Factors Contributing to the Success of Mendel's Experiments

As explained earlier, the plant which Mendel chose for his experimentation was the garden pea, *Pisum sativum*. It proved to be a very satisfactory subject for experimentation on hybridization because of the following reasons:

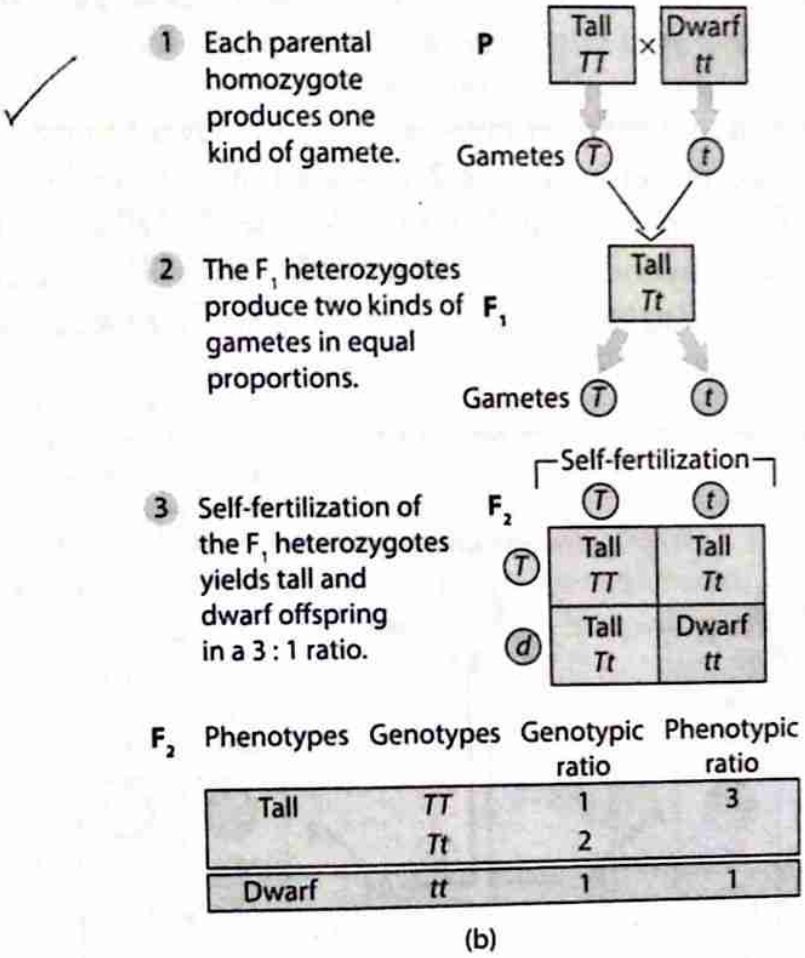
1. It is easy to cultivate this plant and it completes its life cycle in one season only.
2. It has sharply distinct heritable differences seen morphologically.
3. A variety of seeds can easily be purchased that would give rise to plants with distinct characteristics.
4. In pea, the petals of the flower close down tightly, preventing pollen grains from entering or leaving, thus enforcing self-fertilization (male and female gametes from the same flower fuse to produce seeds).

The reasons for the success of Mendel's experiments are stated as under:

1. He had mathematical background, so he kept accurate quantitative and applied statistical analysis and mathematical logic in his experimental analyses.
2. He used large sampling size to give more credibility to the data collected by him.
3. He focused on one character at a time and kept track of each character separately.
4. He focused on contrasting characters of plants that were otherwise part of the same trait, such as, tall vs. dwarf, green vs. yellow and so on.
5. The general rules of inheritance laid down by him were substantiated by the inferences drawn from the experiments conducted on plant samples from successive generations.

Punnett Square Method

For situations involving one or two genes, it is possible to write down all the gametes and combine them systematically to generate the array of zygotic genotypes. This procedure is called the Punnett square method after the British geneticist Reginald C. Punnett. It is a straight forward way of predicting the outcome of crosses by probability. Figure 1 shows the Punnett square method used for cross described above.



F_2	Phenotypes	Genotypes	Genotypic ratio	Phenotypic ratio
	Tall	*TT*	1	3
		Tt	2	
	Dwarf	*tt*	1	1

(b)

Figure 1 Punnett square symbolic representation of the cross between tall and dwarf peas.

Dihybrid Cross

Mendel found that a cross between round yellow and wrinkled green seeds (P_1) produced only round and yellow seeds in F_1 generation, but in F_2 four types of combinations were observed. These are:

(a) Round yellow 9 parental combinations
(b) Round green 3 non-parental combinations
(c) Wrinkled yellow 3 non-parental combination
(d) Wrinkled green 1 parental combination

Thus, the offspring of F_2 generation were produced in the ratio of 9:3:3:1 phenotypically and 1:2:2:4:1:2:1:2:1 genotypically. This ratio is called dihybrid ratio. Four types of gametes with two old and two new combinations, that is, *RY*, *ry*, *Ry*, *rY* are formed from the F_1 hybrid. These four types of gametes on random mating produced four types of offspring in the ratio of 9:3:3:1 in F_2 generation.

Correns converted two of the generalizations of Mendel into two laws of heredity:

1. **Law of segregation (Purity of gametes):** The law of segregation states that when a pair of contrasting factors or genes or allelomorphs are brought together in a heterozygote (hybrid) the two members of the allelic pair remain together without being contaminated and when gametes are formed from

the hybrid, the two-separate out from each other and only one enters each gamete as seen in mono-hybrid and dihybrid cross. That is why the law of segregation is also described as law of purity of gametes.

2. **Law of independent assortment:** If the inheritance of more than one pair of characters (two pairs or more) is studied simultaneously, the factors or genes for each pair of characters sort out independently of the other pairs. Mendel formulated this law from the results of a dihybrid cross.

Test Cross

A cross between an organism of dominant phenotype with an unknown genotype and a known recessive phenotype (homozygous) is called test cross. Figure 2 shows a testcross in which seed characteristics are used. In the figure, "*r*" stands for the recessive allele for wrinkled seeds; "*R*" stands for the dominant allele for round seeds. In this testcross, the hybrid F_1 plants having round seeds (*Rr*) are crossed with plants having wrinkled seeds (*rr*) to obtain 208 seeds:106 round seeds, and 102 wrinkled seeds—a 1:1 ratio, showing F_1 hybrid to be heterozygous for the trait.

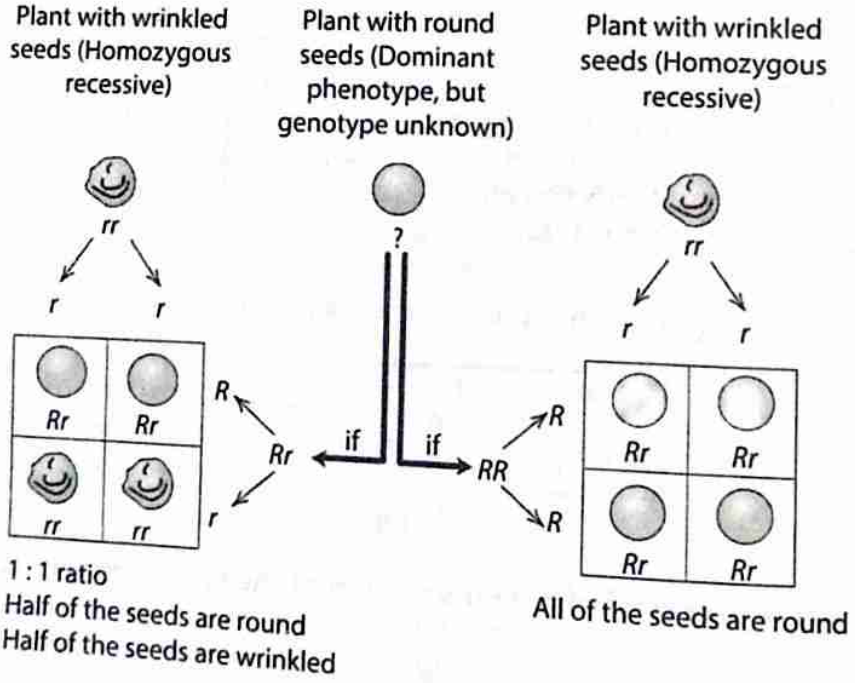


Figure 2 The hybrid F_1 plants having round seeds (*Rr*) are crossed with plants having wrinkled seeds (*rr*).

Back Cross

A genetic cross between a hybrid progeny and any of the parent (or an organism with genotype similar to the P_1 parents) is called a back cross. It is different from reciprocal cross, which is the cross between different parents with sexes reversed is known as reciprocal cross. For example, tall male with dwarf female or dwarf male with tall female. Both back cross and reciprocal cross are used in plant breeding and animal breeding to obtain desired offspring.

6.3 Gene Interacti

Inter-allelic or Intra-genic Gene Interaction

1. **Incomplete dominance (1:2:1 ratio):** Here F_1 hybrids are not related to either of the parents, but exhibit a blending of characters of two parents. This is called incomplete dominance or blending inheritance. For example, a cross between red and white flowers of Four O'clock plant, *Mirabilis jalapa*, produced pink flowers in F_1 generation.
2. **Co-dominance (1:2:1 ratio):** In co-dominance, both the genes of an allelomorphic pair express themselves equally in F_1 hybrids. 1:2:1 ratio both genotypically as well as phenotypically in F_2 generation. For example, MN blood type in man is an example of co-dominance. The persons with MN genotype produce both antigen M and N and not some intermediate product indicating that both the genes are functional at the same time.

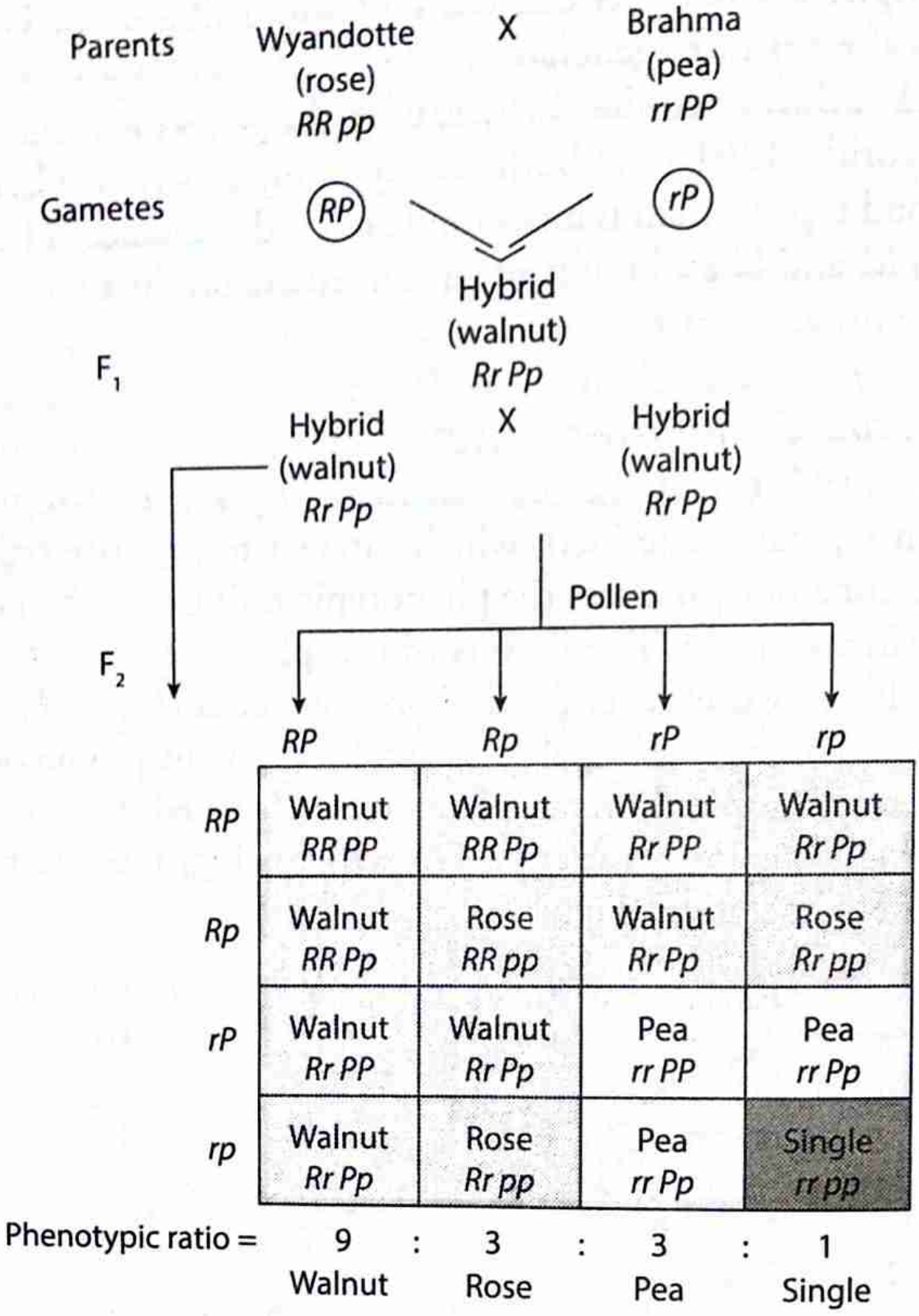


Figure 4 Crosses of supplementary genes in chickens.

3. **Epistasis (Inhibiting genes):** Epistasis is the interaction between non-allelic genes (present on separate loci) in which one-gene masks, inhibits or suppresses the expression of other gene. The gene that suppresses the other gene is known as inhibiting or epistatic factor and the one, which is prevented from exhibiting itself, is known as hypostatic. Although, it is similar to dominance and recessiveness, but the two factors occupy two different loci. Epistasis can be of the following types:
   (a) Dominant epistasis (12:3:1 or 13:3 ratio): In dominant epistasis out of two pairs of genes the dominant allele, (i.e., gene A) of one gene masks the activity of other allelic pair (Bb). Dominant epistasis in dogs.
   (b) Recessive epistasis (9:3:4 ratio): Epistasis due to recessive gene is known as recessive epistasis, that is, out of the two pairs of genes, the recessive epistatic gene masks the activity of the dominant gene of the other gene locus, for example, in mice agouti colour.
4. **Duplicate genes (15:1 ratio):** Sometimes two pairs of genes located on different chromosomes determine the same phenotype. These genes are said to be duplicate of each other. For example, in shepherd's purse (*Bursa bursapastoris*), the seed capsules have two shapes—triangular and oval. If the genotype is *aabb*, that is, only recessive alleles in both genes are present, then oval seed capsules were formed. However, if the dominant allele of either gene is present, triangular seed capsules are formed.

5. **Pleiotropic effect of genes:** Certain genes are known to control the viability of the organism. These genes are known as lethals or semilethals depending upon their influence. Complete lethal genes in homozygous condition kill nearly all homozygous individuals, while in the case of semilethal genes some homozygous individuals are able to survive.

6.9 Genetic Disorders

The various types of genetic disorders are grouped into two broad categories: (1) Mendelian disorders, (2) Chromosomal disorders (described later).

Human Karyotype

The normal diploid number of chromosomes in man is 46 (i.e., 23 pairs). 22 pairs out autosomes and one pair of sex chromosome. A summary of the chromosome constitution of a cell or an organism is known as its karyotype. Study of human chromosomes has helped a lot in correlating various human diseases, malformation and deformities with the abnormalities in the number and structure of chromosomes. These abnormalities may be in the autosomes or sex-chromosomes.

Pedigree Analysis

Pedigree analysis is a method based on Mendelism. A pedigree is a record of inheritance of certain genetic traits for two or more generations presented in the form of a diagram or family tree or case history or genealogy. To find out the possibility of absence/presence of a particular trait in the progeny. It is mainly employed in domesticated animals and man also.

Mendelian Disorders

These disorders are inherited to the offspring following principle of inheritance and are mainly caused by alterations or mutation in alleles of a single gene.

1. Hemophilia is an X-linked disorder in which persons are unable to produce a factor for blood clotting. The principal type of hemophilia in humans is due to a recessive X-linked mutation, and it is most common in males.
2. Color blindness or color vision deficiency is decreased ability to perceive color differences; under normal light conditions. Color blindness is usually a sex-linked condition.
3. Sickle cell anemia is an autosomal recessive disorder that is inherited. People with two sickle cell genes have severe anemia; those with only one defective gene have the sickle cell trait.
4. Phenylketonuria or PKU is a genetic error of protein metabolism characterized by elevated blood levels of the amino acid phenylalanine.
5. Thalassemia is an autosomal recessive form of anemia which occurs due to mutation or deletion of the genes controlling the synthesis of globin chains of hemoglobin. It is mainly of three types—α-Thalassemia, β-Thalassemia, and δ-Thalassemia.
6. Alkaptonuria is an autosomal recessive genetic disease caused by problems in phenylalanine and tyrosine metabolism. It is caused due to a defect in the enzyme homogentisate 1,2-dioxygenase, which catalyzes the degradation of tyrosine.
7. Polydactyly is condition in which the individual develops extra fingers or toes. It is caused by incomplete penetrance of trait, that is, when individuals do not show a trait even though they have the appropriate genotype.

8. Gaucher's disease is an autosomal recessive disorder is caused by deficiency of enzymes lysosomal exoglycosidase and β-glucocerebrocidase which leads to accumulation of glucocerebrocides in white blood cells, causing malfunction of spleen, liver, kidneys, lungs, brain and bone marrow.
9. Tay Sach's disease is caused by a mutant allele located on HEXA gene, which also encodes the enzyme hexosaminidase A and is inherited to homozygous individuals. It leads to accumulation of ganglioside GM2 and blockage of the action of neurons, leading to deterioration of the nervous system and eventually paralysis.
10. Huntington's disease is a neurodegenerative disorder caused due to dominant gene (HD gene) on chromosome 4.
11. Cystic fibrosis is an autosomal recessive mutation of CF gene and leads to abnormal mucus in the lungs, pancreas, and sweat glands.

Chromosomal Disorders

These are rarely inherited to the offspring and are mainly caused by non-disjunction of chromosomes during meiosis.

1. The presence of an extra copy of chromosome 21 leads to Down syndrome.
2. An O gamete from either the mother or the father fusing with an X gamete to form an XO zygote develops into a female with Turner syndrome. The individual has 45 chromosomes.
3. A zygote with an extra Y (XYY) develops into a physically normal male who is likely to be taller than average and has 47 chromosomes, and thus suffers from Klinefelter's syndrome.