

BIT17R241 – 2019-20 Even Semester – Notes

Genetics Terms and Meanings:

Genetics is the branch of science concerned with genes, heredity, and variation in living organisms.

Gene: Genes are parts of DNA and carry hereditary information. It normally encodes a protein.

Allele: Genes come in more than one form. The different forms are called alleles. Different forms (means – different forms of DNA sequence) of a gene at a particular locus of given organism is called as allele.

Genotype: The genotype is the set of genes in our DNA which is responsible for a particular trait.

Phenotype: The phenotype means the physical expression, or appearance of character/trait. Any character that can be observed in an organism is called as phenotype.

Homozygous: At a particular locus in diploid organism, when both copies of a gene having same allele it is referred as homozygous.

Heterozygous: At a particular locus in diploid organism, when both copies of a gene having different allele it is referred as heterozygous

Homologous Chromosomes: In diploid organism, when two chromosomes are having similar size, length and similar genes, they are referred as homologous chromosomes.

Dominant: In heterozygous situation, the allele that is responsible for the final outcome of a character/ trait or phenotype is called as dominance.

Recessive: In heterozygous situation, the allele that is suppressed and not expressing the final outcome of a character/ trait or phenotype is called as recessive. Recessive alleles are expressed only when they are homozygous in a given locus.

Self Pollination: Fertilization of gametes from the same flower or same plant is called as self pollination.

Cross Pollination: Fertilization of gametes between two different plants of same species is called as cross pollination. It is normally done by emasculation of one flower, which means removal of anthers before its maturation. Pollens from a different plant of the same species is collected and dusted over this emasculated plant stigma, allows fertilization.

F1 Generation: The first offspring (or filial) generation. The next and subsequent generations are referred to as F2, F3, etc

Monohybrid Cross: It is a cross between two plants of a same species that are different from each other in one particular character/trait or phenotype.

Dihybrid Cross: It is a cross between two plants of a same species that are different from each other in two characters/traits or phenotypes.

Mendelian Genetics: Law of Dominance:

Mendel Studied Seven Characteristics That Bred True:

Gregor Mendel of Austria (now Czech Republic) was doing experiments with pea plants. He did these experiments in 1856. He did various cross pollination experiments with pea plants for almost 8 years and meticulously recorded all results. He was studying following 7 characters in pea plants.

- 1) Height – Either Tall or Dwarf
- 2) Flower position: Axial or Terminal
- 3) Flower color : Purple or white
- 4) Seed color: Yellow or Green
- 5) Pod Color: Green or Yellow
- 6) Pod shape: Smooth or constricted
- 7) Seed shape: Round or wrinkled

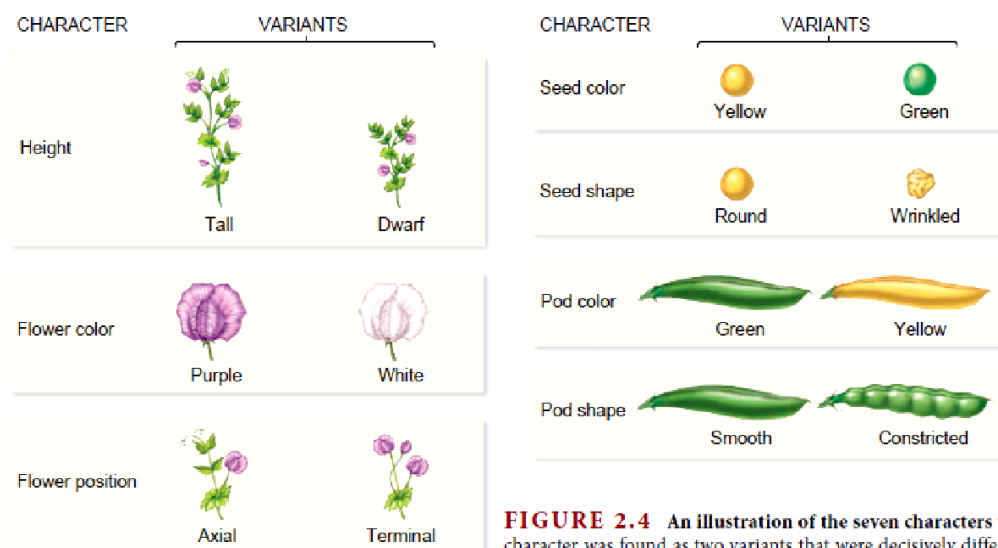


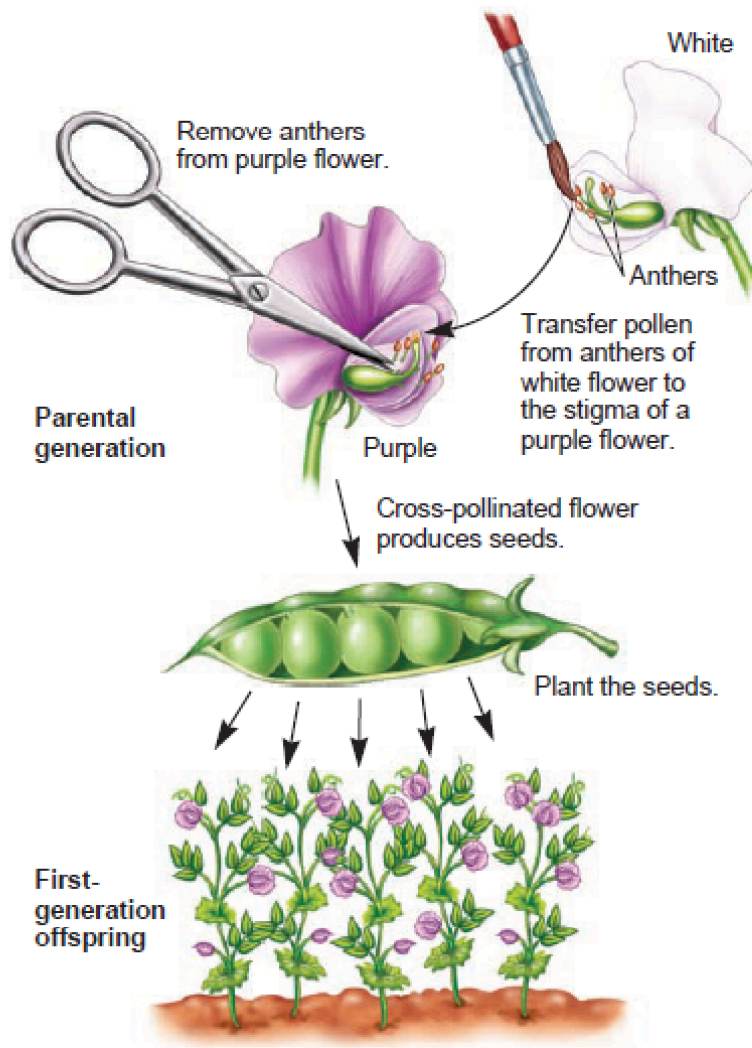
FIGURE 2.4 An illustration of the seven characters that Mendel studied. Each character was found as two variants that were decisively different from each other.

True Breeding lines: “True breeding lines” means these plants are naturally self-pollinated and shows similar characters of parents. There is no observable difference were observed from parents and off springs. True breeding lines are also called as “pure lines”.

When tall plants (True breeding line) are allowed to self pollination and produce seeds, all these seeds give rise to tall plants only. Likewise, true breeding lines always produce similar plants when they are allowed to self pollination. It was true for all 7 characters. When 1000 seeds are collected from tall true breeding line after self pollination, all 1000 seeds produced only tall plants.

Cross Pollination in Pea Plants:

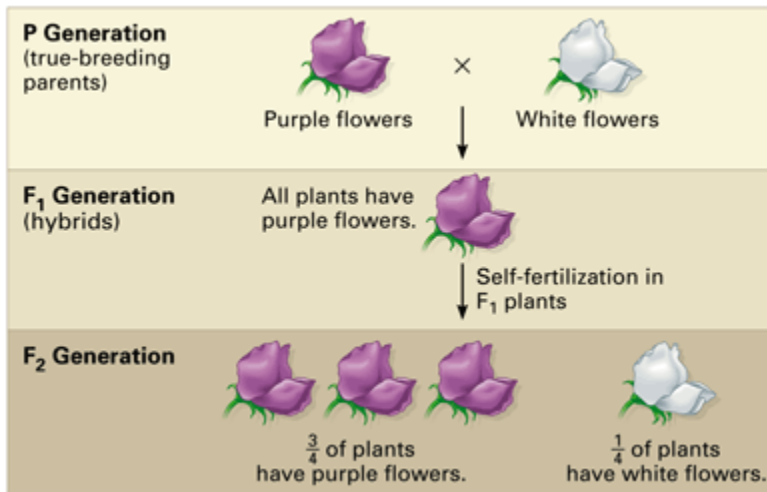
In year 1856, Chromosomes, genes, DNA or meiosis were not known or well understood. Mendel performed cross pollination. Cross pollination is performed between two plants. One plant is considered as male. From male plants mature pollen grains are collected and kept ready for fertilization. In another female plant, the male reproductive organ, stamens are removed completely before they get matured. Hence this flower ovary is not fertilized yet.



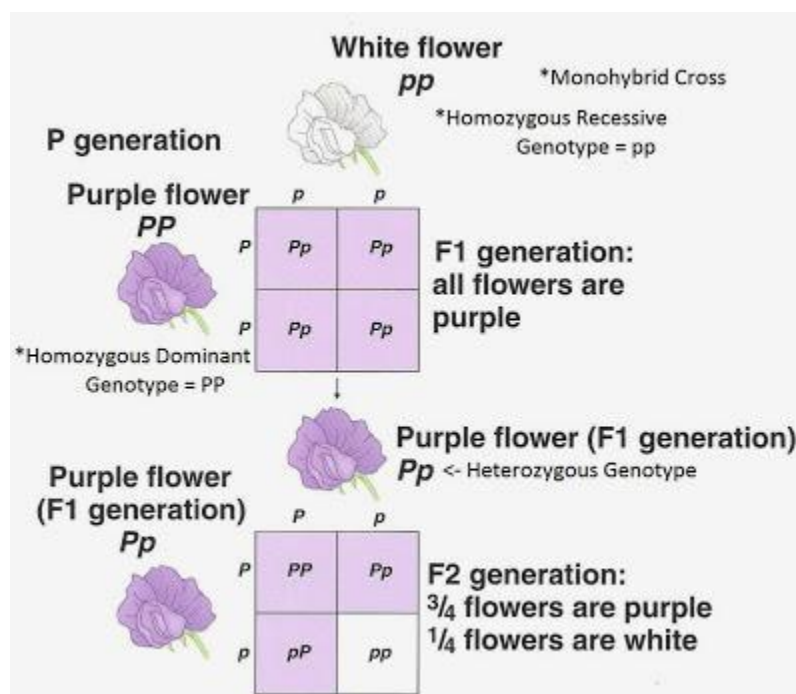
Above diagram shows an example of cross pollination between two pea plants, where one produce purple flowers and another produce white flowers. The cross pollinated

plant produce seeds after the fertilization. When the seeds were planted for germination, all seeds grown and gave purple flowers.

He allowed the F₁ plants for self fertilization and collected seeds from that. When these seeds were germinated he noticed that among 120 seeds, approximately 90 seeds produced purple flowers and around 30 seeds produced white color flowers.



The same can be explained by Punnett square model. The gametes produced by each parent are listed on either side and all the possible combinations are shown below.



Conclusion: Mendel's data (3:1 in F₂ generation) was observed in all 7 different characters he studies. He did not find any blending (or intermediate character) of the any two character. For example, he did not find any intermediate size of plant in case of height character. It was clearly either tall or dwarf. In other character, either it was white or purple. No slightly purple color flowers were found in F₂ generation. Hence he proposed that, there are two unit factors (alleles/genes) control a single trait. One is dominating or masking other character when they are heterozygous. Here, as the purple color was found in all the plants of F₁ generation, it must be dominant character. White color flower was observed only in F₂ generation and it was found in very low ratio. Hence it must be recessive character.

Mendelian Law of Segregation:

Mendel Followed the Outcome of a Single Character for Two Generations:

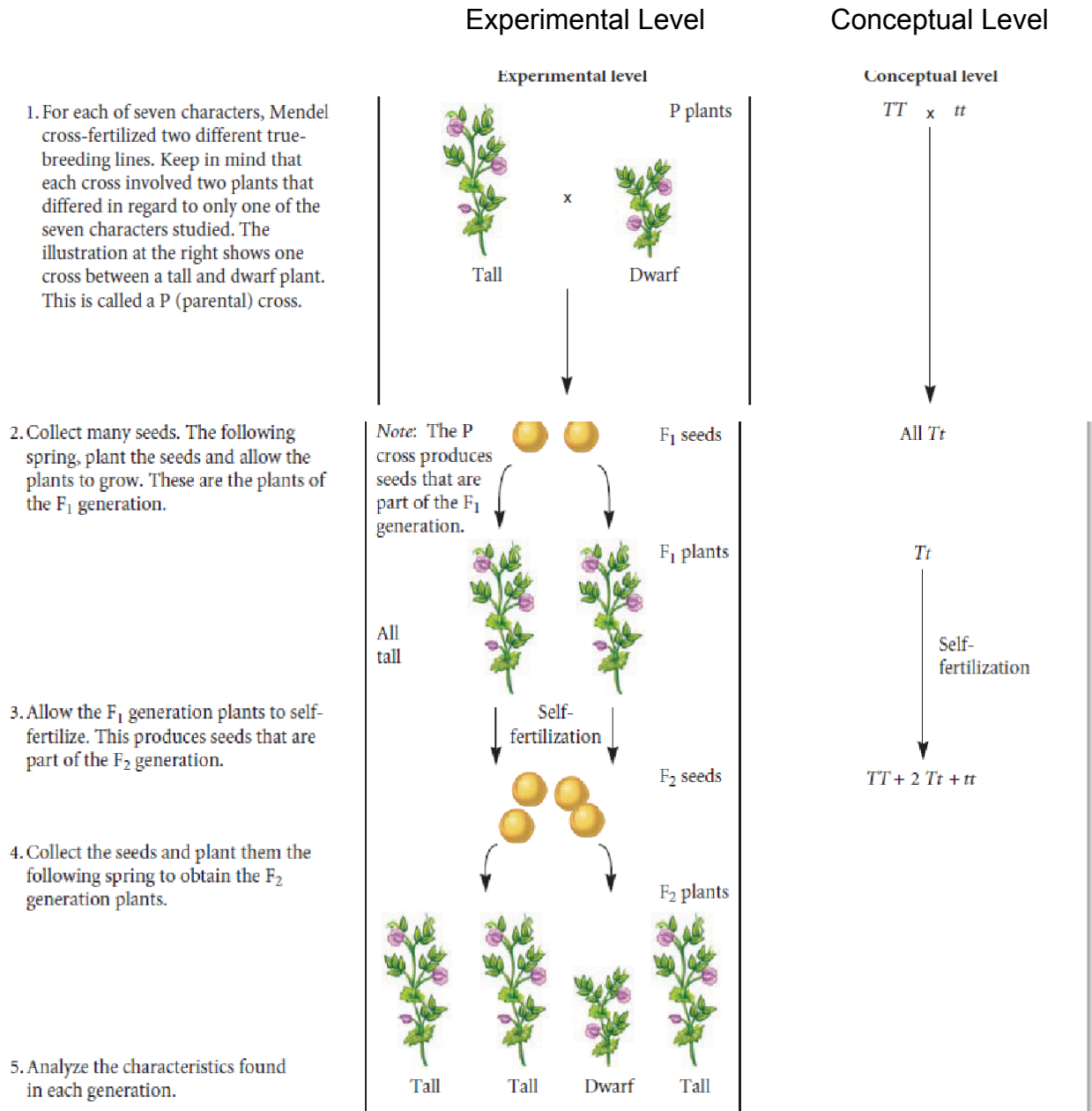
Law of segregations states that:

The two copies of a gene segregate (or separate) from each other during transmission from parent to offspring. Now we know very well that all diploid organisms have two copies for each gene. They segregate (or separate) from one another during the formation of gametes. Once the male and female gametes join together after fertilization, the diploid is formed. Hence, each copy of gene in both chromosomes is inherited from each parent.

Mendel was doing monohybrid cross with seven different characters. He performed experiment up to 2 generations. He chose two parental generations (P) that are different in one particular character. Here, for example he chose two plants that are different in height. One was tall plant and other one was dwarf (short). He cross pollinated these two plants (one is Tall and another is dwarf) by emasculating one of the plants.

The following diagram shows both experimental and conceptual level. He already proposed that two copies of genes are present for each character or trait. Based on this concept, the production of gametes from each parent is denoted here. Tall homozygous nature is denoted as TT. Dwarf homozygous nature is shown as tt. When they are heterozygous it is marked as Tt. Hence, Tall is dominating character; the plants are Tall in phenotype when they are heterozygous in genotype.

In F₁ generation he observed that all the plants were tall. If he germinated 1000 seeds from after the cross, all 1000 were tall plants. Later, he was allowing these F₁ plants to self pollinate and collected seeds. The seeds germination and grown into pea plants and the phenotypic ratio was 3:1. The results were similar for all the 7 different monohybrid cross he performed with pea plants.



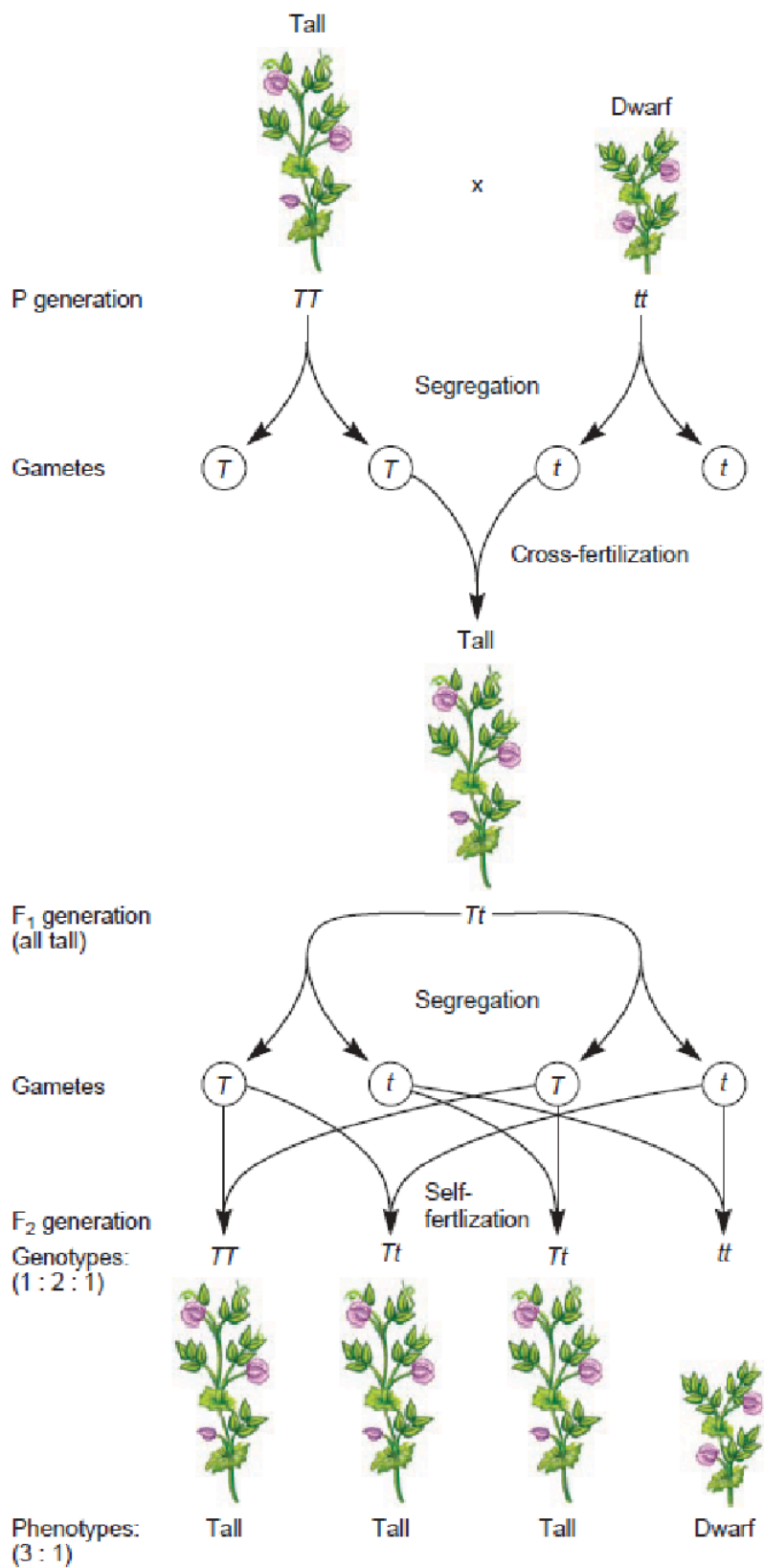
The possible segregation is shown below. Tt heterozygous will become into T and t . Likewise, this will happen in both male and female part of flowers. When they join together randomly, it produces the following genotypes.

TT – Tall plants; (one homozygous – dominant alleles)

Tt – Tall plants (two heterozygous)

tt – Dwarf plants (one homozygous – recessive alleles)

Hence the genotypic ratio is 1:2:1. The diagram is shown below.



The same can be inferred by drawing Punnett square. One side of the square, the male possible gametes should be given. Other side of the square, the possible female gametes should be given. This will help to identify all possible combinations. This will also provide 1:2:1 genotypic ratio.

		Male gametes	
		♂	
		<i>T</i>	<i>t</i>
Female gametes	♀	<i>TT</i>	<i>Tt</i>
	<i>t</i>	<i>Tt</i>	<i>tt</i>

By doing single hybrid cross Mendel proposed that the two copies of the gene separate from each other when inherited from one generation to next generation.

Law of Independent Assortment:

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 other words, the allele for one gene will be found within a resulting gamete independently of whether the allele for a different gene is found in the same gamete.

Mendel proposed this law after performing dihybrid cross with pea plants. It means, he followed the inheritance of two different characters within the same groups of individuals. For example, let's consider an experiment in which one of the characters was seed shape, found in round or wrinkled variants; the second character was seed color, which existed as yellow and green variants. In this dihybrid cross, Mendel followed the inheritance pattern for both characters simultaneously.

Mendel already know from his previous experiments that Yellow colour in seeds is dominant characters whereas green colour is recessive character. Smooth seed coat shape is dominant character and wrinkled seed coat shape is recessive character. As expected in F1 generation, he observed all plants showed Yellow colored and round shape seeds. He was allowing the plants to self fertilized and followed the seed colour and shape pattern in F2 generation. There are two possibilities. One possibility is that both genes are linked in the same chromosomes. Other possibility is both genes are present in two different chromosomes. Outcome of such two possibilities in F2 generation is given below in diagram.

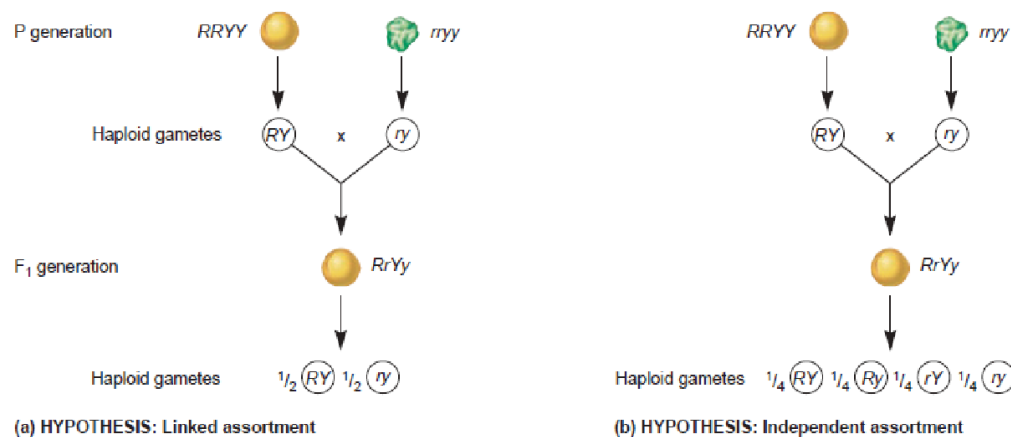


FIGURE 2.7 Two hypotheses to explain how two different genes assort during gamete formation. (a) According to the linked hypothesis, the two genes always stay associated with each other. (b) In contrast, the independent assortment hypothesis proposes that the two different genes randomly segregate into haploid cells.

In F₂ generation, he would be observing 3:1 (or 12:4 – both are same) if both genes are linked. If they are present in two different chromosomes he would get 9:3:3:1. He received such results.

Interestingly, he observed non-parental characters in F₂ generation. Non-parental characters means, new combination of characters that were NOT seen from parental plants. For example, in F₂ generation, he observed four different phenotypes as given below.

<i>P</i> cross	<i>F</i> ₁ generation	<i>F</i> ₂ generation
Round, yellow × wrinkled, green seeds	All round, yellow	315 round, yellow seeds 108 round, green seeds 101 wrinkled, yellow seeds 32 wrinkled, green seeds

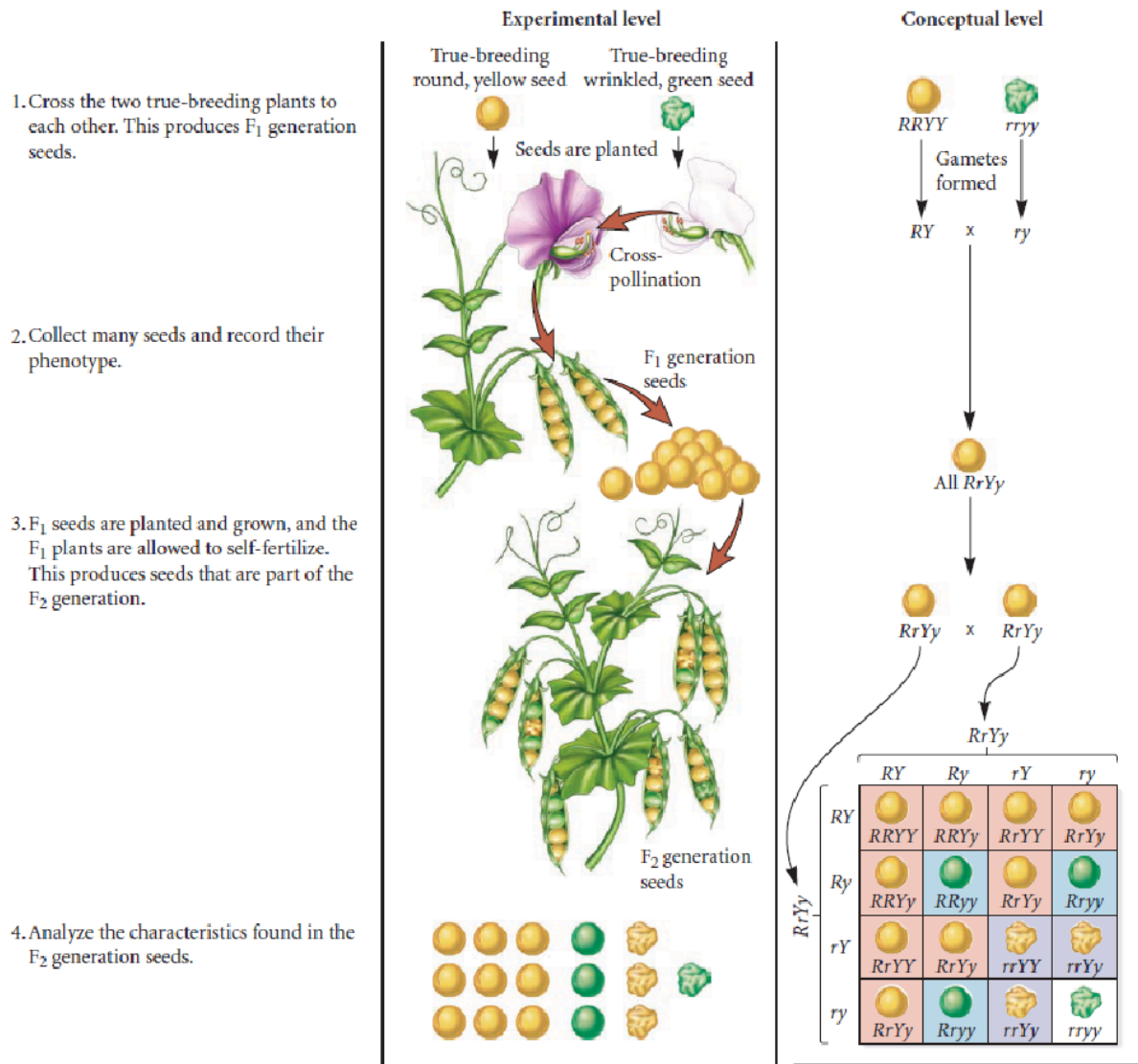
Round Yellow Seeds: 9 Ratio - Parental Combination
 Green seeds : 3 Ratio – Non parental combination
 Wrinkled Yellow : 3 Ratio – Non Parental combination
 Wrinkled Green : 1 Ratio - Parental Combination.

The experimental level and conceptual level diagrams are given below, when these two characters are located in two different chromosomes:

F₁ generated plants will produce 4 different possible gametes: RY, Ry, rY and ry. Both male and female gametes will produce these 4 different games and it gives 16 different possible combinations as given below in Punnett square model.

TESTING THE HYPOTHESES — FIGURE 2.8 Mendel's analysis of dihybrid crosses.

Starting material: In this experiment, Mendel began with two types of true-breeding pea plants that were different with regard to two characters. One plant had round, yellow seeds ($RRYY$); the other plant had wrinkled, green seeds ($rryy$).



Since he observed that, Round Yellow character was found in 9 proportion, the green round seeds are in 3 proportion; the Wrinkled yellow seeds were found in 3 proportion, and green wrinkled was in 1 proportion (9:3:3:1) he proposed that, the inheritance of one character from one generation to next generation is independent of another character.