Chapter 32

MENDELIAN GENETICS

Mendel established principles that describe transmission of genes from parents to offspring. He derived these principles from his experimentation. When Mendel began his studies he knew nothing about chromosomes and role of meiosis in inheritance. Despite of this, he was able to determine the existance of units of inheritance. He also predicted the behaviour of these units during gamete formation. Later investigations established parallel behaviour between chromosomes and Mendel's unit of inheritance during meiosis.

Gregor Mendel —— About Mendel

Gregor Johann Mendel was born to a peasant family in the village of Heinzendori, now part of Czechoslovakia in 1822. Mendel studied Philosophy during his earlier part of educaion. In 1843, he was admitted to Augustinian monastery in Brno. In 1849, he joined as teacher and worked for several years. During 1851-53 he attended University of Vienna, where he studied physics and botany. In 1854 he returned to Brno where he taught physics and natural science for 16 years. In 1856 Mendel performed his first hybridization experiment. He was elected abot in 1868, but continued his research. Mendel died of a kidney disorder in 1884.

Mendel's Experimental Approach

Many workers tried to provide experimental evidence relating to inheritance before Mendel, but Mendel's work is an elegant model of practical work. He showed correct approach and methodology necessary for good experimental work. For instance:

i. He chose an organism easy to grow and interbreed. It grows to maturity in a single season.

ii. He worked with seven characters, each represented by two contrasting traits or forms. For example, he selected tall and dwarf traits for character stem height.

iii. He worked with true-breeding strains, the forms that appeared generation after generation.

iv. He restricted his experimental work to one or very few pairs of contrasting traits in each experiment.

v. He kept quantitative records, a necessity in genetic experiments. From the analysis of his data he derived certain postulates which become basis for principles of transmission of hereditary traits.

The significance of Mendel's work was realised in 20th century when his publications and work was rediscovered by geneticists investigating function and behaviour of chromosomes.

The Choice of Experimental Organism

Mendel selected garden pea plant (*Pisum sativum*) because it showed several sharply contrasting characteristics that were without intermediate forms, and were relatively unaffected by environmental factors. Also, the flowers of garden pea are self-pollinating in nature, and so the crosses can be arranged according to the choice of breeder.

Mendel's Characters

The characters that caught Mendel's attention were:

a. length of stem: *tall or short*

b. position of flowers: *axial or terminal*

c. colour of unripe pods: *green or yellow*

d. form of ripe pods: *inflated or constricted*

e. form of ripe seeds: *round or wrinkled*

f. colour of cotyledons: *yellow or green*

g. colour of seed coat: *grey-brown or white*

Figure 28.1 Characters of the garden pea (*Pisum sativum*) that were studied by Mendel; the character of height (tall and dwarf plants) is illustrated in Figure 28.5

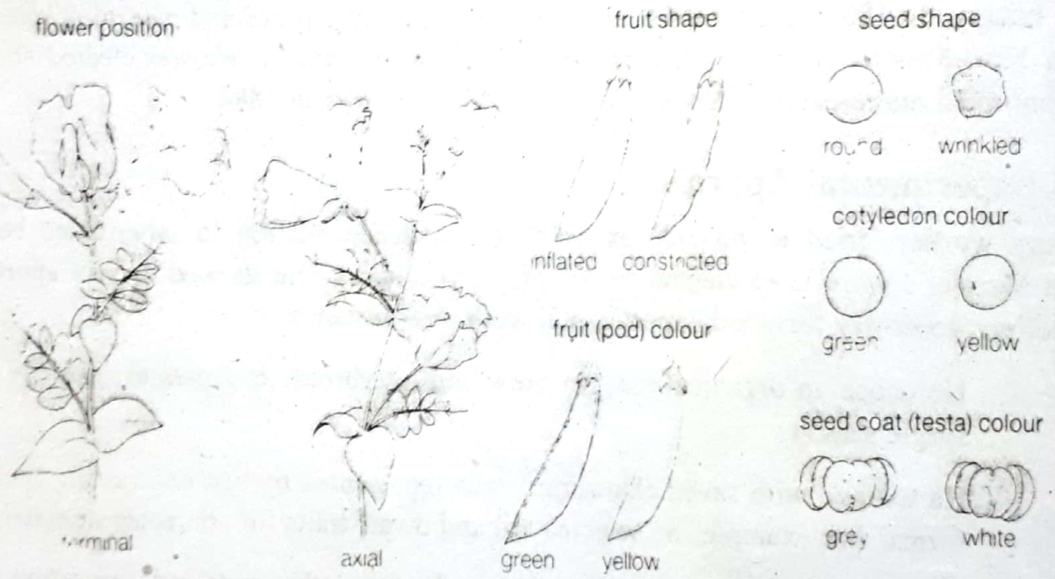

Fig. 32.1: Characters of garden pea (*Pisum sativum*) studied by Mendel.

MENDEL'S WORK

The significance of Mendel's work lies in his ability to formulate a scientific problem. The aim of his experiments was to study the numbers and kinds of offspring produced by hybrid individuals and to determine from the observations whether any stastical relationship existed among these offspring.

Verification of Pure Lines

The pea plants that Mendel used in his experiments were grown from seeds that was obtained from plants that had "bred true" when self-fertilised for at least two generations before the experiment. Such plants are called **true breeding**. When self-pollinated and self-fertilised, the offspring always resemble the parents for the given trait.

MONOHYBRID CROSS

Mendel performed crosses in which only one pair of contrasting allele or alternative traits is being followed. Each such cross is known as **monohybrid cross**.

Two individuals from different parent strains, each exhibiting one of the two contrasting traits of character under study, were mated. For example, Mendel crossed pea plants with tall stems and dwarf stems. Tall and dwarf represent contrasting traits of the character stem height. The original parents are called **P_1** or **parental generation**. The offspring produced was called **first filial generation** or **F_1**. All the F_1 individuals (1064 in number) were tall, regardless whether the parent plant was tall or dwarf. Mendel called these plants **hybrids**. The F_1 hybrids were allowed to self-pollinate. Their offspring was named **second filial generation** or **F_2**. Mendel observed that three-fourth (787 in number) of F_2 plants were tall and one-fourth dwarf (277 in number). Mendel continued with the selfing of the F_2 progeny for four more genrations to confirm his interpretation of the results. Another aspect of monohybrid cross was that the inheritance was similar in F_1 and F_2 when reciprocal crosses were perfomed.

Figure 28.5 Mendel's experiment with tall and dwarf peas

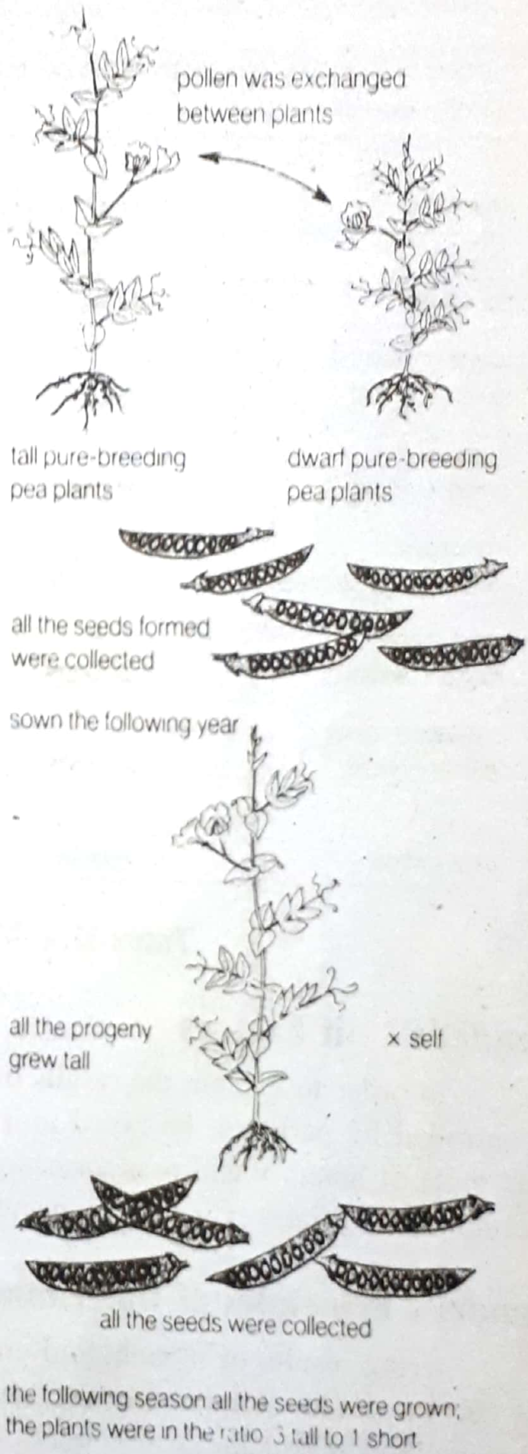

Fig. 32.2: Mendel's experiment with tall and dwarf peas.

Mendel's Mathematical Approach

In collecting results, Mendel recorded the numbers of individuals in each class in the progeny, and established the ratios of the contrasting characters of many subsequnet generations. This involved counting many thousands of seeds. This helped him to formulate a hypothesis to explain the ratios obtained and devise test crosses.

Mendel made monohybrid crosses for each of other pairs of contrasting traits. He found that F_1 offspring were identical to one of the parents. In F_2 an approximate ratio of 3:1 was obtained. Three-fourth appeared like F_1 plants and one-fourth exhibited the contrasting trait, which had dispappeared in F_1.

Many plants were grown in each cross, and therefore large numbers of F_2 progeny were obtained. Mendel recorded the numbers of plants of each type, and expressed the outcome as a ratio.

Character	Progeny of F_1	Progeny of F_2: numbers	ratio
height: tall x dwarf	all tall	787 tall, 277 dwarf	2.84:1
flower position: axial x terminal	all axial	651 axial, 207 terminal	3.14:1
fruit colour: green x yellow	all green	428 green, 152 yellow	2.82:1
fruit shape: inflated x constricted	all inflated	882 inflated, 299 constricted	2.95:1
seed shape: round x wrinkled	all round	5474 round, 1850 wrinkled	2.96:1
cotyledon colour: yellow x green	all yellow	6022 yellow, 2001 green	3.01:1
testa: grey x white	all grey	705 grey, 224 white	3.15:1

Table 32.a: Mendel's seven monohybrid crosses.

Mendel's Unit Factors

In order to explain the results of monohybrid crosses, Mendel proposed that each trait is controlled by particles, he called **unit factors**, now known as **genes**. These factors serve as basic units of heredity and pass unchanged generation to generation. These are responsible for determination of traits expressed by plants.

Mendel's Principles of Inheritance

Using results of monohybrid crosses for various contrasting traits and to explain how unit factors could account for the results of monohybrid crosses, Mendel suggested the following three postulates or principles of inheritance.

i. Pairs of Unit Factors

Genetics characters are controlled by unit factors and these factors exist in pairs in organisms. For example, in monohybrid cross involving tall and dwarf, a specific unit exist for each trait: **T** for **tall** and **t** for **dwarf**. Since these factors appear in pairs, three combinations are possible which determine the stem height. These are **TT**, **Tt** and **tt**.

ii. Dominant *or* Recessive Unit Factors

When two different unit factors for a single character, T and t, for example, are present in an individual. One unit factor suppresses the other. One that dominates is called **dominant** and the other that is suppressed is known as **recessive**. For example, in a cross between tall (T) and dwarf (t), all the F_1 plants were tall, therefore T was dominant and t recessive.

iii. Segregation

At the time of formation of gametes, the paired unit factors seggregate or separate randomly so that each gamete recieves one of them. The reppearance of recessive dwarf plants in F_2 can be explained on the basis of this postulate.

The Chromosome Theory

Now that we know the structure of chromsomes, and about the way they behave during nuclear divisions, the interpretation of Mendle's experimental results present no difficulties.

Mendel's factors, **now called genes** are found on chromosomes in the **nucleus of the cells.** There are at least two forms of a gene, and these are called **alleles**. In Mendel'experiment, the allele in pea plants for "tallness" was **dominant** over the allele for "dwarfness", which is **recessive**. A pure-breeding tall pea plant is **homozygous** for the "tall" allele, and a pure breeding dwarf pea plant is homozygous for dwarf allele. The F_1 progeny are **heterozygous** "tall", with one allele for "tall" and one for "dwarf". In the heterozygous tall individual, we say that the recessive gene is not **expressed**. Conventionally, alleles are represented by the same letter to show that they are variants of a single gene, for example **T and t** for tall and dwarf plants. The dominant allele is given the capital letter.

The terms "homozygous" (TT or tt) and "heterozygous" (Tt) refer to *the genetic constitution of the individual called* **genotype**. The visible expression of gene, i.e., *the actual appearance of an organism, is called its* **phenotype**. The distinction between genotype and phenotype is important, since the environment may sometimes profoundly affect the appearance of an organism, for example a "tall" plant may actually quite short if unfavourable conditions have stunted its growth. The environment does not alter the genotype of the organism, however.

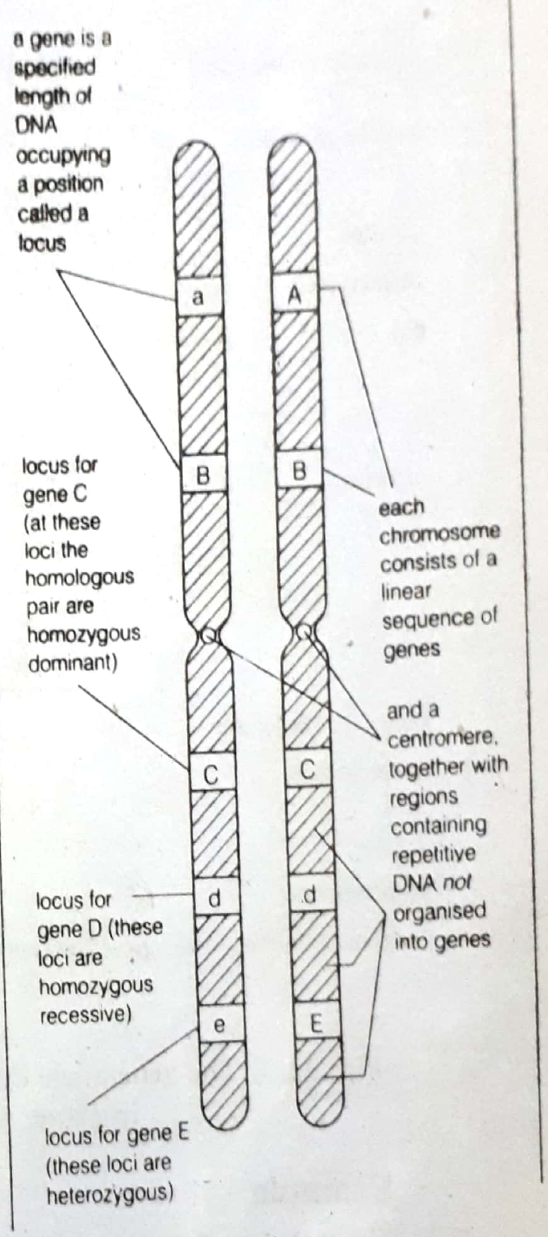

Fig. 32.3 Genes on a homologus pair of chromosomes.

Mendel's Law of Segregation of Gametes

The data of monohybrid crosses helped Mendel to recognise a pattern of heredity and formulate **Mendel's first law or law of segregation of gametes**. It states that:

"The hereditray characteristics are determined by **particulate units** *or* **factors**. *These unit factors occur in pairs in an individual, but in the formation of germ cells, these*

factors are seggregated so that only one member of a pair is transmitted through any one gamete. When the male and female gametes unite, the double number of factors are restored in the offspring". The word factor is substituted by the word gene now-a-days and rest of the statement is read same almost.

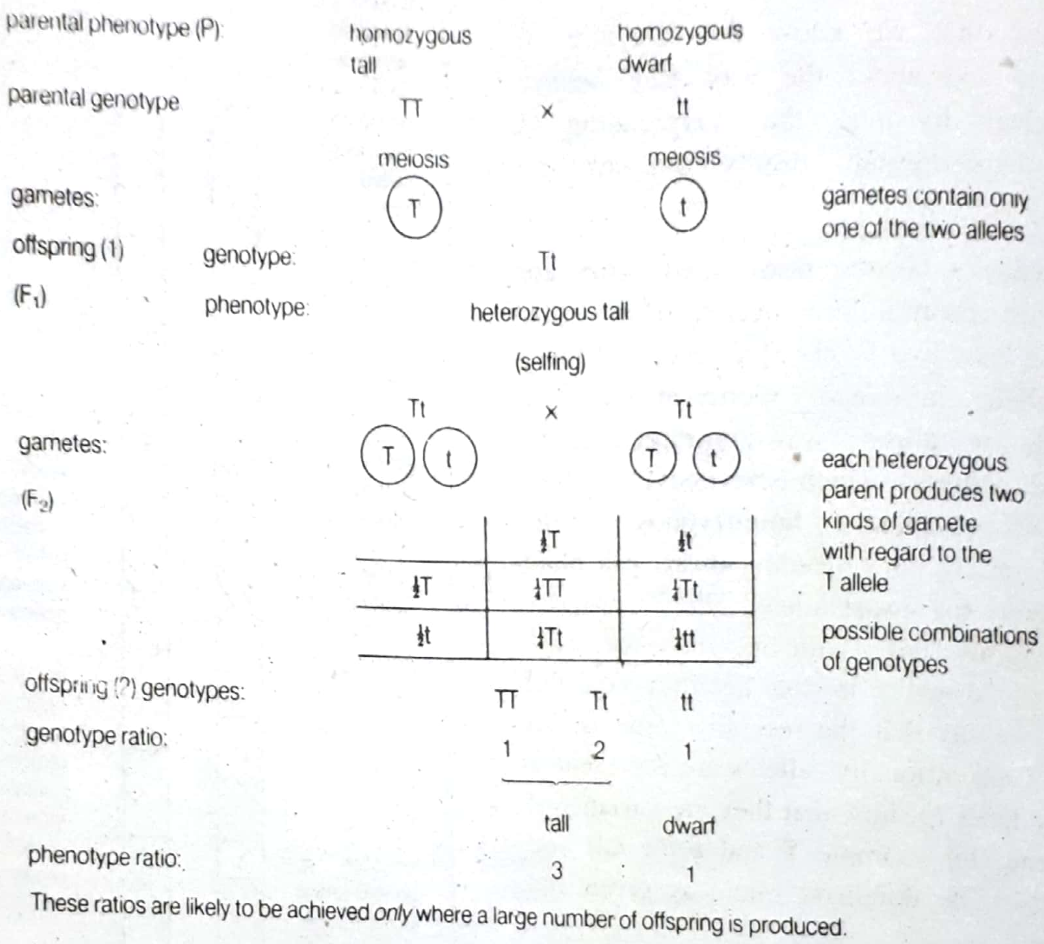

Fig. 32.3: The gene/allele interpretation of monohybrid cross. The gene for height in *Pisum sativum* has two alleles (T,t).

Example

The **tall/dwarf cross** explains the law of segregation of gametes. According to Mendel all the P_1 tall (TT) and dwarf (tt) plants contained similar unit factors (genes). During gamete formation the ova and pollen of tall plants received one tall unit factor (T) as a result of segregation. Similarly ova and pollen of dwarf plants received one dwarf unit factor (t). After fertilisation all F_1 plants received one of each unit factor (T and t), re-establishing the paired nature. Because tall is dominant to dwarf, all F_1 plants were tall.

When F_2 plants form gametes, each gamete receive either tall or dwarf unit factor. Random fertilisation result in formation of four possible combinations:

i. ***tall/tall*** **(TT);** **ii.** ***tall/dwarf*** **(Tt)**

iii. ***dwarf/tall*** **(tT);** **iv.** ***dwarf/dwarf*** **(tt)**

Combinations (i) and (iv) will result in tall and dwarf plants, respectively. Similarly combinations (ii) and (iii) will yield tall plants due to presence of dominant factor for tallness. Therefore, F_2 consists of three-fourth tall and one-fourth dwarf or a ratio of 3:1.

Punnet Square --- Prediction of Genotypes and Phenotypes

The genotypes and phenotypes resulting from the recombination of gametes during fertilisation are more easily visualised by constructing a **Punnet square.** All possible gametes are assigned to a column or a row, with the vertical columns representing those of the male parent and horizontal row those of the female parent. After entering the gametes in the row and columns, the new generation is predicted by combining the male and female gametic information for each combination and entering the resulting genotypes in the boxes. This process represent all possible random fertilisation events. The genotypes and phenotypes of all potential offspring are ascertained by reading the enteries in the boxes. Punnet square is also known as **checkerboard.**

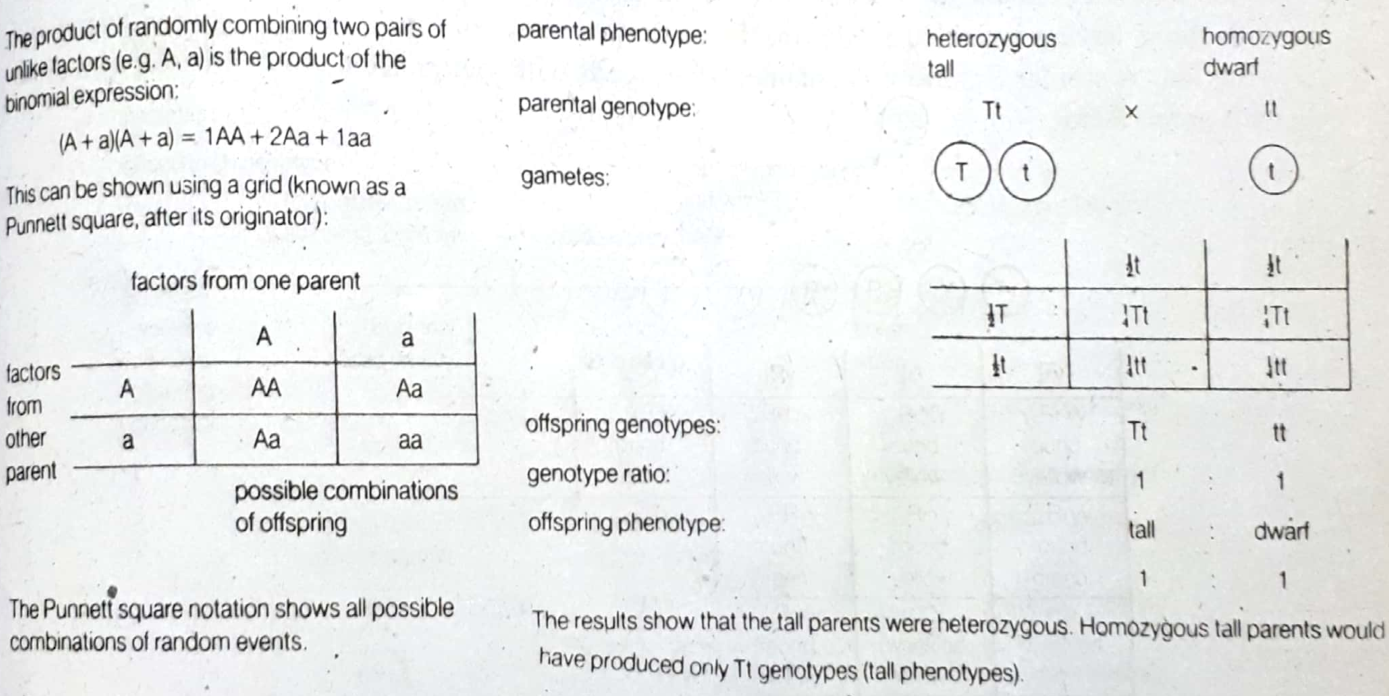

Fig. 32.4: The Punnet square and monohybrid testcross.

Test Cross

Tall plants produced in F_2 generation may either have TT or Tt genotypes. Mendel devised a simple way to distinguish the genotype. It is known as **test cross**. The organism of dominant phenotype (tall) but unknown genotype is crossed to homozygous recessive individual. For example, a tall plant is crossed with a dwarf plant. If the tall plant is homozygous (TT), all the F_1 plants would be tall. But if the tall plant is heterozygous (Tt), one-half of the F_1 plants would be tall (Tt) and one-half dwarf (tt), exhibiting a ratio of 1:1. This ratio is called **test cross ratio.**

Backcross

The term test cross is often used interchangeably with back cross. However, the two are not necessarily the same. The test cross is a cross of an individual of dominant phenotype to homozygous recessive. On the other hand backcross is a mating of an individual of known phenotype to any one of homozygous parents. For example in the above mentioned example the cross between F1 hybrid to homozygous recessive individual is a test cross, but if we cross the F1 hybrid to a homozygous tall (TT), it is a backcross. The backcross may have special value in some genetic analysis such as in case of incomplete dominance. Because the homozygous recessive individual is also one of the parents, therefore test and back crosses are regarded same.

DIHYBRID CROSS

In next phase of his experiments Mendel designed experiments in which two characters were examined simultaneously, in order to know how two pairs of factors behave in relationship to one another when followed in the same cross. ***A cross involving two pairs of contrasting traits is called* dihybrid cross.**

The shape of the *pea* seeds can be either round or wrinkled. The round factor (**R**) is dominant to wrinkled (**r**). Similarly the colour of seeds may be yellow or green. The yellow factor (**Y**) is dominant to wrinkled (**y**). When plants homozygous for round seeds are crossed with those having wrinkled seeds, the F_2 generation showed a ratio of 3.1 between round and wrinkled. A similar F_2 ratio was obtained when seeds with yellow factor were crossed with seeds with green factor.

Mendel's test cross — hybyrid round, yellow peas × pure-breeding wrinkled, green peas

	round, yellow peas	round, green peas	wrinkled, yellow peas	wrinkled, green peas
phenotype ratio:	1 :	1 :	1 :	1

the gene/allele interpretation

parental phenotype: heterozygous round, yellow × homozygous wrinkled, green

parental genotype: RrYy × rryy

gametes: RY Ry rY ry × ry

	¼RY	¼Ry	¼rY	¼ry
1ry	¼RrYy	¼Rryy	¼rrYy	¼rryy

offspring phenotype: round, yellow peas; round, green peas; wrinkled, yellow peas; wrinkled, green peas

The offspring genotypes are shown in the Punnett diagram.

Fig. 32.5: The dihybrid testcross.

Mendel crossed a plant with *round yellow seeds* (**RRYY**) to a plant with *wrinkled green seeds* (**rryy**). All F_1 dihybrids produced *round yellow seeds* (**RrYy**). This is a case of dominance and the results were expected. But whether genes for round and wrinkled, and yellow and green would stay together when F_1 dihyrids are allowed to self-pollinate. The most suitable way to check it was to make a testcross between F_1 dihybrids and double recessive parents. A ratio of 1:1:1:1: was obtained when the cross was made. Formation of four classes of gametes suggest that genes for seed shape and seed colour are behaving independently. In addition to old combinations the F_1 dihybrids also produce new combinations Ry and rY. Thus, the factors are free to form new combinations, therefore it can be said that the factors assort independently. Knowing that dihybrid individuals form four different kinds of gametes in equal proportions, 16 possible combinations are expected. When Mendel crossed the F_1 hybrids, he obtained: .315 *round yellow* (**RY**); 108 *round green* (**Ry**); 101 *round wrinkled* (**rY**); and 32 *wrinkled green* (**ry**). Stastical analysis showed a ratio of 9:3:3:1. This is called **dihybrid ratio.**

When crosses were made for remaining five pairs of genes, they followed the same rule and produced same results, i.e., the dihybrid and testcross ratios were established.

parental phenotype: homozygous round and yellow — wrinkled and green

parental genotype: RRYY × rryy

gametes: (RY) (ry)

offspring (1) genotype: heterozygous round and wrinkled RrYy × self

gametes: (RY) (Ry) (rY) (ry) (RY) (Ry) (rY) (ry)

offspring (2) genotype:

	$\frac{1}{4}$RY	$\frac{1}{4}$Ry	$\frac{1}{4}$rY	$\frac{1}{4}$ry
$\frac{1}{4}$RY	$\frac{1}{16}$RRYY round, yellow	$\frac{1}{16}$RRYy round, yellow	$\frac{1}{16}$RrYY round, yellow	$\frac{1}{16}$RrYy round, yellow
$\frac{1}{4}$Ry	RRYy [illegible]	$\frac{1}{16}$RRyy round, green	$\frac{1}{16}$RrYy round, yellow	$\frac{1}{16}$Rryy round, green
$\frac{1}{4}$rY	[illegible]	$\frac{1}{16}$RrYy round, yellow	$\frac{1}{16}$rrYY wrinkled, yellow	$\frac{1}{16}$rrYy wrinkled, yellow
$\frac{1}{4}$ry	$\frac{1}{16}$Rr[illegible] round, yellow	$\frac{1}{16}$Rryy round, green	$\frac{1}{16}$rrYy wrinkled, yellow	$\frac{1}{16}$rryy wrinkled, green

progeny phenotype:	round, yellow	round, green	wrinkled, yellow	wrinkled, green
phenotype ratio:	9	3	3	1
progeny genotypes:	RRYY RRYy RrYY RrYy	RRyy Rryy	rrYY rrYy	rryy

Fig. 32.6: The gene/allele interpretation of the dihybrid cross.

Law of Independent Assortment

These results led Mendel to formulate his second law of inheritance, the **law of independent assortment**. It states that:

"The members of one pair of factors (genes) segregate independently of members of other pair at the time of gamete formation --- as pair Rr segregated independent of Yy"

TRIHYBRID CROSS

Mendel demonstrated that processes of segregation and independent assortment apply to three pairs of contrasting traits as well. Such a cross was named **trihybrid cross.**

Example

When **RRYYTT**(round, yellow and tall) individuals were crossed to **rryytt** (wrinkled, green and dwarf) individuals. All F_1 were dominant for traits: shape and colour, and height of plant, i.e., RrYyTt. When F_1 hybrids were allowed to self-pollinate. Each F_1 parent formed 8 gametes, RYT, RYt, RyT, Ryt, rYT, rYt, ryT, ryt, as a result of independent assortment of three pairs of genes. F_2 progeny consisted of 64 individuals in the ratio of **27:9:9:9:3:3:3:1**. This ratio is called trihybrid ratio. The **test cross** ratio for a trihybrid cross is **1:1:1:1:1:1:1:1**.

Determination of Gametes

An easy way to determine different gametes is **branching method**. This helps in determination of gametes in a dihybrid and a trihybrid cross.

Let us consider a dihybrid cross between round and yellow and wrinkled and green individuals. The genes for traits: seed shape and colour, segregate from one another as well as assort independentl of each other. Therefore, gene for round has an equal chance of entering a gamete with either the gene for yellow or the one for green. The same is true for gene for wrinkled seed. The branching method is also used to derive the different kinds of gametes formed by the trihybrid.

Prediction of Offspring in a Cross

The knowledge of independent assortment enable us to predict the kinds of offspring in any crosses involving two or more pairs of factors on separate chromosomes. Punnet square method help us to visualize it. It also provides a picture of different genotypes, but when more than two pairs of genes are involved in a cross, Punnet square method becomes cumbersome, because a large number of squares is required, for example 64 squares are needed in a trihybrid cross since each trihybrid forms eight different classes of gametes.

Forked-Line Method

A more simple and easy method called **forked-line method** can be used to predict the kinds of offspring. The forked-line method is a much more direct and time saving method than the Punnet square and accomplishes the same thing in telling us the different phenotypes to be expected and their frequencies.

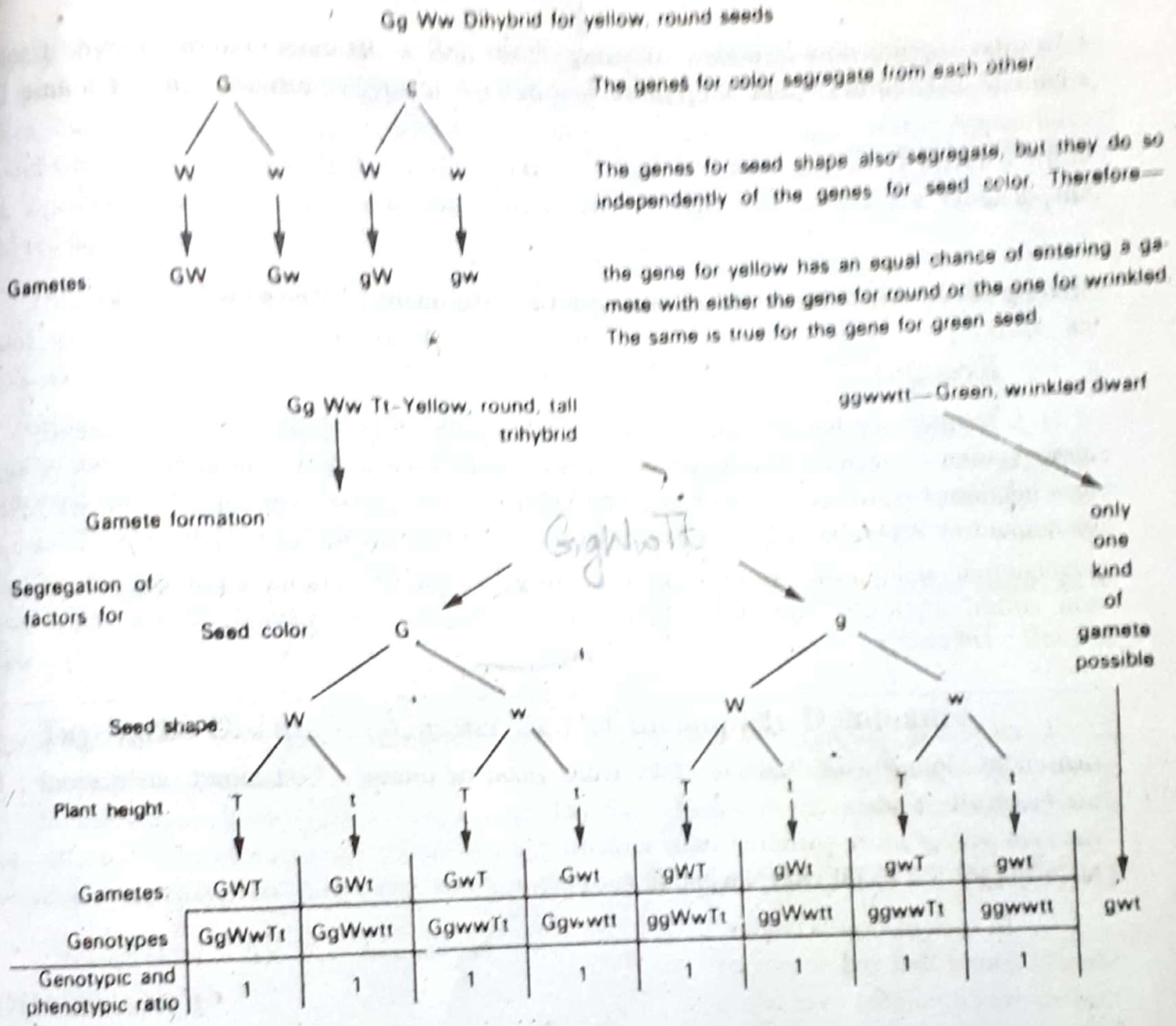

Fig. 32.7: ***Branching Method*: One way to determine the different types of gametes which can be formed by a dihybrid and a trihybrid.**

Law of Probability

This fork line method is based upon law of probability established for the dihybrid cross. It states that:

"if we know the chances of two indépendent events happening separately, the chance of two occurring together is the product of separate probabilities"

Example 1 --- A Dihybrid Cross

A dihybrid cross (**RrYy x RrYy**) can treated as if it were two monohybrid crosses, i.e., a cross between one kind (**Rr x Rr**) and a cross between the other kind (**Yy x Yy**). Our knowledge of monohybrid crosses tells us that in the first case we would expect 3/4 round and 1/4 wrinkled and in the second cross 3/4 yellow and 1/4 green. We know that the ratio is an expression of probability, i.e., the chance of a certain event taking place at one time. So in this dihybrid cross the separate chances for round vs wrinkled at any one time and similarly for the independent occurrences of yellow vs green. Therefore, to determine the different combinations which are possible in a dihybrid cross we can multiply together the separate probabilities:

3/4 *round* : 1/4 *wrinkled*

3/4 *yellow* : 1/4 *green*

9/16 *round yellow* : 3/16 *wrinkled yellow* : 3/16 *round green* : 1/16 *wrinkled green*

Example 2 ---- A Trihybrid Cross

Suppose we are following three traits in a cross: **seed shape** (round or wrinkled), **seed colour** (yellow or green), and **height of plants** (tall or dwarf); and want to know the result of a cross between two trihybrids: **RrYyTtX RrYyTt**. Let us consider these as separate monohybrid crosses and multiply the result of above dihybrid cross by the chances for tall vs dwarf:

9/16 *round yellow* : 3/16 *wrinkled yellow* : 3/16 *round green* : 1/16 *wrinlked green*

3/4 *tall* : 1/4 *dwarf*

27/64 *tall round yellow* : 9/64 *tall wrinled yellow* : 9/64 *tall round green* : 3/64 *tall wrinkled green* : 9/64 *dwarf round yellow* : 3/64 *dwarf wrinkled yellow* : 3/64 *dwarf round green* : 1/64 *dwarf wrinkled green*

INCOMPLETE DOMINANCE

In case of flower colour, Mendel found that red colour of flowers was dominant over white colour of flowers. The white coloured flowers reappeared in F_2 following Mendel's principle of seggregation.

However, later a deviation to this rule was observed in a plant species commonly called **four-o'clock** (*Mirabilis jalapa*). The plant produces flowers with red and white colours. When pure-breeding **red (R) flowered plants** were crossed with pure-breeding **white (r) flowered plants**, the F_1 plants were **pink (Rr) flowered**. Appearance of pink flowers, an intermediate shade between red and white, provided relief to those who believe in blending inheritance.

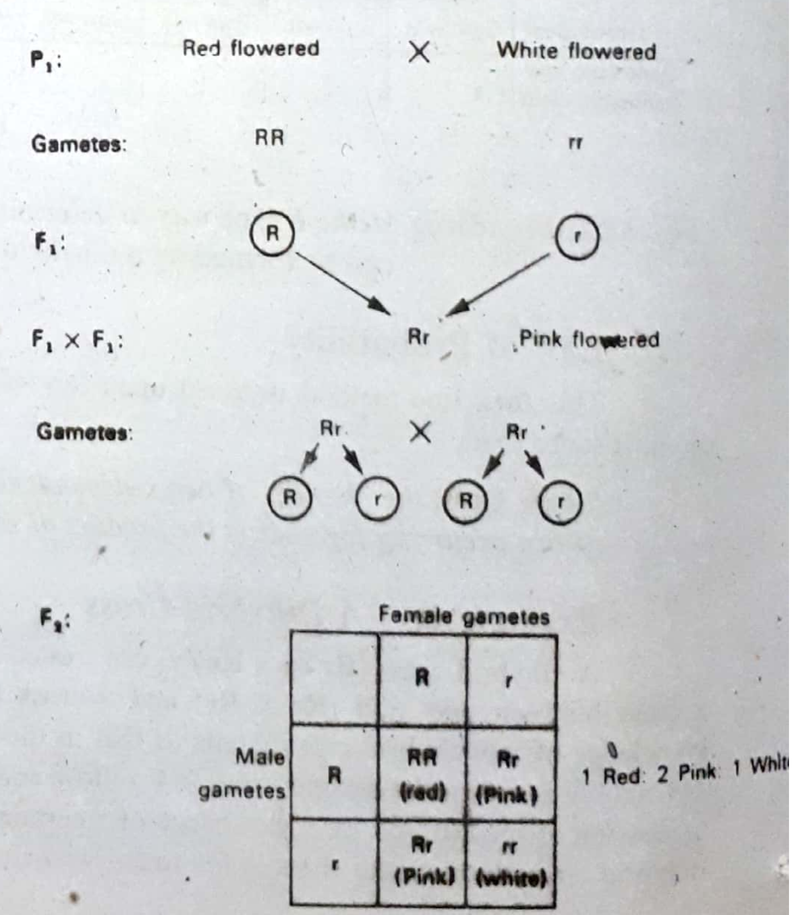

Fig. 32.8: Incomplete dominance in the four-o'clock.

But when F_1 hybrids were crossed (Rr x Rr), the F_2 generation showed a phenotypic ratio of **1 red : 2 pink : 1 white,** instead of typical 3:1 ratio. Although the ratio contradicted Mendel's principles, but it provides a strong support to the concept of particulate inheritance. Appearance of red and white factors (genes) in F_2 indicate that these have not been altered or blended while present together in the pink flowered individuals. The F_2 reds and whites are same as the parental red and white.

This was declared a case of **incomplete dominance**. In this case neither of the gene is dominant to the other. Each expresses itself in the presence of its allele to produce an intermediate effect.

Another important aspect of incomplete dominance is that phenotypic ratio (1:2:1) is the same as genotypic ratio (1 homozygous dominant : 2 heterozygous dominant : 1 homozygous recessive) resulting from cross when two monohybrids are crossed. So when dominance is incomplete, a cross of two monohybrids (Rr x Rr) gives a phenotypic ratio of 1:2:1 which is identical to the genotypic ratio. The heterozygous individual shows incomplete dominance. Whereas when the dominance is complete the phenotypic (3:1) and genotypic ratios are complete.

Tay-Sachs Disease --- Another case of Incomplete Dominance

Incomplete dominance is found in many other cases as well, for example in human beings, the individuals homozygous recessive for Tay-Sach disease (a biochemical disorder) are severely affected. The children die before the age of three after suffering from severe nervous degeneration. Heterozygotes suffer less and homozygous dominant individuals do not suffer at all.

CODOMINANCE

In case of blood types in humans, both genes (A & B) produce an effect in a heterozygous individual. This is called **codominance**. The genes which govern A and B blood types are alleles. Each control the formation of a different red blood cell protein or antigen. **Antigen a** in case of person having blood group A and **antigen b** in individuals with blood group B. Neither gene is dominant to the other. The heterozygous individuals with blood group AB contains both antigens a and b. Both proteins ae detected in equal amounts in the red cells.

The cases of incomplete dominance and codominance suggest that the dominance is not universal and absolute.