritance of the other gene. This analysis became the basis of Mendel's second law, **the law of independent assortment** which states that during gamete formation, different pairs of alleles segregate independently of each other.

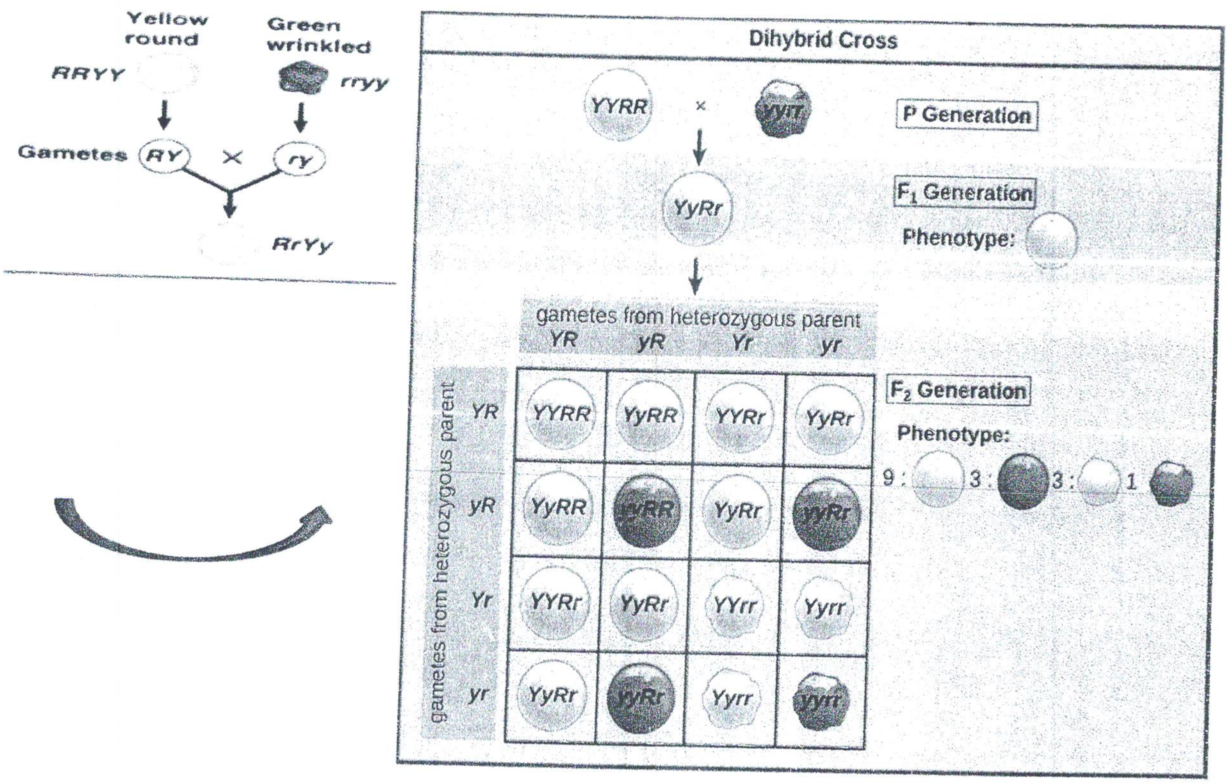
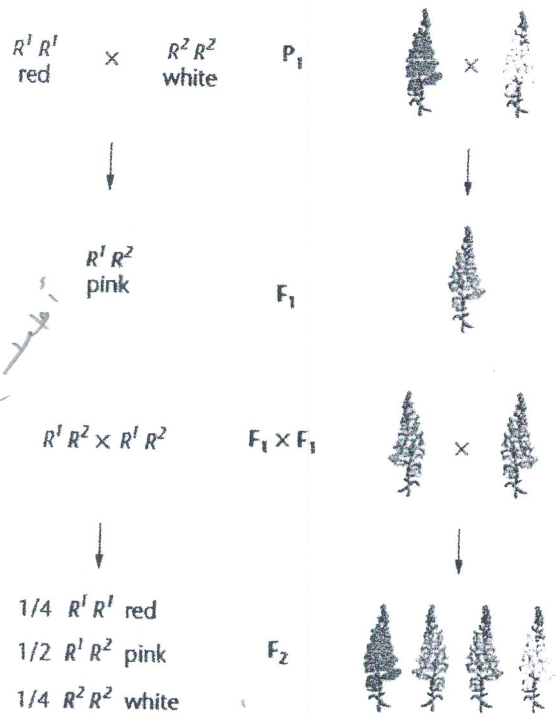


Figure A dihybrid cross produces parental types and recombinant types: pure-breeding parents (P) produce uniform generation of F1 dihybrids; Self-pollination of F1 yields the characteristic F2 phenotypic ratio of 9:3:3:1

Non Mendelian inherited

1- Incomplete dominance

For some characteristics, The F1 hybrids have an appearance in between the phenotypes of the two parental varieties, this inherited pattern called incomplete dominance in which one alleles of a pair isn't fully dominant over its partner, so neither parental allele is dominant or recessive to the other and expression of both alleles is observed as intermediate phenotype in heterozygous individual. In a cross between a true breeding red flower snapdragon plant with a true breeding white one, all F1 plants are pink. The explanation of this result is that the two red alleles in the red plant produce a double dose of the red-producing enzyme which generates enough pigment to make the flower look fully red. In the heterozygous form (pink) only one copy of the red allele results in only enough pigment to make the flower look pink. In the white flowers, the alleles don't produce any functional enzyme (no red pigment) and the flowers appear white. In this example three genotypes R1R1, R1R2 and R2R2 have three distinct phenotypes.



2- Codominance

In this case, the heterozygous phenotype exhibits the phenotype of both parents' traits together which means that non of the two alleles that the parents carry are dominant or recessive to the other. In human, some of the complex molecules that are fixed in the red blood cells membrane show codominance. For example, one gene (I) with alleles (I^A) and (I^B) control the presence of a sugar polymer that extends from the red blood cell membrane. Each allele encodes for an enzyme that causes the production a slightly different form of the complex sugar. In heterozygous individuals, the red blood cells carry both the (I^A) and (I^B) determined sugar on their surface, where as the cell of homozygous individuals show the products of either (I^A) or (I^B) alone. So the person with both (I^A) allele and (I^B) exhibits a codominant case because it's red blood cells have both kinds of sugar at their surface.

Genotype	Antigen	Phenotype
$I^A I^A$	A	A
$I^A i$	A	
$I^B I^B$	B	B
$I^B i$	B	
$I^A I^B$	A, B	AB
$i\ i$	Neither	O