06 Genetics

Scientific basis of Mendel's experiments

Mendelian Heredity (Mendelism)

Principles in heredity were first formulated by an Austrian Augustinian monk named Gregor Mendel, who is now considered the father of modern genetics. Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments.

His experiments were conducted decades before the concept of chromosomes. The later discovery of chromosomes as the carriers of genetic units supported Mendel's two basic laws of genetics which are now known as **Mendelism.**

Vocabulary in genetics

There are numerous heritable variations among individuals of a population such as brown, green, or blue eyes or black, brown, or blond hair in human population. A heritable feature that varies among individuals of a population, such as hair colour or eye colour is called a **character** in genetics. These heritable variants of a character in an organism, such as brown or blond hair or blue, brown or black eye colour in human are called as **traits**. These traits are transmitted from parents to offspring. Observable traits of an organism is known as phenotype.

Mendel has described about 'heritable factors' in explaining his experimental results. These heritable factors are identified to be **genes** in modern genetics. **Gene** is the basic unit by which genetic information is passed from parent to offspring. It is a DNA sequence residing usually at a specific **locus** on a particular chromosome and contributes to the development of one or more traits by coding for specific proteins or peptides. Locus (Loci in plural) is a fixed position on a chromosome.

There are alternative versions of genes which are called **alleles.** Alleles reside on the same locus of different chromosomes. Alleles vary in their sequence of nucleotides. This change can affect the function of the protein encoded by the gene and thus the phenotype of the organism. Each diploid organism has at least two copies of each gene, residing on the chromosomes received by the two parents. These copies could be identical or could differ from one another. The condition of

having two identical alleles for a given gene is known as **homozygous** state. Alternatively, having two different alleles for a given gene is referred to as **heterozygous** state.

Phenotype is brought about by the interaction between genotype of the individual with its environment. The genetic make up, or set of alleles, of an organism is known as its **genotype.** An individual's genotype could be either homozygous or heterozygous with respect to a given gene.

At heterozygous state, the allele which determines the organism's phenotype by masking the expression of the other is referred to as the **dominant allele**. The trait produced by the dominant allele is known as the **dominant trait.** The allele which does not exhibit any noticeable effect on the organism's phenotype at heterozygous state is referred as the recessive allele. The trait hidden on the recessive allele is the **recessive trait.** However, they express their trait when they exist in homozygous state.

Mendel tracked only those characters that occurred in two distinct, contrasting phenotypic forms, such as tall stem length vs. short stem length or purple flower colour vs white flower colour. Such traits are referred to as **contrasting traits.**

Mendel used only the **pure breeding** (sometimes called **true breeding**) varieties for his experiments. Pure breeding plants are obtained by self-pollinating over many generations, producing only the same variety as the parent plant. These uniform lines produced from self-fertilization of pure breeding varieties over many generations are called **pure lines.**
During his experiments, Mendel cross-pollinated pure-breeding garden pea plant varieties which shows contrasting **traits**. For example, purple-flowered plants were cross bred with white-flowered plants. Mating or crossing of two pure-breeding varieties with contrasting **traits** is called **hybridization**. Parental generation is referred to as the **P generation** (parental generation). Plant progeny resulted from these hybridization events are referred to as **F_1 generation** (First Filial generation, the word filial from the Latin word for "son"). The progeny that results from the self or cross pollination between these F1 generaion plants are known as **F_2 generation** (Second Filial generation).

An organism that is heterozygous with respect to a single gene of intrest resulting from a cross between parents having homozygous condition for different alleles of specific gene is referred to as a '**monohybrid**'. Breeding experiment conducted between two organisms with heterozygous condition for a specific character is referred to as **'monohybrid cross'.**
An organism that is heterozygous with respect to two genes of intrest resulting from a cross between

parents having homozygous conditions for different alleles of two specific genes is referred to as a **'dihybrid'**. Breeding experiment conducted between two organisms with heterozygous conditions for two specific characters is referred to as a **'dihybrid cross'.**

Breeding an organism having unknown genotype for a specific dominant trait with an organism having homozygous recessive condition for the same specific trait is called a **test cross.** This is usually done to reveal the unknown genotypes for specific dominant traits in an organism.

Monohybrid Cross

Mendel derived his first law of inheritance by following only a single character in one breeding experiment, such as flower colour. IIe started by crossing pure breeding parents with contrasting **traits**. All the F_1 progeny produced from pure breeding parents are **monohybrids,** meaning that they are heterozygous for the particular character being followed in the cross.

F_1 hybrid pea plants were then self- or cross-pollinated and F_2 generation was produced to explore the traits resulting from a monohybrid cross (**Figure 6.1**).

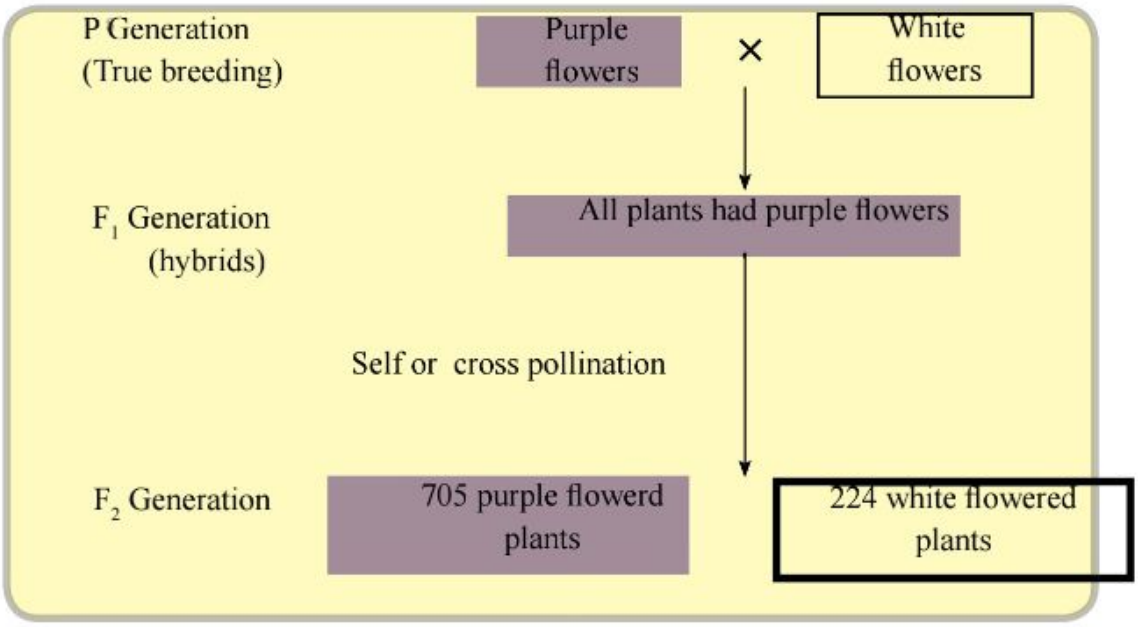

Figure 6.1: Mendel's experiment to investigate the inheritance patterns of a single character over two generations

Mendel, during his experiment, crossed pure breeding purple flowered plants and white flowered plants. Then he allowed the resulting F_1 hybrids to self and cross-pollinate with other F_1 hybrids. Finally, he observed the F_2 generation plants for the colour of the flowers.

During his observation, all F_1 plants produced purple colour flowers. However, in the F_2 generation, both purple and white flowered plants appeared in a ratio of approximately 3: 1.

Among the heterozygote resulted in the F_1 generation, the "heritable factor" responsible for producing white flowers were suppressed in presence of the "heritable factor" that produces purple flowers. As a result, the heterozygotes were all producing purple colour flowers. Therefore, the 'heritable factor' for purple flower colour is dominant to the white (dominant trait). Accordingly, the factor for white flower colour is referred as the recessive trait.

Mendel observed that the same pattern of inheritance consistently occured in six other characters; position of the flower, colour of the seed, shape of the seed, shape of the pod, colour of the pod and the length of the stem.

Mendel's first law of inheritance: The law of segregation

Mendel's first law was put forward to explain the 3:1 inheritance pattern observed among the F_2 offspring in his monohybrid experiments using Pea plants.

As per his hypothesis, each 'heritable character' is determined by two "heritable factors" which are known as alleles. During the formation of gametes, the alleles for a 'heritable character' are separated and get in to each of the gametes formed. This is now known as **Mendel's law of segregation or Mendel's first law in inheritance.**

Analyzing genotype and phenotype ratios using Punett square

In Pea plants, Mendel observed two different **traits** based on stem lengths; tall and dwarf. For his experiments, pure breeding tall and dwarf Pea plants were selected for cross pollination. Thereafter the F_1 generation was self pollinated in order to obtain F_2 generation.

During self pollination of F_1 hybrids, gametes carrying different alleles fuse randomly. Such rdom fusion of gametes produces zygotes with four genetic combinations. A Punnett square can be used to illustrate these genetic combinations. A Punnett square is a graphical representation of the possible geneotypes of an offspring arising from a particular cross or breeding event. An example is given figure 6.2.

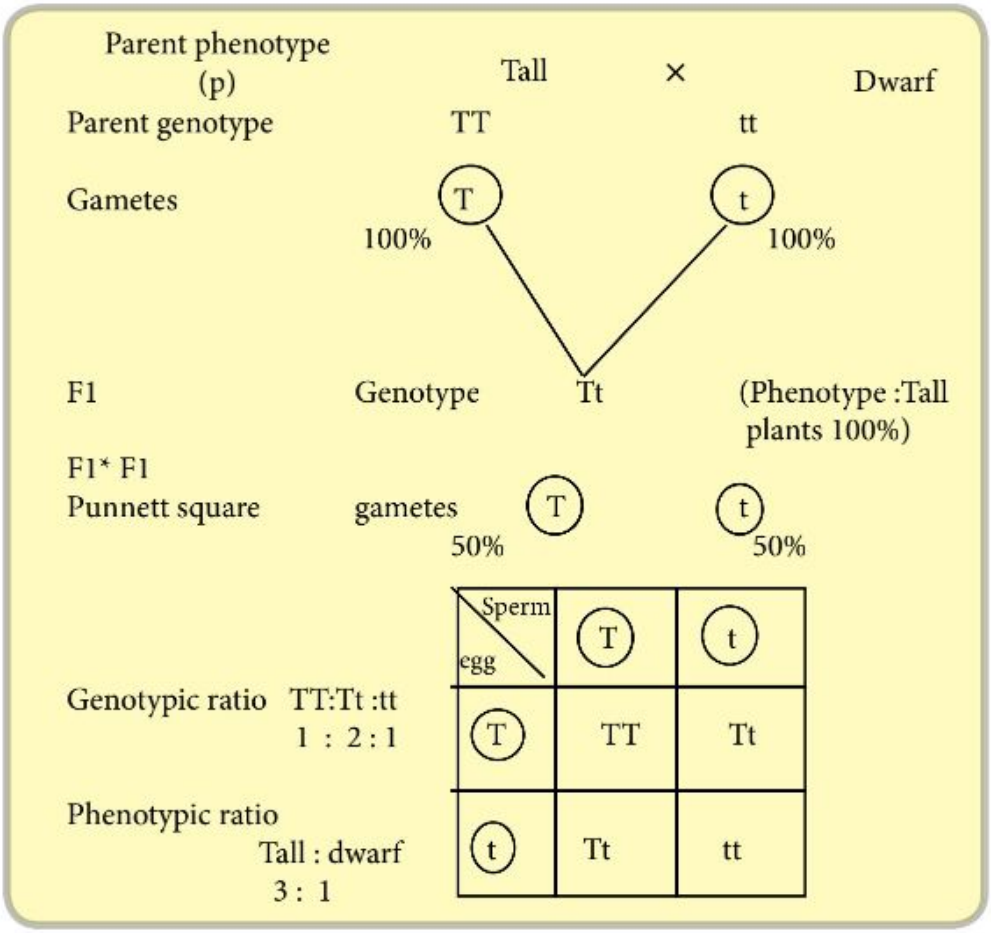

Figure 6.2: Mendel's law of seggregation shown with the aid of the punnett square

Dihybrid Cross

Mendel identified his second law of inheritance by following two characters at the same time, using **dihybrid crosses.** A cross between two homozygous (true breeding) organisms with contrasting traits for two specific characters being followed is known as a **dihybrid cross.** The aim of the Mendel's dihtybrid cross experiment was to find out whether the alleles for one character assort into gametes dependently or independently of the alleles of the other character.

Mendel crossed a true-breeding plant with yellow-round seeds with a true breeding plant with green-wrinkled seeds (Figure 6.3). The cross produced dihybrid F_1 plants, all of which have yellow-round seeds. As shown by the monohybrid crosses, the allele for yellow seeds is dominant (Y) over the allele for green seeds (y) which is recessive. likewise, the allele for round seed is dominant (R), and the allele for wrinkled seed is recessive (r). The F_1 hybrids, are heterozygous for the two characters being followed in the cross (YyRr). The cross between F_1 dihybrids produced the F_2 generation.

This lead towards two alternative hypothesis for inheritance which predict different phenotypic ratios as shown in **Figure 6.3.**

1. The two characters could be transmitted from parents to offspring as a package. The dominant Y and R alleles or the recessive y and r alleles are passed together, generation after generation. This is called **dependent assortment** of alleles.
   According to this hypothesis only two types of gametes are possible; i.e. YR and yr. Thus the phenotypic ratio of the F_2 generation would be similar to that of a monohybrid cross (3:1)
2. The two characters (seed colour and seed shape) could be transmitted from parents to offspring independent of each other i.e. Y allele could be passed either with R or r allele vice versa. This is called **independent assortment** of alleles.
   This hypothesis predicts four different allelic combinations for a bi-allelic locus and thus four different types of gametes from F_1 generation; i.e, YR, Yr, yR and yr. According to this, both male and female gametes have four possibilities for each. Therefore, during the union of male and female gametes, there are 16 (4 × 4) equally probable ways in which the alleles can combine to produce the F_2 generation. As shown in Figure 3, these combinations would give rise to four different phenotypes with a ratio of 9:3:3:1 (nine Yellow round to three Green-round to three Yellow-wrinkled to one Green-wrinkled).

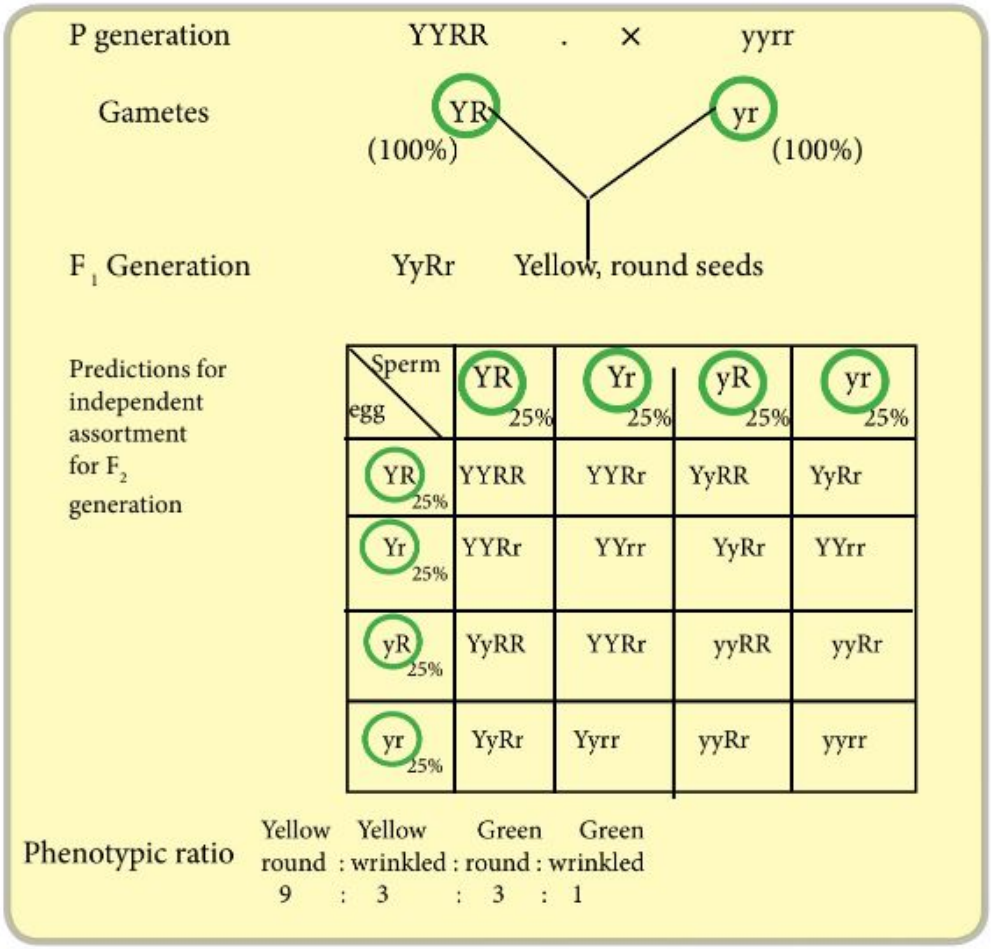

Figure 6.3: Alternative forms of inheritance patterns possible in a dihybrid cross

Mendel's experiment produced the four distinct phenotypes (Yellow-round, Green-round, Yellow-wrinkled and Green-wrinkled seeds) predicted in the second alternative hypothesis in the ratio of 9:3:3:1. This showed that, the alleles responsible for each trait assorted independently of those of the other.

Mendel's second law of inheritance (The law of independent assortment)

Based on these experiments, Mendel put forward his **second law of inheritance - the law of independent assortment.** The law states that, alleles separate and pair up independently during the formation of gametes. As a result of that, two or more genes assort independently irrespective of the other.

However, according to current knowledge, this condition applies to two circumstances only;

- To genes located on different chromosomes (genes on non-homologous chromosomes)
- To genes located far apart on the same chromosome

Success of Mendel's experiments

Mendel followed a scientific approach in conducting his experiments. The following features in his experiments helped him to unravel the two fundamental principles of heredity.

- Mendel carried out thousands of genetic crosses of any given kind. This allowed his results to closely resemble the probability predictions. Usually, the larger the sample size, the closer the results to the value predicted based on the probability.
- He kept accurate records of his results. These records helped him to trace the patterns which otherwise would go unnoticed.
- He usually followed up each cross for at least two offspring generations (F_1 and F_2). This helped him to uncover some of the traits hidden in the F_1 generation.
- He did a quantitative analysis of the phenotypes of the resulting offspring.

Desirable properties in garden peas for genetic experiments

Garden peas (*Pisum sativum*) carry following desirable properties which makes it a suitable organism to study patterns of inheritance.

- Pea plants are available in many varieties with contrasting traits.
- The generation time is short.
- A large number of offspring is produced from each cross.
- Crossing between the plants could be strictly controlled (self/ cross pollination).

Probability laws and Mendelian inheritance

Mendel's laws of segregation and independent assortment reflect the same rules of probability that applies to tossing coins, rolling dice, and drawing cards from a deck. **Probability** measures

how likely an event is to occur out of the number of possible outcomes. It is calculated by dividing the number of events of interest by the number of total possible outcomes.

1. The probability scale ranges from 0 to 1.
   An event that is certain to occur has a probability of 1, while an event that is certain not to occur has a probability of 0.
   During allele segregation in a F_1 plant (heterozygous) of a monohybrid cross,
   -probability of each egg carrying the dominant allele = 1/2
   -probability of each egg carrying the recessive allele = 1/2

2. The probabilities of all possible outcomes for an event add up to 1.
   During allele segregation in a heterozygous F_1 plant, probability of all events (having dominant and recessive alleles) = 1/2 +1/2= 1

3. When the occurrence of an event does not affect the occurrence of another event (independent events), the probability of simultaneous occurrence of both events can be obtained by multiplying the probability of one event by the probability of the other event. This is known as the **Multiplication Rule** or **Product rule in Probability.**

   In Mendel's monohybrid crosses, for a F_2 plant to have wrinkled seeds (rr), both the egg and the sperm that come together must carry the r allele.
   The probability that the egg will have an r = 1/2
   The probability that the sperm will have an r = 1/2
   The probability of both gametes at fertilization carrying r allele = ½ x ½ = ¼

4. The probability that any one of two or more mutually exclusive events will occur is calculated by adding their individual probabilities. This is the addition rule or sum rule of probability.
   There are two possible mutually exclusive ways for producing F_2 heterozygotes.
   i. The dominant allele come from the egg and the recessive allele from the sperm; the probability of the event = ¼ (according to example in above 3rd sentence)
   ii. The recessive allele from the egg and the dominant allele from the sperm; the probability of the event = ¼ (according to example inabove 3rd sentence)
   So, the probability of getting an F_2 heterozygote = ¼ + ¼ = ½

Prediction of the inheritance patterns in multifactorial crosses

When the pattern of inheritance of two or more characters of an organism is being traced during a genetic cross, it could be called as multifactorial cross. Finding out the outcomes of a multifactorial cross through a Punnett square may be a difficult task. Therefore, applying rules of probability may be useful to predict the outcomes of a multifactorial crossing.

According to the law of seggeregation, a multifactorial cross can be considered equivalent to multiple independent monohybrid crosses occurring simultaneously.

E.g. 1: Dihybrid cross for seed color and seed shape,

Probabilities for seed color (based on the Punnett square for monohybrid cross)

Seed color	
Genotype	Probability
BB	¼
Bb	½
bb	¼

Seed shape	
Genotype	Probability
RR	¼
Rr	½
rr	¼

B: dominant allele for black colour seed b: recessive allele for brown coloured seeds

R: dominant allele for round shaped seeds r: recessive allele for wrinkled shaped seeds.

The probability of each of the genotypes in the F_2 generation can be determined by using the multiplication rule.

Probability of BbRr = ½ (probability of Bb) × ½ (probability of Rr) = 1/4

Probability of bbRr = 1/4 (bb) × 1/2 (Rr) = 1/8

Probability of bbrr = 1/4 (bb) × 1/4 (rr) = 1/16

E.g. 2: A trihybrid cross for flower color, seed color and seed shape.

Y: Dominant allele for yellow colour petals y: Recessive allele for white coloured petals

B: Dominant allele for black coloured seeds b: Recessive allele for brown coloured seeds

R: Dominant allele for rond seeds r: Recessive allele for wrinkled alleles

YyBbRr x yyBbrr

(YyBbRr): Yellow coloured petals with black round seeds

(yyBbrr): white coloured petals with black round seeds seeds

Flower color	
Genotype	Probability
YY	0
Yy	½
yy	½

Seed shape	
Genotype	Probability
BB	¼
Bb	½
bb	¼

Shape of the seeds	
Genotype	Probability
RR	0
Rr	½
rr	½

Assume that unknown pea plant is having genotype of TT

The above crossing has resulted 640 plants in the F1 generation. Determine the number of plants exhibits dominant phenotype for at least two characters.

1. Possible genotypes with their individual probabilities for above conditions

YyBBRr: ½ (probability of Yy) X ¼ (BB) X ½ (Rr)= 1/16

YyBbRr: ½ X ½ X1/2 = 1/8

YyBBrr ½ X ¼ X ½ = 1/16

YyBbrr ½ X ½ X ½ = 1/8

YybbRr ½ X ¼ X 1/2 = 1/16

yyBbRr ½ X ½ X ½ = 1/8

yyBBRr ½ X ¼ X 1/2 = 1/16

2. Probability of exhibiting at least two dominant characters= 1/16 + 1/8 + 1/16 + 1/8 + 1/16 + 1/8 +1/16 = 10/16= 5/8

3. Number of plants expected to exhibit at least two dominant characters = 5/8 × 640= 400 plants

The testcross

This is a deliberate breeding process performed in order to determine the unknown genotypes. The genotype of an individual showing dominant trait may be due either to double dominant genotype or to heterozygous status. This involves the crossing of an organism having unknown genotype for a selected phenotype along with another organism from same species having homozygous recessive condition for same character.

Testcross performed during the monohybrid cross is called as monohybrid testcross. On the other hand, test cross performed during the dihybrid cross is called as dihybrid testcross.

Monohybrid testcross

Let's consider an example for monohybrid testcross. In this example we want to know the genotype of the given tall pea plants. In order to do that, we will cross the tall pea plant with the dwarf pea plant. Since dwarf is a recessive trait the genotype of it will be tt.

There may be two possible genotypes for tall pea plants;

1. TT
2. Tt

Assume that unknown pea plant is having genotype of TT

Cross between TT and tt will result the following.

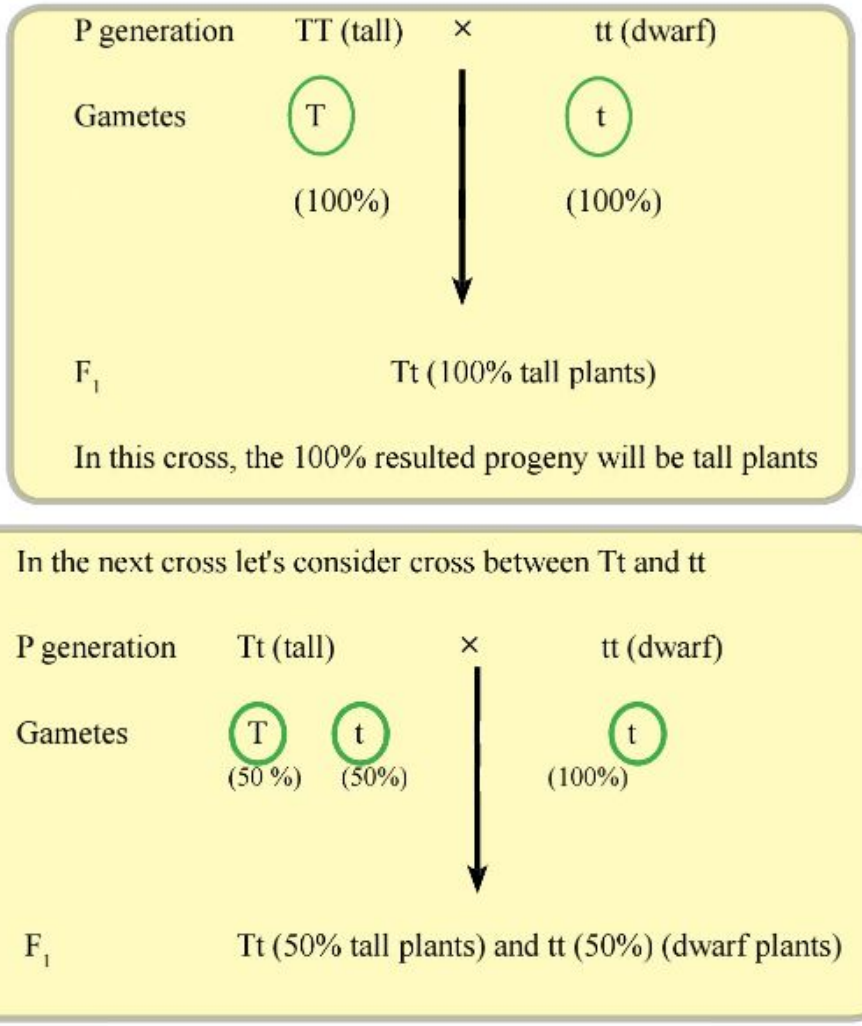

Figure 6.4:Two possible outcomes of a monohybrid test cross

Dihybrid testcross

When an individual with dominant traits for two characters (e.g. RrBb) is crossed with a pure recessive for both characters (wwbb), it is known as a **dihybrid testcross.**

For example, let's consider a cross between plants having red coloured flower petals with black coloured seeds and white coloured flower petals with brown coloured seeds.

The possible genotypes for red coloured petals with black coloured seeds are RrBb/ RRBb/RrBB/ RRBB and for white coloured petals with brown coloured seeds is wwbb.

If the unknown phenotype is RrBb;

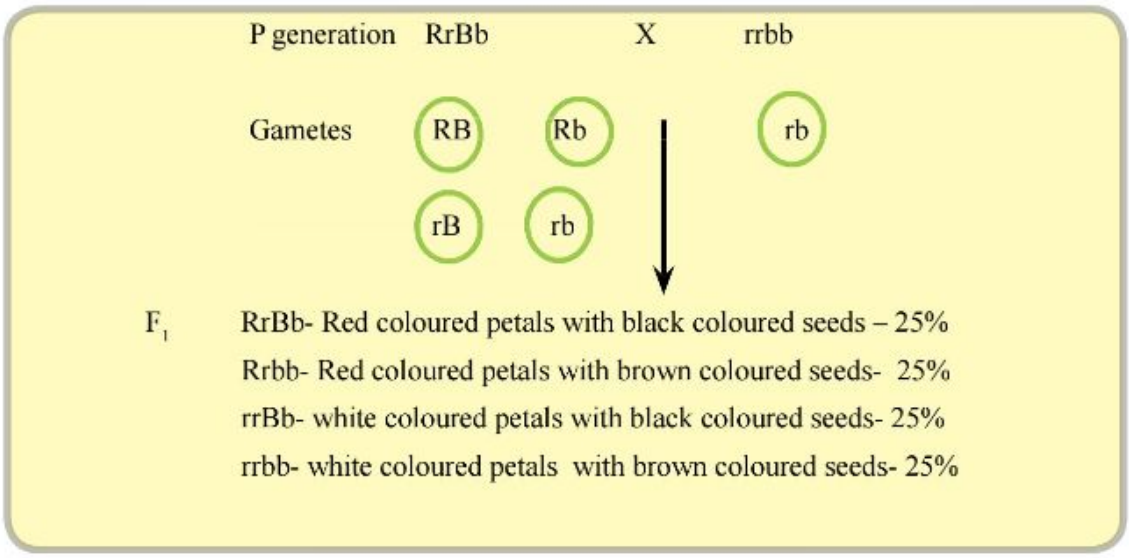

Figure 6.5: (a) Possible outcomes of a dihybrid test cross

In this example, above four phenotypes are possible with 1:1:1:1 ratio.

If the unknown phenotype has the RRBb genotype, the resulting dihybrid cross will produce following phenotypes;

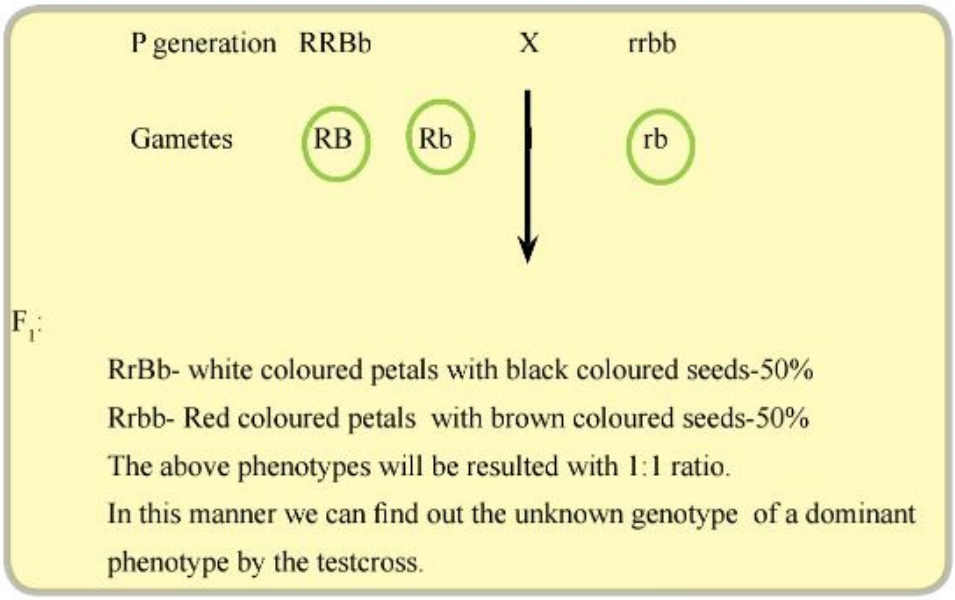

Figure 6.5: (b) Possible outcomes of a dihybrid test cross

Patterns of inheritance of Mendelian characteristics in humans

Common Mendelian characters

Many human traits follow Mendelian patterns of inheritance. Some common examples are stated below;

Attached or detached earlobe:

The extent to which the earlobe is attached to the head is inherited in the Mendelian pattern. The attached earlobe is a recessive trait. Presence of both copies of the recessive allele (homozygous recessive condition) for ear lobe attachemnt would result in attached ear lobe.

Widow's peak:

The pointed contour of the hairline on the forehead is known as Widow's peak. It is due to a dominant allele, W. Therefore, all individuals who lack a widow's peak must be homozygous recessive (ww).

Dimples on cheek:

Cheek dimples are a genetically transmitted trait found in the muscle of the cheek.

When a person smiles, the shorter muscle on the face pulls up the facial skin. This, in turn, creates a slight depression in the skin, which is called dimple. Dimples often occur on both the cheeks. A single dimple on one cheek is a rare phenomenon. Dimple is a dominant trait and inherited in Mendelian fashion.

Bent thumb (Hitchhiker's thumb) and Straight thumb

Hitchhiker's thumbs is a condition where thumb bend backwards while stretching due to the hyper extensibility of interphalangeal joints. Having the dominant 'S' allele would produce the dominant phenotype of straight thumb. The absence of the dominant alleles would allow the thumb to bend.

Rolling or non-rolling tongue

The ability to roll the lateral edges of the tongue upwards into a tube is known as tongue rolling. The tongue's intrinsic muscles allow some people to form their tongues into specific shapes. Rolling the tongue into a tube shape is a dominant trait with simple Mendelian inheritance.

Pedigree analysis

Diagramatic representation of the inheritence of a particular trait within a given family tree, is called pedigree chart. It is constructed by collecting data for many generations within a given family, so that the pattern of inheritence can be understood.

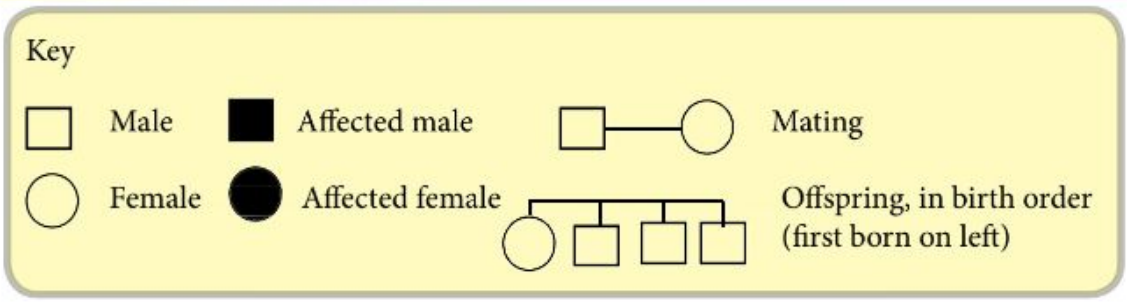

Figure 6.6: Standard Pedigree Symbols

Common Mendelian characteristics in humans analysed with pedigree charts

Widow's peak:

The inheritence of the trait, widow's peak, over three genertations in a particular family is represented in the pedigree chart given below. As shown in figure 6.7, only one grand parent had widow's peak, out of the two pairs. Since widow's peak is a dominant character, grand parents without widow's peak should be homozygous recessive (ww) for the trait. In the next generation some individuals showed the widow's peak, while the others did not. The two grand parents, who express the widow's peak should be heterozygous (Ww) for the condition . Likewise, the two parents of the third generation, who are showing widow's peak should be heterozygous for it, as one of both their parents (1st generation) are homozygous recessive (ww). the third generation with widow's peak can carry either WW or Ww geneotype, as both their parents are having the trait.

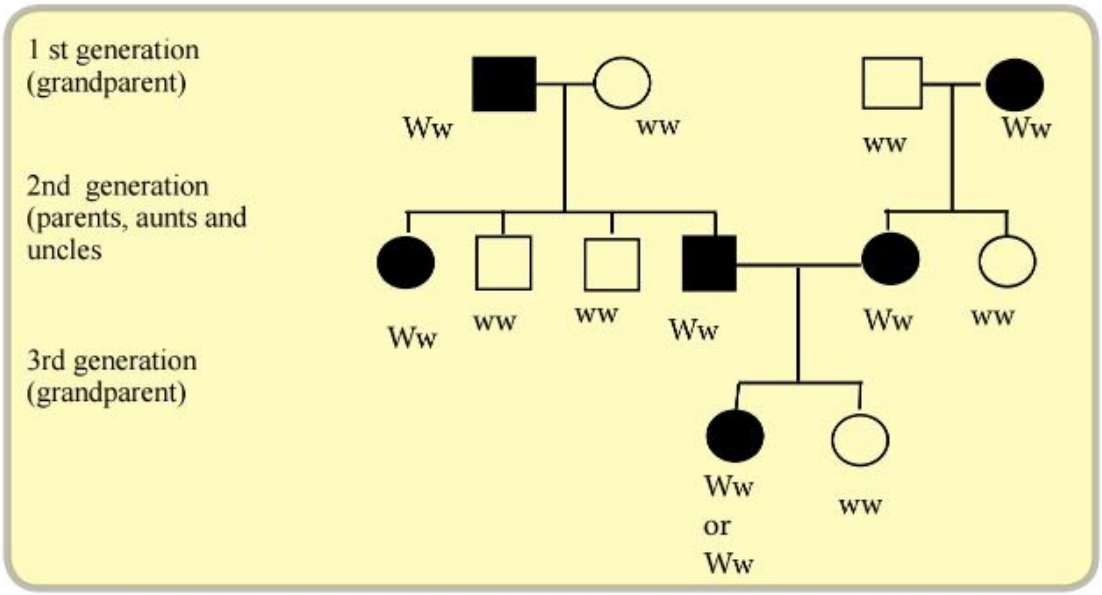

Figure 6.7: Inheritance of Widow's peak

Attached ear lobe

Attached ear lobe, as explained earlier, is a recessively inherited character. In the pedigree chart given below, the inheritence of the tarit is analysed in the same family that was used to study the widow's peak. The dominant allele, which is causing the free ear lobe is denoted with 'F' while recessive allele is denoted with 'f'.

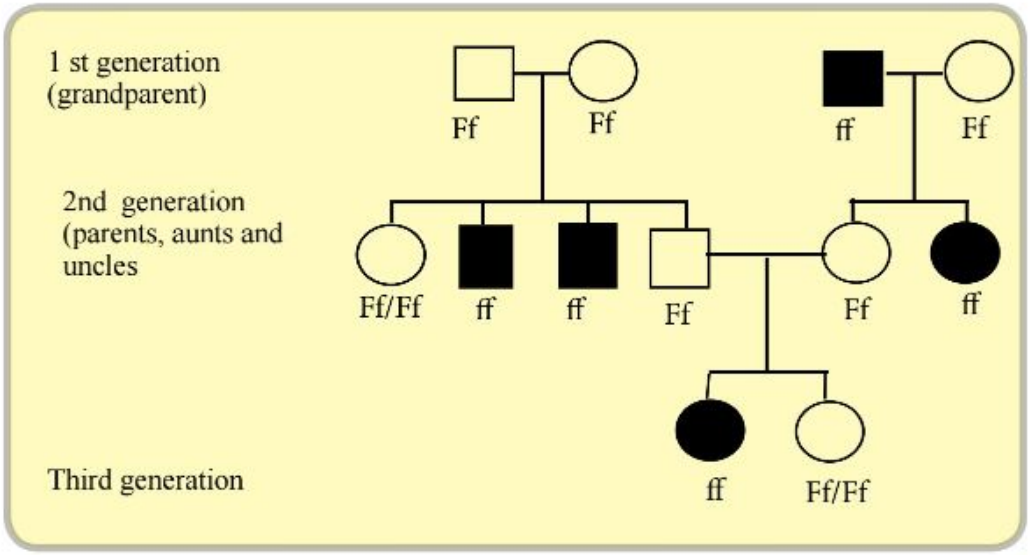

Figure 6.8: The pedigrce chart showing the inheritance pattern of attached lobe

In the first generation, both parents lacking the attachment of ear lobes, resulted mixed progenies with attached and free ear lobes. This tells us that those two parents were heterozygous (Ff), and among the progeny, two males with attached ear lobes carry homozygous recessive alleles (ff) and a male and female with free ear lobe may carry heterozygous (Ff) or homozygous dominant alleles (FF or Ff). Another cross happened at the first generation between a male with attached ear lobe and a female with free ear lobe. This resulted one daughter with attached ear lobe. Therefore, she must possess ff and the other may be FF or Ff. A male from one family and a female from another family, at the second generation expressing free ear lobe phenotype, had resulted progeny having two females, one with attached ear lobes and the other with free ear lobes for the third generation. Therefore, the second generation male and female crossed with free year lobes must be Ff. Third generation female with attached ear lobes must carry ff genotype and the other may carry either FF or Ff.

The probability that another child from the same family will have attached earlobes could be calculated using a monohybrid cross (Ff X Ff). Since homozygous recessive (ff) genotype is causing the condition, the probability is 1⁄4 for each child.

The chance that a child born to the family will have both the widow's peak and attached earlobes can be calculated using probability rules. Assuming that the alleles corresponding to the two

characters are on different chromosomes, the two pairs of alleles assort independently in this dihybrid cross (WwFf X WwFf).

Thus according to multiplication rule,

Chance of having both widow's peak (WWff/Wwff)

and attached ear lobe = Chance of widow's peak x chance of attached earlo be

= 3/4 x 1/4

= 3/16

Non-Mendelian inheritance

Non-Mendelian inheritance refers to inheritance patterns in which traits do not segregate in accordance with ***Mendel's*** laws of inheritance. Phenotypes that do not appear in ratios predicted by Mendelian genetics are the indicators of Non-Mendelian inheritance.

Examples for non-mendelian patterns

- when alleles are not completely dominant or recessive (incomplete dominance and co-dominance),
- when a particular gene has more than two alleles (polyallelism)
- when a single gene produces multiple phenotypes (pleiotropy)
- sometimes two or more genes are involved in determining a particular phenotype (epistasis and polygenic inheritance)
- gene linkage
- genes which are located in sex chromosomes exhibit a different pattern of inheritance in males and females due to the unequal distribution of genes in their sex chromosomes.

Incomplete dominance

The phenomenon of dominant allele completely masking the recessive phenotype, resulting similar phenotypes for both homozygous dominant zygote as well as heterozygous zygote is called **complete dominance.**

On the other hand, at the heterozygous state, the phenomenon of expressing blend phenotypes from both alleles is called **incomplete dominance.** Degree of expression of each allele in the blend phenotypes may vary based on the nature of the alleles.

In *Mirabilis jalapa* (Four o'clock plant), there are several types of flower colours. When red flowered plants are crossed with white flowered plants, all the F_1 hybrids (heterozygotes) have pink flowers (Figure 6.8). This third, intermediate phenotype results from flowers of the heterozygotes having less red pigment than the red homozygotes.

When these F_1 pink flowers are self-pollinated or crossed among themselves to raise F_2 generation, they produce red (C^{RR}), pink (C^{RW}) and white (C^{WW}) flowers giving 1:2:1 ratio. This phenotypic ratio is identical with genotypic ratio because heterozygotes are phenotypically intermediate between two homozygous types.

Note: Since, neither allele is dominant, instead of upper- and lowercase letters, a superscript is used to indicate the trait; i.e. C^R for red colour and C^W for white colour.

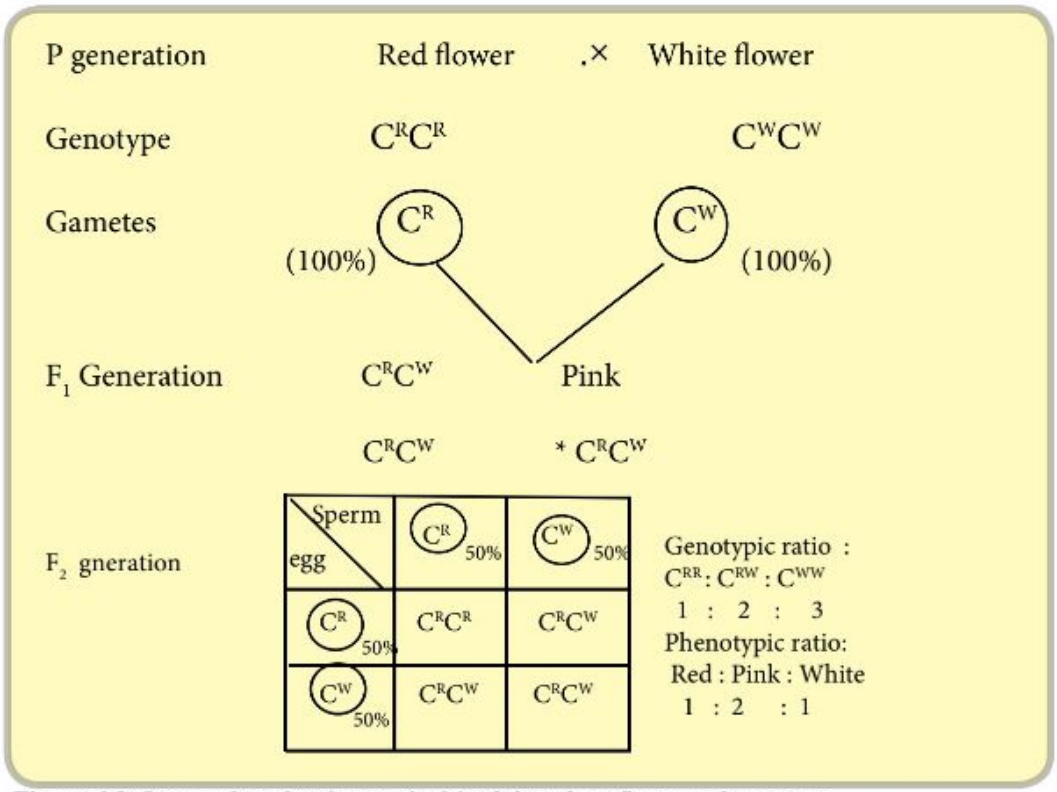

Figure 6.9: Incomplete dominance in *Mirabilis jalapa* flower colour gene

Codominance

In certain traits, at heterozygote state, expression of both alleles contributes equally to the phenotype. Such phenomenon is called **co-dominance.**

For example, a person with AB blood group type has both A and B carbohydrates on the surface of red blood cells at the same time. The two carbohydrates are added to the surface of RBC by enzymes encoded by the I^A and I^B alleles of a single gene. A heterozygous individual would express both carbohydrates (I^AI^B) in an equal manner.

Likewise,

Homozygotes for I^A allele (I^AI^A) will carry only A carbohydrate on RBC.

Homozygotes for I^B allele (I^BI^B) will carry only B carbohydrates on RBC.

As shown below, the F_1 progeny resulting from the mating of two homozygous individuals for each allele would consist of individuals only of AB blood group. F_2 generation produced by the possible mating with in F_1 progeny or heterozygous individuals for the AB alleles, would produce the three phenotypes, blood type A: AB: B at a ratio of 1:2:1.

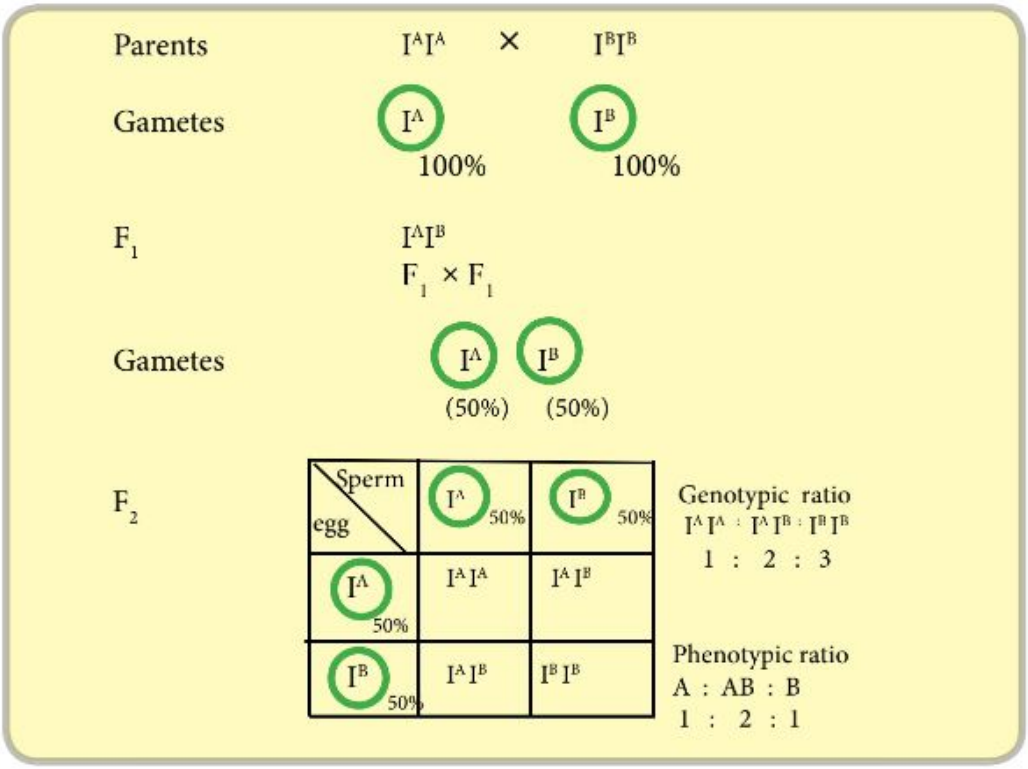

Figure 6.10: Codominance in ABO blood grouping

As shown, the F_2 phenotype ratios in both incomplete dominance and co-dominance are 1:2:1. Hence the two phenomena cannot be differentiated using F_2 phenotypic ratios.

The key to differentiate between incomplete dominance and co-dominance is that in incomplete dominance F_1 generation shows a different phenotype than both the parents whereas in co-dominance F_1 generation shows both the parental traits together.

Polyallelism (Multiple alleles)

Polyallelism refers to the presence of multiple alleles for a single genetic locus, a phenomenon where certain traits are determined by the combination of more than two types of alleles.
E.g. there are three alleles called $\mathbf{I^A}$, $\mathbf{I^B}$, and **i** for a single genetic locus which at different combinations determine ABO blood groups in humans.

In any diploid individual there are only two of the several alleles are present.

As mentioned earlier, the alleles $\mathbf{I^A}$ and $\mathbf{I^B}$ code for enzymes that add A and B carbohydrates to the surface of red blood cells. The two alleles are in a co-dominant relationship. However, the allele 'i' results in lack of these carbohydrates on the red cell surface and it is recessive to both I^A and I^B alleles. Therefore, both $\mathbf{I^Ai}$ and $\mathbf{I^Bi}$ genotypes will result in dominant phenotypes; i.e. having either the A or B carbohydrates. The **ii** genotype will result in the recessive trait of not having either of the carbohydrates.

Thus, based on the presence of the two carbohydrates, a person's blood group may be one of four types as follows: type A (carbohydrate A present), type B (carbohydrate B present), type AB (both carbohydrates A & B present), or type O (neither of the two carbohydrates present).

F_1 and F_2 generations resulting from a mating between individuals with different blood groups are shown below

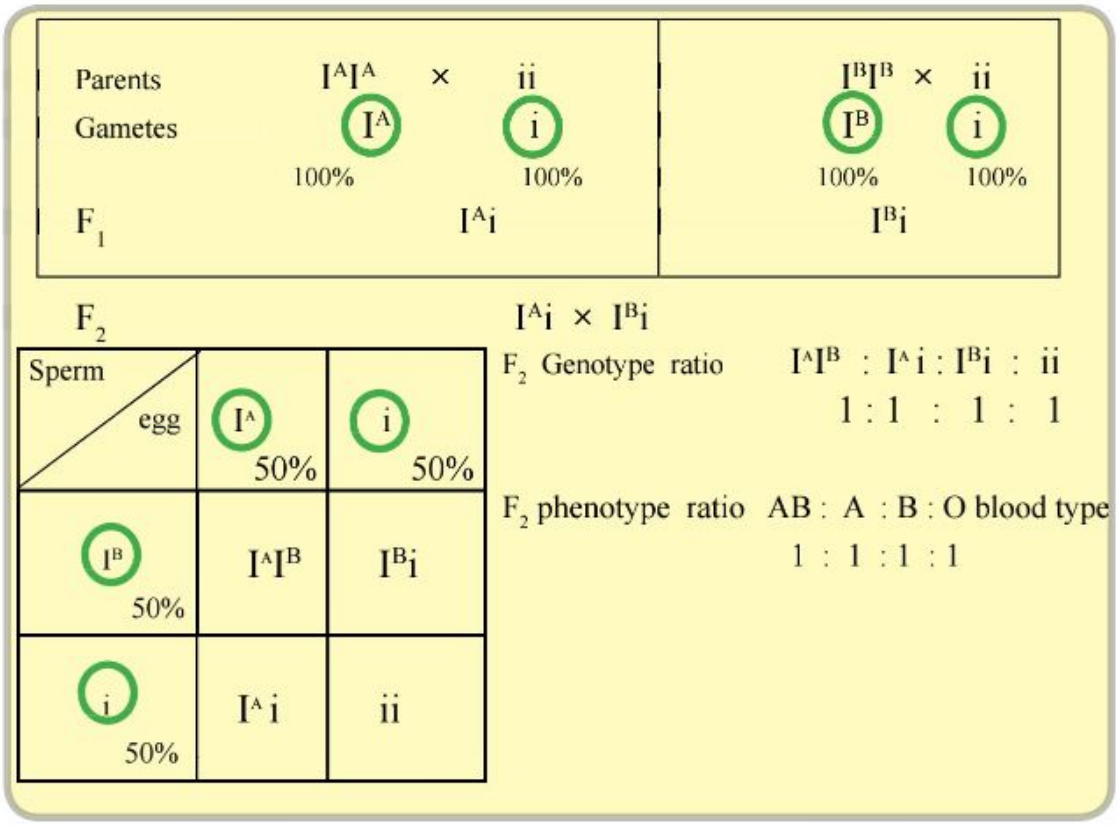

Figure 6.11: Multiple alleles for the ABO blood groups

Epistatis

Epistasis is the phenomenon resulting from interactions between genes of different loci. The alteration in the phenotypic expression of a gene at one locus is due to the interference of another gene at a different locus.

Epistasis could be categorized into two types; dominant epistasis and recessive epistasis, based on the nature of the gene interaction. This causes deviation from the phenotype ratios of Mendelian principles.

Dominant epistasis

When a dominant allele at a specific lous alters the expression of a separate gene at a different locus, it is referred to as **dominant epistasis.** Dominant epistasis is seen in the plumage colour of house fowls. Consider the following experiment.

A cross between a known homozygous, double dominant, white housefowl and a homozygous double recessive white house fowl results a 100% white F_1 progeny. F_2 generation produced from a cross between these F_1 individuals consists of white and coloured fowls in the ratio of 13:3.

The colour of these two fowl varieties are determined by two separate genes;

- The gene 'C'/ 'c' is responsible for producing colour in the feathers. The dominant 'C' allele produces colour while the recessive 'c' allele result in the absence of pigmentation.
- The gene **'I'** is epistatic to gene **'C'** and suppresses the expression of the colour. The dominant **'I'** allele prevents production of pigments while recessive 'i' allele is unable to prevent colouration.

As a result, the double dominant homozygous (CCII) house fowl is white (dominant **I** allele prevents colouration). The double homozygous recessive house fowl (ccii) is white (recessive c allele couldn't produce colour).

The F_1 generation all consist of heterozygous (CcIi) fowls. Due to the inhibitory effect of the dominant **'I'** allele, all F_1 individuals are white. When these fowls are allowed to interbreed, the F_2 generation genotypes carrying inhibitory **'I'** allele will give rise to white colour plumage, despite the presence of the dominant **'C'** allele. The presence of dominant **'C'** allele in the absence of inhibitory **'I'** allele gives rise to coloured fowls.

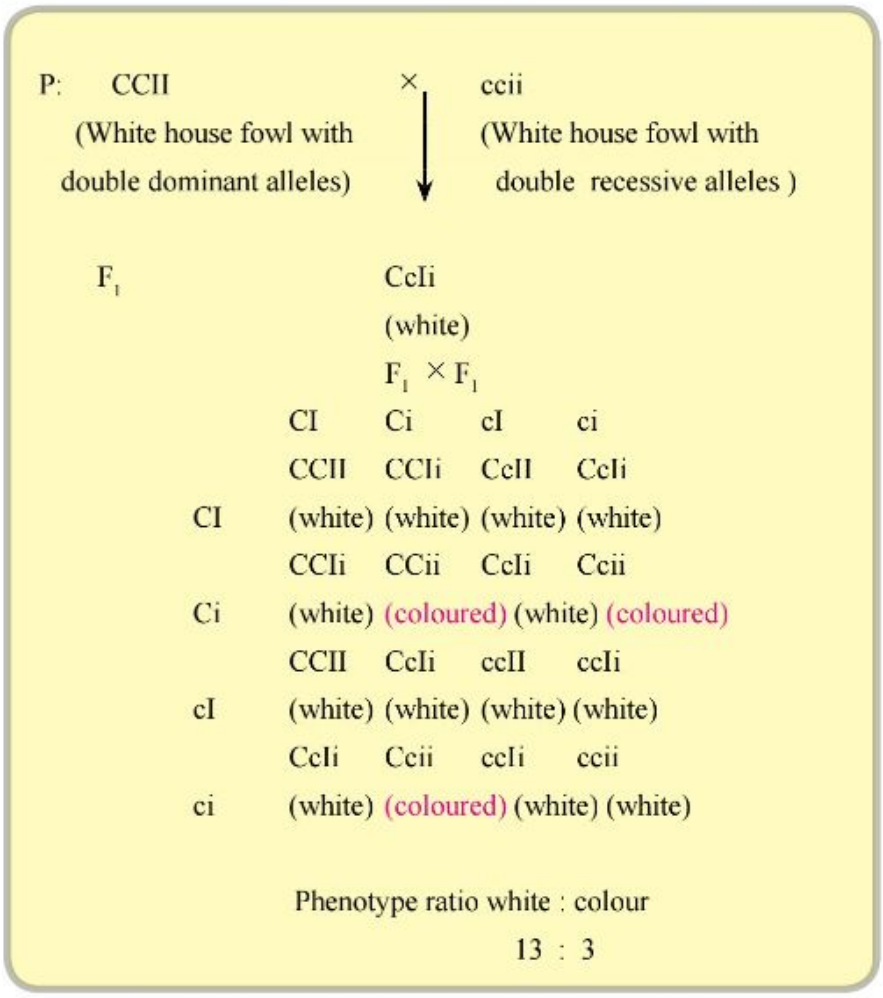

Figure 6.12:An example of dominant epistasis-Plumage colour of house fowl

As per the Mendelian principles, genotype ratios in both F_1 and F_2 generation are expected to be similar to the genotype ratios seen in a normal dihybrid cross. However, due to the effect of epistasis, phenotype ratio would deviate from Mendel's principles for normal dihybrid cross. F_2 phenotypic ratio may get altered from 9:3:3:1 to 13:3.

Recessive epistasis

When a homozygous recessive genotype of a particular chromosomal locus alters/ masks the expression of a separate gene at a different locus, it is referred to as **recessive epistasis.**

A good example for recessive epistasis is found for flower colour in sweet pea (*Lathyrus*) plant. There are purple flowered and white flowered varieties.

A cross between homozygous dominant purple flower (AABB) and homozygous recessive white flower (aabb) strains produced 100% purple colour flowers in F_1 generation. Inter breeding of F_1 plants produced F_2 generation with purple and white flower plants in a ratio of 9: 7.

The purple colour in sweet pea flower is governed by two dominant genes, A and B. Both A and B alleles are coding for compounds that are necessary for the expression of purple colour. Hence, purple colour will be there only when both the dominant alleles (A and B) are present.

Double recessive genotype at any locus (AAbb, aaBB, Aabb or aabb) results white flowers by masking the expression of purple colour. Thus, double recessive genotype at any of the above locus is epistatic to either homozygous dominant (AA and BB) or heterozygous condition (Aa and Bb) of the other. (AAbb, Aabb, aaBB, aaBb, aabb- White and AaBb, AaBB, AABb, AABB- Purple)(Figuer 6.13)

All of the F_1 generation plants were found to express purple flowers due to the presence of heterozygous condition at both loci (AaBb).

In F_2 generation, plants having genotypes with both **A** and **B** alleles (9/16) express purple flowers, and plants having genotypes with **'aa'** and a **'B'** allele (3/16) or **'A'** allele and **'bb'** alleles (3/16) and **'aabb'** genotype (1/16) produce white flowers, thus only two phenotypic classes are expressed; purple and white. Thus, the normal dihybrid phenotype ratio as per Mendelian principles **9:3:3: 1** is changed to 9: 7 ratios in F_2 generation.

Parents Purple × White Flower

Flower Flower

AABB aabb

F_1 AaBb (Purple Flower)

$F_1 \times F_1$ F_2

	AB	Ab	aB	ab
AB	AABB [P]	AABb [P]	AaBB [P]	AaBb [P]
Ab	AABb [P]	AAbb [W]	AaBb [P]	Aabb [W]
aB	AaBB [P]	AaBb [P]	aaBB [W]	aaBb [W]
ab	AaBb [P]	Aabb [W]	aaBb [W]	aabb [W]

P = Purple Flower, W = White Flower

Figure 6.13: an example of recessive epistasis - flower colour of sweat pea plants

Polygenic inheritance

Inheritance of a phenotype such as quantitative characters; height, skin colour, intelligence quotient etc. which results from a cumulative expression of two or more genes is called polygenic inheritance.

e.g: Skin colour in humans is determined by many genes. For simplicity only three genes are considered. Each gene (A, B, or C) has a dark-skin allele contributing one "unit" of darkness to the phenotype and being incompletely dominant to the other allele (a, b, or c). Therefore
AABBCC person: very dark skin
aabbcc person: very light skin
AaBbCc person: intermediate between skin colour
Because the dominant alleles from various loci have a cumulative effect, on the skin colour. (Campbell et al, 2015)

Based on the number of genes involved in determining a polygenic character, the phenotypic and genotypic combinations in the progenies may vary. Data for a polygenic character representing a population may result in a **normal distribution curve.** The majority of offspring would be expected to have intermediate phenotypes (skin colour in the middle range).

Genetic linkage

Some genes coding for particular characters are located on the same chromosome and also at a closer distance. Thus, they escape from crossing over and independent assortments occur during the meiotic cell division at gametogenesis and inherit together. This results in deviation from Mendel's law of independent assortment. The above phenomenon is called genetic linkage.
E.g. Inheritance of body colour and wing size in the fruit fly Drosophila

In *Drosophila,* wild-type flies are found to have gray bodies and normal-sized wings. Due to mutation for the above traits body colour becomes black and wings become vestigial. Both characters are determined by genes of autosomal chromosomes.

In this example, the mutant alleles are recessive to the wild-type alleles. The alleles for body colour are indicated as G (gray) and g (black), and those for wing size are indicated as N (normal) and n (vestigial).

To examine the above, wild type flies were crossed with flies which are mutant to both body color as well as wing size and followed by a dihybrid test cross.

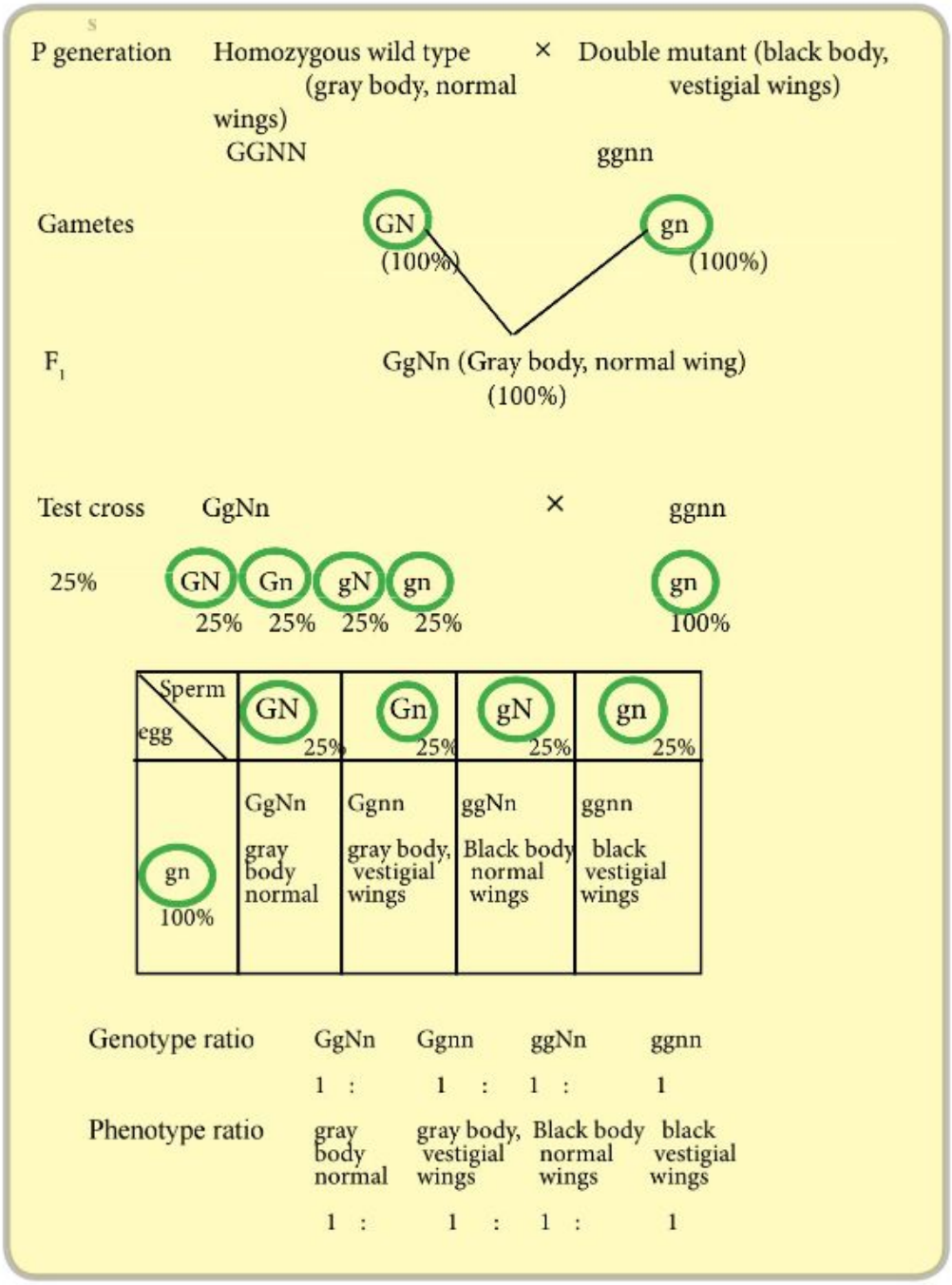

Figure 6.14: Prediction of inheritance of body colour and wing size in the fruit fly *Drosophila* as per Mendelian principles

Most offspring had a parental (P generation) genotype, indicating that the genes for body colour and wing size are genetically linked on the same chromosome.

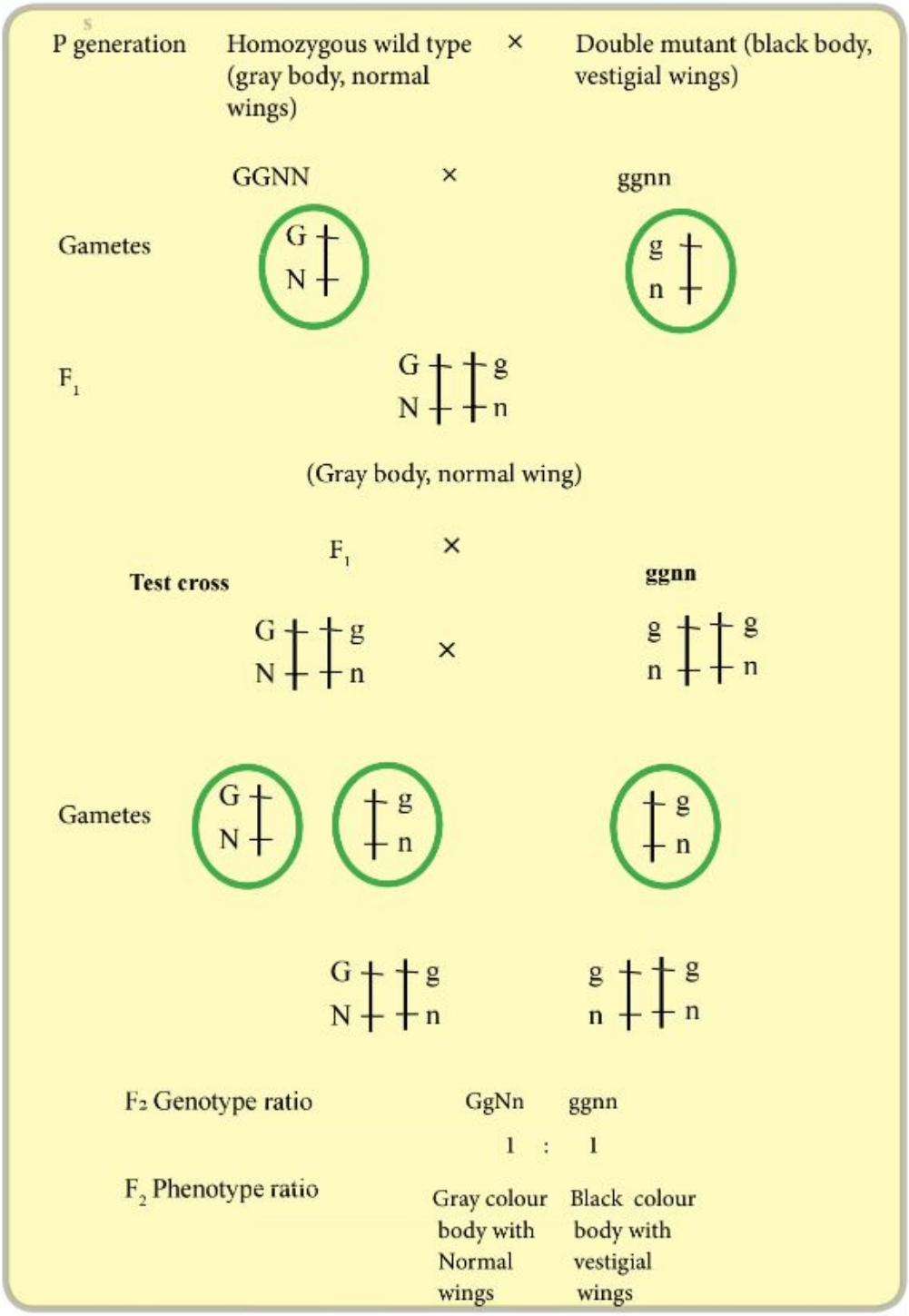

Figure 6.15: Inheritance of body colour gene and wing size gene in the fruit fly *Drosophila*

Though, the genes for body colour and wing size are linked, in some occassions, they get assorted independently due to crossing over. Therefore, the above test cross may results recombinant offsprings in lower frequency. for example, in Morgan's experiment recombinant phenotypes Gray colour body with vestigial wings (Ggnn) and Black colour body with normal wings (ggNn) were observed in lesser numbers.

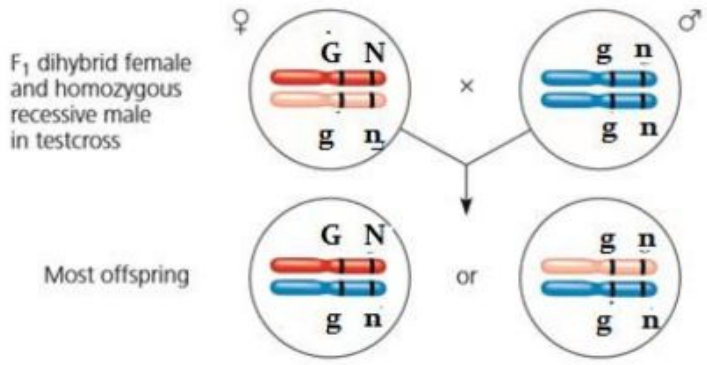

Figure 6.16: Linkage of genes responsible for body colour and wing size in the fruit fly Drosophila

The production of a relatively small number of offspring with non-parental phenotypes indicated occasional breaks in the genetic linkage. This is due to the crossing over occurs between the homologous chromosomes.

Human sex determination

Sex is determined by the expression of sex chromosomes. In humans, each individual carries 22 pairs of autosomal chromosomes and one pair of sex chromosomes. Type of sex chromosomes expressing male traits are named Y chromosome and the other as X chromosome. Comparatively X chromosome is bigger than Y chromosome. They both code for different traits except their homologous regions. When X and Y chromosomes pair up, they remain homologous only in specific regions. On the other hand when chromosome X, X pair up, they both remain homologous to each other.

On the occurrence of gametogenesis in females, meiosis yields haploid eggs carrying 100% X chromosomes, whereas in males, half the number of the haploid sperms produced carry X and the remaining half Y chromosomes. During the fertilization of male and female gametes, the occasion where both egg and sperm carry X chromosomes results in a female zygote, and on the other hand, an occasions where an egg fuses with a sperm carrying Y chromosome results in a male zygote.

Thus, any mating occasions between male and female organisms of same species leading to fertilization can have fifty percent chance for yielding either male or female zygotes.
Anatomical sex signs develop in humans based on the expression of XX and XY chromosomal combination.

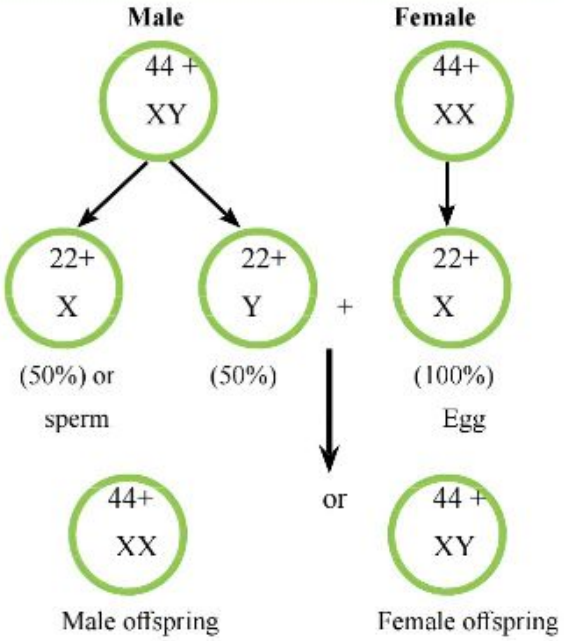

Figure 6.17: Sex determination of humans

Human sex linked characteristics

Certain characters of humans are carried on the genes located on the sex chromosomes. Those genes located on the sex chromosomes are called sex linked genes and the characters expressed by them are called sex linked characters.. Characters expressed by or carried on the X chromosome are called X- linked characters and the genes expressing or carrying those characters are called X- linked genes. On the other hand, characters which are expressed by or carried on the Y chromosome are called Y linked genes and the genes expressing or carrying those characters are called Y linked genes. Y chromosome carry only few genes other than those related to the sex. Some disorders carried on the Y-linked genes are transferred and expressed only through male progeny.

e.g. absence of certain Y-linked genes causes inability to produce normal sperm.

In addition to sex related characters, X chromosomes carry many other characters which are not relevant to individual sex.

e.g.

Red green colour blindness: *An X-linked recessive disorder characterized by the difficulty in perceiving differences between red and green colours.*

Haemophilia: *An X-linked recessive disorder where one or more of the proteins required for blood clotting are absent. Haemophilic person run the risk of severe bleeding during injuries due to the delay in clot formation. Campbell et al, 2015)*

Inheritance of X-linked genes

The inheritance of sex linked characters or genes differ for male and female due to the XX genotype of female and the XY genotype of male. During the fertilization, X chromosome from both of their biological parents result a female zygote and on the other hand, X chromosome from the female parent and Y chromosome from the male parent results a male zygote. Thus, for females, X-linked recessive disorders are expressed only at their homozygous genotype. However in males, due to the presence of only one X chromosome, have only one recessive X-linked allele. Therefore, having a recessive X-linked allele with disorder is sufficient for expression.

Pleiotropy

In some occasions, expression of a single gene affects the expression of multiple traits which are not related to each other. The above phenomenon is called as Pleiotropy. Pleiotropic alleles are responsible for the multiple symptoms associated with certain hereditary diseases in humans, such as cystic fibrosis and sickle-cell disease.

Sickle-cell disease

Sickle cell disease is caused by an alteration in the haemoglobin protein of red blood cells. A single gene mutation is responsible for the above condition. In homozygous recessive individuals, all the haemoglobins are of the sickle-cell variety. People living in high altitudes or under physical stress experience low oxygen content in their blood. Low oxygen content in the blood may induce the sickle-cell haemoglobin proteins to get accumulated and results sickle shape in red blood cells. Sickle cells may clump and clog small blood vessels causing tissue and organ damage in several body parts. This may result renal failure, heart failure and thrombosis.

Cystic fibrosis

Cystic fibrosis is a disease condition causing thicker and stickier mucus than its normal nature. As a result, mucus get accumulated in the pancreas, lungs, digestive tract, and reproductive organs which cause lung infections, respiratory failure, poor digestion, and infertility.

The thickening of mucus is due to the excess chlorine secretion of defected chloride channels of the plasma membrane. The defect in the trans-membrane chloride channel occurs as a result of the Cystic Fibrosis Trans-membrane Regulator (CFTR) protein. The altered CFTR protein is due to the mutation of CFTR gene. This is identified as autosomal recessive disorder.

Epigenetics

Study of occurence of certain phenotypes of certain characters controlled by factors other than their DNA sequence or genetic code is called epigenetics. This is due to 'switching on' and 'switching off' of certain genes by modifying nucleotides of a DNA sequence by methylation and demethylation, where methyl groups are added to wild type DNA sequence or else removed from a methylated DNA sequence. The above random occasions result different modified expression for a single DNA sequence.

Epigenetics results due to either inherited signals from parents or signals arising due to the environmental factors. Inheriting epigenetic traits from parents to the children's generation is called epigenetic inheritance. This may get reversed by various external stimuli from the environment. Some epigenetic influences result in inappropriate gene expressions leading to cancers.

Schizophrenia is a mental disorder that occurs due to the genetic defects. In some identical twins, only one of them gets schizophrenia and the other does not get it. This is due to two types of expressions for same DNA sequence, called epigenetics.

Population genetics

Hardy-Weinberg Equilibrium

Hardy-Weinberg Equilibrium is used to assess whether a population is evolving with respect to a particular characteristic/ genetic locus. The genetic makeup of a trait in a population would remain unchanged, if they are not evolving at that genetic locus. Therefore, the predicted data for a particular trait of a population can be compared with the actual data obtained from the same population. The comparison of both data as mentioned above may help to determine whether the population evolves or not for the considered trait.

Hardy-Weinberg Equilibrium Principle

In 1908 British mathematician **G.H. Hardy** and German physician **W. Weinberg,** independently showed that, in a population that is not evolving, allele and genotype frequencies will remain constant from generation to generation. This is now considered as key concept in population genetics and referred to as **Hardy-Weinberg Equilibrium Principle.**

To determine whether the allele and genotype frequencies have changed in consecutive generations, a Punnett square can be drawn, considering the combination of alleles in all possible crosses in a population.

The following example can be used to work out the Hardy Weinberg equilibrium.
A wild flower plant population showing incomplete dominance for the flower colour alleles have distinct phenotypes indicating their genotypes. i.e.

Plants homozygous for the C^R allele (C^RC^R) produce red pigment and have red flowers. Plants homozygous for the C^W allele (C^WC^W) have white flowers .
Heterozygous plants (C^RC^W) produce some red pigment and have pink flowers.

In the population of 500 flowers, there were 800 C^R alleles and 200 C^W alleles. Since the flower colour of the above plant type is determined by pair of alleles, 500 flowers may consist of 1000 alleles for their flower petal's pigmentation.
Therefore,

- allele frequency of C^R alleles (p) = $\frac{800}{1000}$ = 0.8
- allele frequency of C^w alleles (q) = $\frac{200}{1000}$ = 0.2

If the gametes are formed at random, the probability that an egg or sperm contains a C^R or C^W allele is equal to the frequency of each of these alleles in the population. Thus, each egg has an 80% chance of containing a C^R allele and a 20% chance of containing a C^W allele; the same is true for each sperm.

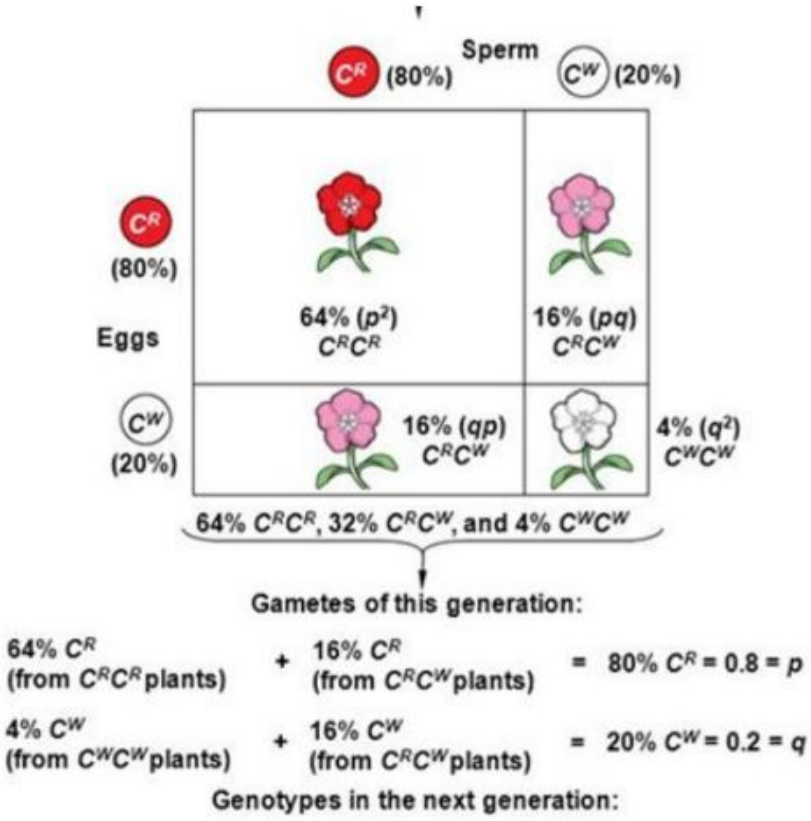

Figure 6.18: The Hardy-Weinberg Principle (Campbell et al, 2015)

During random fertilization, gametes fuse together randomly. Therefore, rule of multiplication can be applied to calculate the probability for each genotype combination.

According to Hardy-Weinberg equilibrium, if a character is determined by two alleles, the three genotypes will appear in the following proportions;

P_2 = frequency of dominant homozygotes

q_2 = frequency of the recessive homozygotes

2pq = frequency of the heterozygotes

The probability that two C^R alleles will come together, $p \times p = p^2 = 0.8 \times 0.8 = 0.64$

Hence the proportion of C^RC^R genotype in the progeny = 64%

The probability that two C^W alleles will come together, $q \times q = q^2 = 0.2 \times 0.2 = 0.04$

Hence the proportion of C^WC^W genotype in the progeny = 4%.

C^RC^W heterozygotes can arise in two different ways.

If the sperm provides the C^R allele and the egg provides the C^W allele,

the resulting C^RC^W heterozygotes in the progeny, $p \times q = 0.8 \times 0.2 = 0.16 = 16\%$

If the egg provides the C^W allele and the sperm the C^R allele,

the resulting C^RC^W heterozygotes in the progeny, $q \times p = 0.2 \times 0.8 = 0.16 = 16\%$

Thus, the total frequency of heterozygote in the progeny

$$pq + qp = 2pq = 0.16 + 0.16 = 0.32, \text{ or } 32\%$$

In the above example, only three kinds of genotypes are possible. When conditions suits for Hardy-Weinberg equilibrium, the sum of frequencies of all three genotypes equals 1. Thus, the equation for Hardy-Weinberg equilibrium can be written as below;

$$P^2 + 2pq + q^2 = 1$$

Conditions for Hardy-Weinberg Equilibrium

The Hardy-Weinberg approach describes a hypothetical population that is not evolving and fulfilling the following conditions.

1. **Absence of mutations.** Mutations result changes in alleles. Insertion, deletion, or substitution of nucleotides result altered alleles. This leads to modified gene pool.
2. **Occurrence of random mating.** Breeding occurs randomly without any influence that causes selectiveness. Mating of closely related individuals may alter the allele frequencies.
3. **Absence of natural selection.** All genotypes of the progeny are expected to survive irrespective to their differences, abilities and the environmental conditions. Variations in survival and reproduction of some genotypes may alter the frequency of alleles.
4. **Size of the population is extremely large.** In small populations, particular genotypes may disappear due to death or infertility. Therefore, larger the population, more likely to favor the Hardy-Weinberg equilibrium.

5. **Absence of immigration or emigration.** Individuals moving in and out of the populations may cause appearance of new genes and disappearance of existing genes. This is is called gene flow and it may alter allele frequencies.

Naturally, most populations do deviate from Hardy-Weinberg equilibrium except for certain genetic loci. Slowly evolving populations may also do not deviate much from Hardy-Weinberg equilibrium and therefore, they remain as predicted for a non-evolving population.

Evolution and change in gene frequency

Evolution can be explained in terms of changes in allele (gene) frequencies over generations. A species evolves when changes in gene frequencies drive the species into a higher level of adaptation for a specific ecological niche.

Genetic variation within the population is the key to evolution. Mutation will create new alleles and migration will include it into the population to increase variation. Natural **selection will then choose the better adapted individuals based on their phenotypic variations, causing population to evolve.** As a result, after the evolution the population is at a higher adaptive level compared to the level of adaptation they showed before being evolved.

This concept can be illustrated with the evolution of the peppered moth in England during the time of industrialization. The moth had two phenotypic varieties based on their colour; dark and light. Prior to the industrialization of central England, the light-coloured allele was most prevalent. The light-coloured moths had an advantage over the dark coloured ones as they could hide on the white-barked trees to avoid predation from birds.

Due to the pollutants generated parallel to the industrilalization, the light-coloured trees were stained dark. This exposed the light- coloured moths to predation, reducing their numbers. As a result, the light- colour allele became less prevalent. In its place, the dark-colour allele became more predominant, because dark moths could camouflage themselves better on the stained trees and avoid being eaten by their bird predators. The population evolved to a higher adaptive state with the change in gene frequencies (light – colour allele frequency went down while dark – colour allele frequency went up).

Plant and animal breeding

People have intervened in the reproduction and genetic make up of plants and animals since the dawn of agriculture, eight to ten thousand years ago. Early farmers selected the best looking plants and seeds and saved them to plant for the next season. Likewise, the best farm animals were allowed to mate with each other to preserve and improve their desirable traits.

This phenomenon wherein human beings interfere in the process of reproduction to allow only selective mating to occur, so that offspring with improved characters are produced is called **breeding** (as against natural reproduction).

With the science of genetics became better understood, plant and animal breeders used what they knew about the genes of a plant or an animal to select for specific desirable traits to develop improved plant varieties or animal breeds. The selection for features such as faster growth, higher yields, pest and disease resistance, larger seeds or sweeter fruits in crop plants, colour and pattern of the skin, hair or feathers in animals have now dramatically changed domesticated species compared to their wild relatives.

Importance of plant and animal breeding

In breeding programmes, the attributes, structure and composition of plant and animals are manipulated in such a way to make them more useful to humans. Accordingly, plant and animal breeding has a significant impact of world's agro-economy as discussed below;

Addressing world food and feed quality needs

An estimated 800 million people in the world, including 200 million children, suffer from malnutrition and associated health issues. Plant and animal breeding helps to enhance the value of food by improving their nutritional quality. For example, rice, which is the most widely eaten staple food, lacks many essential vitamins.

Another problem encountered in major food crops is the presence of toxic substances within them such as alkaloids in yam, cynogenic glucosides in cassava (manioc), trypsin inhibitors in pulses, and steroidal alkaloids in potatoes. Plant breeding is useful in reducing these toxic components and making them safer to eat. Plant breeding is also useful in making some plant products more digestible. For example, a high lignin content of the plant material reduces its value for animal feed which can be overcome with the use of breeding techniques.

Addressing food supply needs for a growing world population

It is anticipated that an additional three billion people will be added to the world population within the next three decades. Aligned with this population growth, an expansion in world food supply should be required to meet the projected needs. Unfortunately, land for farming is scarce and therefore more food will have to be produced on less land. This calls for improved and high yielding animal and plant varieties to be developed. In response plant breeding has produced super rice which has 50% more yield compared to the normal rice, super wheat which boost the

harvest by 20-40% of normal wheat and several high yielding corn, maize and soya bean varieties. The total production of meat and milk has also increased considerably *o*ver the years due to careful use of selective breeding techniques.

Need to adapt to environmental stresses

Weather and soil conditions can have a major impact crop yield. Climate changes and global warming are partly responsible for modifying the crop production environment (e.g., some regions of the world are getting drier and others saltier). To meet the increasing demand for food new cultivars need to be bred which can sustain these adverse conditions. For example, it is necessary to develop new plant types that can resist various biotic (diseases and insect pests) and other abiotic (e.g., salt, drought, heat, cold) stresses in the production environment. In response, genetically modified, pest resistant cotton, maize, and potatoes which carry Bt toxin, salinity tolerant rice varieties, cold tolerant tobacco, potato and strawberry varieties etc. are now available in agricultural industry. Likewise, both crop plants and farm animals (cattle, pig sheep goat etc.) with increased immunity to pathogens have also been produced through various breeding techniques.

Satisfying industrial and other end-use requirements

Consumers are having different requirements based on the texture, colour and composition of a particular food item irrespective of its taste or nutritional value. These diverse demands for the same food can be now successfully met through breeding procedures. For example, potato is a versatile crop used for food and industrial products. Different varieties are being developed by breeders for baking, cooking, fries (frozen), chipping, and for starch. These cultivars differ in size, specific gravity, and sugar content, among other properties. High sugar content is undesirable for frying or chipping because the sugar caramelizes under high heat to produce undesirable browning of fries and chips. Likewise, there is a high demand for seedless fruits such as grapes, melon and strawberries and also for leaner meat. Depending on these end-user requirements, it is possible to develop quality added products using animal and plant breeding techniques.

Developing animal and plant varieties with aesthetic values

Aesthetics is of major importance in horticulture as well as in the industry of ornamental and pet animals. The ornamental plant industry depends to a large extent on the development of new varieties that exhibit new flower/ leaf colours, varying sizes and attractive shapes etc. using plant breeding.

The pursuit of novelty has spurred a similar explosion of types in pet animals as well. Today selective breeding for numerous morphological features and functional abilities have given rise to nearly 400 dog breeds making them one of the most diversified species on earth. Likewise, there

are close to 50 rabbit breeds, vast number of bird varieties and an extensive range of ornamental fish.

Breeding techniques

Plant and animal breeders use numerous techniques to create new varieties with enhanced features. Many of these techniques have been successfully practiced over centuries even without the knowledge of the underlying genetics. Following section summarizes some of these traditional breeding techniques which has caused significant improvements in agriculture and farming.

Artificial selection

Artificial selection is the earliest form of biotechnology and has been used by humans for thousands of years. It, is a process of selective breeding, where plants or animals with specific traits were selected to breed so that their desired traits could be passed to the next generations to produce a high performing new variety. The method has made a huge impact on agriculture by way of improving plant and animal products before the discovery of more sophisticated technologies like genetic engineering.

The first prerequisite of artificial selection is the availability of variation with respect to the desirable characters. Once a population with a desirable variation is recognized, the best performing individuals for the desired feature are selected. For example, when selecting for fruit size in plants, only those giving the biggest fruits are chosen for the breeding programme and the rest of the population is discarded or rejected. The progeny of the selected individuals is grown further and again screened for the desired feature. This process is repeated sometimes for many generations, until a uniform plant population is attained which has the best-desired characters. Eventually, a new uniform crop variety with the desired characteristic is produced by this successive selection, followed by multiplication of the selected individuals.

The advantage of selective breeding is that it uses the processes of natural selection, but under direct supervision from carefully selected animals or plants with the desired traits. There are no genetic modifications or other forms of tampering that could potentially harm people and the risk to the plant or animal is often minimum.

Crops like corn and wheat are commonly selectively bred in order to obtain the highest yielding plants. Breeding animals with higher protein and lower fat percentages, as well as plants that have higher nutritional values, had been used to create food sources with a higher quality of nutrition. In addition, selective breeding has, effectively removed undesirable traits such as low resistance to disease, in some animals and plants.

However, selective breeding among animals can take a long time for the process to work. In horse breeding, for example, the given standard to establish a new breed is to have offspring with the desired traits to be produced over the course of 7 generations. This means it may take 25-50 years for the desired traits to become a foundational component of an animal.

Inbreeding and out breeding

Inbreeding

The breeding among genetically similar individuals are known as inbreeding.

Among plant breeders the term "inbreeding" is commonly used to mean self-fertilization, i.e. the fertilization of a flower with its own pollen or with pollen from a different flower on the same plant. This is done to produce an inbred variety, which is exactly the same generation after generation. Many important crops, such as wheat, oats, barley, and tobacco, are produced from seeds which are habitually self-fertilized.

However, in animal breeding the term " inbreeding " is used to refer to the mating of closely related individuals, as, for instance, the mating of father and daughter, brother and sister, or cousins. In both crop plants and farm animals, inbreeding brings uniformity of the required type while preserving the desired characters. Inbreeding is used for developing pure lines in agriculture as well as for research.

As a rule, inbreeding increases homozygosis and thus exposes harmful recessive genes which would have otherwise stay hidden among heterozygotes. Continued inbreeding, therefore, reduces genetic fitness of the population. As a result, the growth and fertility of the inbred population would go down with adverse effect on their productivity. Prevalence of genetic disorders might also increase among the inbred population. This phenomenon of having a reduced genetic fitness in a given population as a result of inbreeding is called **inbreeding depression.**

However, in agriculture and animal husbandry, positive effects of inbreeding will be harnessed as much as possible. To ensure this, only those offspring that are exhibiting the desired trait, without other negative ones, will be used for future breeding. The negatively affected individuals in the progeny are removed or are not be allowed to be bred. Thus, inbreeding is used in agriculture to help accumulation of superior genes.

Outbreeding

When plants or animals of different breeds (races) are mated with each other, it is known as **outbreeding** or **cross breeding.** This allows the desirable characters of the exotic parent, which the indigenous parent does not have, to be transmitted to the progeny.

For example, cross breeding is carried out by animal breeders to enhance milk and meat production. In India zebu breeds of cows and nondescript cows are crossed with exotic breeds like Holstein Fresian, Brown Swiss and Jersey bulls or their semen, to enhance the milk production potential of the progeny. Likewise crop plants like corn and hemp, are normally cross-fertilized.

Hybrid breeding

When genetically unrelated pure-bred plants or animals in the same species are mated with each other it is known as **hybridization** or **outcrossing.**

Generally, this is carried out with plants and animals who do not share common ancestors on either side of their pedigree up to four to six generations. The offspring of such a mating is known as the **out cross** and will possess stable characteristics and hybrid vigour. **Hybrid vigour,** also called **heterosis,** is the increase in such characteristics as size, growth rate, fertility, and yield in the hybrid organism over those of its parents.

Plant and animal breeders exploit heterosis by mating two different pure-bred lines that have certain desirable traits. The first-generation offspring generally show, in greater measure, the desired characteristics of both parents. This vigour may decrease, however, if the hybrids are mated together; so the parental lines must be maintained and crossed for each new crop or group desired.

In plant breeding, because creating hybrids involves many years of preparation to create pure lines that have to be constantly maintained so that F_1 hybrid seeds can be harvested each year, the seeds then become more expensive. Nevertheless, hybrid seeds have had a tremendous impact on agricultural productivity. Today, nearly all corn and 50% of all rice are hybrids. In the US, the widespread use of corn hybrids, coupled with improved cultural practices by farmers, has more than tripled corn grain yields over the past 50 years from an average of 35 bushels per acre in the 1930s to 115 bushels per acre in the 1990s. No other major crop anywhere in the world even comes close to equaling that sort of success story.

Many cultivars of popular vegetables or ornamental plants are hybrids. In terms of improved plant characteristics, tropical vegetable breeders can point to some rather clear achievements over the last two decades:

- **Yield improvement.** Hybrids often out yield traditional true breeding varieties (inbred varieties) by 50-100% due to its improved vigor, improved genetic disease resistance, improved fruit setting under stress, and higher female/male flower ratios.
- **Extended growing season**. Hybrids often mature up to 15 days earlier than local true breeding varieties. For many crops, the hybrid's relative advantage over the true breeding is most pronounced under stress conditions.
- **Quality improvement.** Hybrids have helped stabilize product quality at a higher, and more uniform level – this implies improved consumption quality (e.g. firm flesh of wax gourd, crispy taste of watermelon).

Interspecific breeding

In this approach, which is also known as interspecific hybridization, male and female organisms of two different species are mated. The progeny obtained from such a mating are usually different from both the parental species and may be fertile, partially fertile, or sterile.

Plants hybridize much more frequently and successfully than animals do. Pollen from flowering plants disperses widely and may land on flowers of other species allowing natural interspecific breeding to take place. Plant forms are less stringently controlled than animal forms, and so the intermediate form of a plant hybrid is more likely to be physiologically successful.

Often interspecific hybrids are sterile or for some other reason cannot interbreed with the parental species. Occasionally sterile interspecific hybrids can undergo a doubling of their chromosome set and become fertile tetraploids (four sets of chromosomes). For example, the bread wheats that humans use today are a result of two hybridizations each followed by chromosome doubling to produce fertile hexaploids (six sets of chromosomes). In such instances, the hybrids can become new species with characteristics different from either of the parents. Crop yields increase dramatically when hybridization is used to exceed one or more of the parents in size and reproductive potential. For example, boysenberries (Rubus ursinus x idaeus) were developed at Knott's Berry Farm in California. They are a result of a set of crosses between blackberries (*Rubus fruticosus*), European raspberries (*Rubus idaeus*) and loganberries (*Rubus* × *loganobaccus*).
For many fruit crop species, the use of interspecific breeding is increasing, in order to utilize naturally occurring sources of pest and disease resistance, fruit-quality components, etc. within the available germ plasm. For example, winter hardiness of apple was improved by making a hybrid species *Malus* × *domestica* from *Malus* × *asiatica* and *Malus pumifolia.*

However, among animals interspecific breeding is restricted to few species. Common examples include Mule (male donkey x female horse), Hinny (male horse x female donkey) and Liger (male lion x female tiger).

Note that in mules and hinnies, the common genus the parents belong to is Equus and in liger, its Panthera. Other examples are zebra/donkey cross resulting in an offspring called zonkey, zebra/ horse cross resulting in zorse. The offspring from this cross could develop into adults, but may not develop functional gametes. Sterility is often attributed to the different number of chromosomes the two species have, for example, donkeys have 62 chromosomes, horses have 64 chromosomes.

Genetic principles of breeding techniques

From the beginning of plant and animal breeding, farmers made use of principles of genetics, with or without awareness of these concepts. Following section explains, three most widely used genetic principles applied in animal and plant breeding at present.

Polyploidy

Polyploidy refers to the presence of more than two complete sets of homologous chromosomes per cell nucleus. This is a widely used principle in plant breeding. In plants, polyploidy can be induced artificially using antimitotic agent, colchicine.

One of the most important consequences of polyploidy for plant breeding are the increment in plant organs ("gigas" effect) caused by the larger number of gene copies. Polyploid individuals may thus exhibit larger organs compared to their diploid counterparts, such as roots, leaves, tubercles, fruits, flowers and seeds. Polyploid plants also have lower growth rates, and tend to flower later or over a longer period of time than related diploids, which is a desirable feature for ornamental breeding.

In addition, polyploidy often results in reduced fertility due to meiotic errors, allowing the production of seedless varieties such as the triploid watermelon. On the other hand, when the crossing between two species is not possible because of differences in ploidy level, polyploids can be used as a bridge for gene transferring between them. Similarly, the genome doubling in a newly formed sterile hybrid allows the restoration of its fertility.

Genome redundancy (having additional gene copies due to increase ploidy) have other benefits as well. It promotes a "buffering" effect in which the deleterious alleles are masked by the extra copies of wild-type alleles. At the same time, it allows functional diversification of redundant gene copies, in which one member of a duplicated gene pair mutates and acquires a novel function, without compromising essential functions.

The increment in heterozygosity is another feature that accompanies polyploidy. Higher levels of heterozygosity have been positively related to vigor increment in maize, potato and alfalfa improving the product quality and increasing the tolerance to both biotic and abiotic stresses.

Mutation Breeding

Methods for inducing mutation have the potential of producing new sources of genetic variability for crop breeding. These methods can be employed when it appears there is little, or no, variability for the character to be improved available within the gene pool of the species. This method of inducing desirable mutations in crop plants using either chemical or physical agents was termed **mutation breeding.**

Several agents can be used to cause mutations. This include ionizing radiation such as gamma rays, protons, neutrons, alpha and beta particles and chemicals such as sodium azide and ethyl methanesulphonate. Since the desirable mutations induced by these treatments are found at a very low frequency (0.1% of total mutations), breeders have to screen a large population to select a desirable mutation. In addition, most mutations act in a recessive fashion and are likely to be masked by their dominant allelic counterparts making the screening procedure even harder.

The effectiveness of using induced mutation depends on the breeding system of the plant. Its use in self-pollinated plants is likely to be more successful than in crosspollinated ones. Populations of cross-pollinated plants usually possess stores of genetic variability in the recessive condition and it would not be likely that induced mutation would produce significant amounts of new variability. Further, induced mutation is potentially useful in the improvement of asexually propagated crop plants.

Despite these limitations, mutation breeding efforts continue around the world today. It has improved both morphological and physiological characteristic of both crop and ornamental plants such as flower colours, seed size, crop yield, disease resistance and salinity tolerance, drought tolerance and early maturity. Examples of plants that have been produced via mutation breeding include wheat, barley, rice, potatoes, soybeans, and onions.

Genetic modification

Genetic modification, also called **genetic engineering,** is the direct manipulation of an organism's genes to change the genetic makeup of cells. In this method, genetic material is obtained from one organism showing a desired trait and will be inserted to another second organism using recombinant DNA technology, so that the receiving organism will also show the same desirable trait. Thus, by transferring genes within and across species, improved or novel organisms are produced.

In traditional plant breeding techniques transfer of genes is limited to the closely related species or genera. For example, traditional breeding techniques could not be used to insert a desired gene from daffodil into rice because the many intermediate species between rice and daffodil and their common ancestor is extinct. With genetic engineering, however, such gene transfers can be done more quickly, more specifically, and without the need for intermediate species. The term **transgenic** or **genetically modified organism (GMO)** is used to describe organisms that have been engineered to express a gene from another species. Advocates for plant biotechnology believe that the genetic engineering of crop plants is the key to overcoming some of the most pressing problems of the 21st century, including world hunger and fossil fuel dependency. Examples of transgenic plant varieties include transgenic papaya that is resistant to ring spot virus, "golden rice," with increased levels of beta-carotene and salinity resistant rice varieties among others.

Natural or artificial breeding: Advantages and disadvantages

Although artificial breeding is extensively practiced today with remarkable economic gains, there are serval drawbacks in the method in comparison to the natural breeding.

The artificial breeding is geared towards producing a uniform set of plants or animals with traits desirable to humans. The development of this uniformity needs compromising the variability within the species. This reduction of genetic diversity will have adverse consequences on the evolutionary fitness of the species leading to low resistance to infections, higher prevalence of congenital anomalies and reduced fertility. For example, when a population of animals or plants with the same genetic traits are attacked by a pathogen to which they are susceptible, the entire population is likely suffer due to the absence of the resistant trait within the gene pool. This limited opportunities for the natural selection to act upon the population is likely to drive them towards a lower fitness plateau.

On the other hand, natural breeding can rule out weaknesses and disabilities which affect survival by allowing natural selection to act upon the species. This will produce fitter and stronger individuals in the long run. However, the natural selection would not guarantee a productivity increase in the consumer perspective, despite the increase in genetic fitness.

As discussed earlier, sometimes inbreeding is practiced as an artificial breeding technique. This would result in an increase in homozygosis which would in turn increase the expression of recessive deleterious mutations that would otherwise stay masked within heterozygotes. This can cause the population to undergo inbreeding depression with adverse effects on the overall fitness.

Sometimes artificial breeding can exhibit negative correlated responses. This refers to the fact that while artificial breeding is improving certain characters in the population, simultaneously and unintendedly it could lead towards deterioration of other characters that are not under direct

observation. For example, the shape of the skull in some dog breeds has made it difficult for them to eat normal food because of the upper jaw being much shorter than the lower jaw, such as in the case of Boxer or the Bulldog. Likewise, selection for large offspring has resulted in a high fraction of difficult births, sometimes requiring caesarean sections in the Texel sheep, and even almost as a standard way of delivering in the beef cattle breeds Belgian White-and-Blue cattle and the Dutch Improved Red-and-White. This kind of negative responses are difficult to predict in advance and usually only visible after the new breed is established.

Although there are negative impacts artificial breeding is still preferred over natural breeding for the numerous advantages it could bestow upon overall animal and plant productivity as discussed earlier.

Notes:

This is to acknowledge that some of the diagrams used in this book have been taken from various electronic sources using internet . This book is not published to make profit and sold only to cover cost.

The resource book is prepared according to the subject content and learning outcomes of the G.C.E. (A.L) Biology new syllabus which is implemented from 2017.

The content of this Resource book declares the limitation of the G.C.E. (A.L) Biology new syllabus which is implemented from 2017.

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