LESSON 5

PATTERNS OF INHERITANCE. MENDEL'S LAWS

Basic concepts and terminology of modern genetics

allele - an alternative form of a specific gene.

backcross refers to a cross of F1 hybrids to individuals that have genotypes of the parental generation.

cross-fertilization has same meaning as *cross.* It requires that the male and female gametes come from separate individuals.

dihybrid cross - a cross in which an experimenter follows the outcome of two different traits.

dominant - an allele that determines the phenotype in the heterozygous condition

genotype - the genetic composition of an individual, especially in terms of the alleles for particular genes.

heterozygote - an individual who is heterozygous.

homozygous describes a diploid individual that has two identical alleles of a particular gene.

hybrid - an offspring obtained from a hybridization experiment; (2) a cell produced from a cell fusion experiment in which the two separate nuclei have fused to make a single nucleus.

independent assortment - the random distribution of alleles to the gametes that occurs when genes are located in different chromosomes

monohybrid - an individual produced from a monohybrid cross.

monohybrid cross - a cross in which an experimenter is following the outcome of only a single trait.

phenotype - the observable traits of an organism.

Punnett square - a diagrammatic method in which the gametes that two parents can produce are aligned next to a square grid as a way to predict the types of offspring the parents will produce and in what proportions.

recessive - a trait or gene that is masked by the presence of a dominant trait or gene.

self-fertilization – **a** fertilization that involves the union of male and female gametes derived from the same parent.

testcross - an experimental cross between a recessive individual and an individual whose genotype the experimenter wishes to determine.

trait - any characteristic that an organism displays (morphological, physiological, behavioral traits).

Mendel's Experiments

Gregor Johann Mendel, born in 1822, is now remembered as the father of genetics. In 1856, Mendel began his historic studies on pea plants (Pisum sativum). In the pea plants, Mendel found certain clear advantages such as: pea plant has a few contrasting characters (yellow-green seed color, round-wrinkled seed shape, purple-white flower color etc.); under natural conditions the pea plants exhibited only self-pollination; the duration of life cycle in the pea plants was very short; every pea plant produced a large number of seeds.

For 8 years, he grew and crossed thousands of pea plants. Mendel made two innovations: *he developed pure lines and counted his results and kept statistical notes.*

Mendel's analysis of monohybrid crosses

Mendel began his experiments with true-breeding pea plants that varied with regard to only one of seven different contrasting characters.

In the first set of experiments, Mendel conducted cross-pollination between a pure breeding pea plants with round seeds (RR) and pea plants with wrinkled seeds (rr). All seeds in F1 generation were found to be round (Rr). Similar results were obtained with reference to all the pairs of contrasting characters (fig.19).

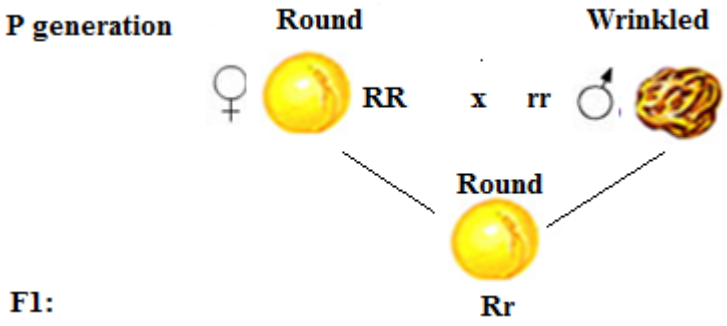

Fig. 19. The first Mendel's experiment

Based on these results, Mendel came to the conclusion that in a cross-involving two contrasting characters, only one character expresses itself in the next generation. Mendel

called the character, which expressed as ***dominant*** character and the character, which failed to express, as ***recessive*** character. This idea came to be known as the ***Law of dominance***.

At this stage, Mendel wanted to know whether the round seeds resulting from a cross between round and wrinkled seeds, were similar to the round seeds of the P_1 generation. Hence, he allowed the round seeds of the F_1 generation to undergo self-pollination. In the next generation, Mendel found both round seeds and wrinkled seeds, approximately in the ratio 3:1. The results were most surprising since the recessive character of wrinkled seeds had reappeared in the next (F2) generation (fig.20).

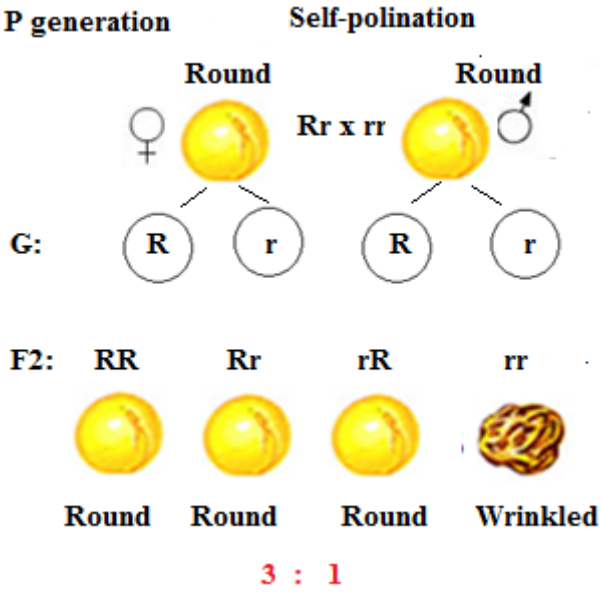

Fig.20. The second Mendel's experiment

From the results, it was clear that the round seeds of the F1 generation were different genetically from the round seeds of the P1 generation. Similar results were obtained by Mendel for the other contrasting characters also. Based on these results, Mendel came to the conclusion that certain factors are involved in the expression of each of these contrasting characters. He presumed that if the F1 round seeds on selfpollination could give rise to both round and wrinkled seeds, this plant should have contained two factors, one responsible for round and the other responsible for wrinkled seeds. Similarly, he presumed that the P1 also round seeds should have contained two factors, both responsible for round shape.

Based on these results Mendel arrived at another conclusion, which is known as the second law of inheritance or ***Law of segregation*** (or the ***Law of purity of gametes***). The law states that: In a cross involving a pair of contrasting characters, the factors responsible for

the two opposite characters stay together in the F1 generation but segregate (separate) during the formation of gametes.

Using the modern notion of genes Mendel's law of segregation states that: *The two copies of a gene segregate (or separate) from each other during transmission from parent to offspring.*

Mendel's analysis of dihybrid crosses

Mendel decided to examine the inheritance of two characteristics at once and he tested this idea. Based on the concept of segregation, he predicted that traits must sort into gametes separately. First, he generated plants that were purebred for two characteristics, such as seed color (yellow and green) and seed shape (round and wrinkled). Mendel crossed the plants with wrinkled and yellow seeds (*rrYY*) with plants with round, green seeds (*RRyy*). Then F1 dihybrids (*RrYy*) are allowed to self-fertilize. Mendel's results from this cross (fig.21):

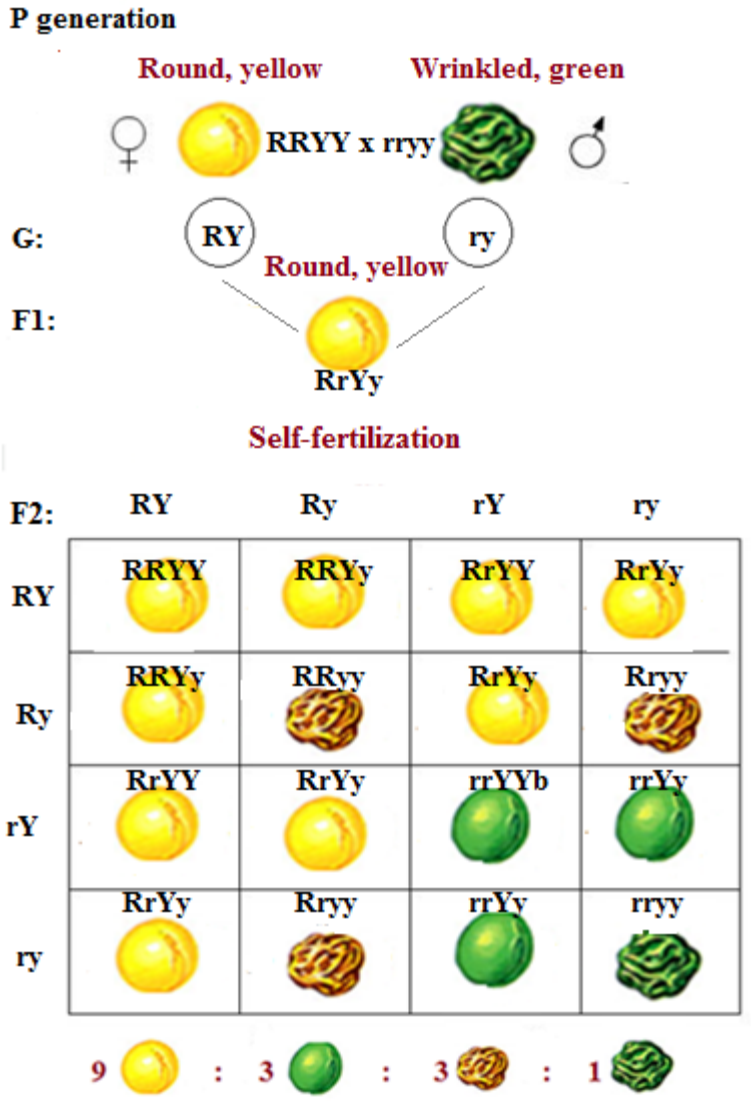

Fig. 21. The third Mendel's experiment

In the crossing between two parents that are heterozygous (*RrYy* × *RrYy*) each from them gives four types of gametes: *RY, Ry, rY,* and *ry*. These four types of gametes are the result of the independent assortment of the seed shape and seed color alleles relative to each other. During fertilization, gametes of opposite parents can combine between each other resulting in 16 types of offspring, each one containing two copies of the seed shape gene and two copies of the seed color gene. Thus, the various phenotypes were present in a 9:3:3:1 ratio.

Mendel's results supported the hypothesis that different characters assort themselves independently. Using the modern notion of genes, ***Mendel's law of independent assortment*** states:

Two different genes will randomly assort their alleles during the formation of haploid cells.

In 1866, Mendel published his work, entitled "Experiments on Plant Hybrids". This paper was largely ignored by scientists at that time, possibly because of its title. Another reason his work went unrecognized could be tied to a lack of understanding of chromosomes and their transmission. Nevertheless, Mendel's ground-breaking work allowed him to propose the natural laws that now provide a framework for our understanding of genetics.

In 1900, the work of Mendel was independently rediscovered by three biologists with an interest in plant genetics: *Hugo de Vries* of Holland, *Carl Correns* of Germany, and *Erich von Tschermak* of Austria. Within a few years, the influence of Mendel's studies was felt around the world.

Mendelian inheritance in Humans

Some human traits show Mendelian pattern of inheritance. It means that a gene has only two possible versions: one dominant and one recessive. According to Mendel's rules, a dominant allele reveals itself in phenotype of either homozygote or heterozygote whereas recessive allele may manifest only in homozygous condition. There are only a few examples human traits controlled by a single gene. Table 1 lists some of the most common single-gene traits in humans. As it shown in the table very often single-gene traits in human involve an abnormality that is disabling or life-threatening. Some traits were previously believed to be Mendelian, but their inheritance is probably based on more complex genetic models, possibly involving *more than one gene*: eye color, *hair color*, *Morton's toe*, tongue rolling,

widow’s peak, *earlobes*, *Hitchhiker's thumb* and etc. Much more traits arise from the interaction of many genes.

Table 1

Some of the most common single-gene traits in humans

Trait	Effect
Caused by a dominant allele	
Advanced Sleep Phase Syndrome	An inherited abnormal sleep pattern in which the individual is a "morning lark" and consistently goes to sleep very early and is very early to rise. The individual's blood melatonin level and the body core temperature rhythm that are preordained by our daily biologic (circadian) clock are phase-advanced by 3 to 4 hours.
ACHOO Syndrome (Autosomal Dominant Compelling Helioopthalmic Outburst)	Uncontrollable sneezing in response to the sudden exposure to bright light, typically intense sunlight.
Hypercholesterolemia	Missing protein that removes cholesterol from the blood; heart attack by age 50.
Huntington disease	Progressive mental and neurological damage; neurologic disorders by ages 40–70.
Caused by a Recessive Allele	
Thalassemia	Reduced amounts of hemoglobin anemia, bone and spleen enlargement
Sickle-cell anemia	Abnormal hemoglobin; sickle-shaped red cells, anemia, blocked circulation; increased resistance to malaria.
Cystic fibrosis	Defective cell membrane protein; excessive mucus production; digestive and respiratory failure.
Tay-Sachs disease	Missing enzyme; buildup of fatty deposit in brain; buildup disrupts mental development.
Phenylketonuria (PKU)	Missing enzyme; mental deficiency.
Albinism	Failure the production of melanin, the pigment that colours the skin, hair and eyes.