Chapter

9 Genetics

REMEMBER

Before beginning this chapter, you should be able to:

- Differentiate asexual mode from sexual mode of reproduction
- Remember structure and function of nucleus
- Recall the DNA structure and chromosome numbers

KEY IDEAS

After completing this chapter, you should be able to:

- Understand the concepts of heredity and variation
- Explain Mendel's monohybrid and dihybrid crosses
- Understand concept of sex determination
- Discuss different types of genetic disorders

INTRODUCTION

The branch of biology that deals with the study of heredity and variations is called genetics. The study of genetics enables us to understand how the characters of parents are transmitted to the children and then to the subsequent generations. Genetics also helps us to understand how and why the individuals of the same species show variations in their characters. The progress of genetics resulted in major advancements in various fields such as agriculture, medicine, forensic science, and so on. Though genetics is relatively a new branch of science, it is considered as the most important area of science that leads to dramatic breakthrough in the modern world as it contributed to the transformation of society in a big way.

Info Box!

The word 'genetics' originated from the Greek word '*genesis*' that means to grow into or to become.

Table 9.1 Contributors in the field of genetics

Scientist	Year	Contribution
Gregor Johann Mendel	1865	Breeding experiments on garden pea; laws of heredity; Father of genetics
Hugo de Vries	1886	Mutations
J. Kolreuter and F. Galton	1883	Quantitative inheritance
Sutton and Boveri	1902–1903	Chromosome theory of inheritance
Thomas Hunt Morgan	1908	Genetic experiments on fruit fly (*Drosophila melanogaster*); Father of experimental genetics
H.G. Khorana	1960s	Synthesis of artificial gene
W. Bateson	1905	Coined the term 'genetics'

HEREDITY AND VARIATION

Reproduction is the process by which all the living organisms produce their offspring. The characters are passed on from one generation to the next by this process. Asexual mode of reproduction produces offspring that are almost exactly similar to the parents. They show very little differences from their parents. As only one parent is involved in asexual mode of reproduction, their offspring are called clones. The clones produced inherit all the characters of the parent, and hence closely resemble the parent organism. However, in the higher organisms, sexual reproduction is the principal mode of reproduction. It results in offspring that show some differences from their parents. This degree of differences between offspring and the parents is called variation. That means, some variations are produced during the process and these variations may be transmitted from one generation to the next. This transmission of characters and variations along the forthcoming generations is known as inheritance. This phenomenon of exhibiting inheritance is called heredity.

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The inheritance of characters from the parents to the offspring is known as heredity.

Heredity results in some resemblances and differences between the parents and their offspring. The characters that are transmitted to the succeeding generations are called inherited traits. Some commonly observed inherited characters in human beings include skin colour,

eye colour, hair colour, height, shape of the ear lobes, eye brows, etc. Apart from these observable features, there are certain anatomical features such as blood group that offspring inherit from their parents. Some behavioural patterns such as hand use, pattern of walk and intelligence also seem to be inherited across the generations.

Units of Heredity

The chromosomes present in the nucleus of living cells serve as the vehicles of transmission of characters. The chromosomes possess DNA that is the genetic material and is passed on from one generation to the next. The segment of DNA with a specific sequence of nucleotides is called gene. Hence, genes are the physical units of heredity and DNA is the molecular or chemical basis of heredity. Nucleus in the mature cells of all higher organisms contains chromatin material in the form of inter-woven network of fibres. The chromatin material consists of DNA as the genetic material. The chromatin material gets organized into chromosomes at the time of cell division. A particular species of organisms is characterized by a certain number of chromosomes as counted at the time of cell division.

Table 9.2 Number of chromosomes in selected organisms

Organism	Number of Chromosomes
Garden pea (*Pisum sativum*)	14
Fruit fly (*Drosophila melanogaster*)	6
Female Honey bee	32
Tobacco plant	48
Human beings	46

Info Box!

The word 'chromosome' comes from the Greek word, *chrome* meaning colour. Chromosomes got their name owing to being stained by dyes.

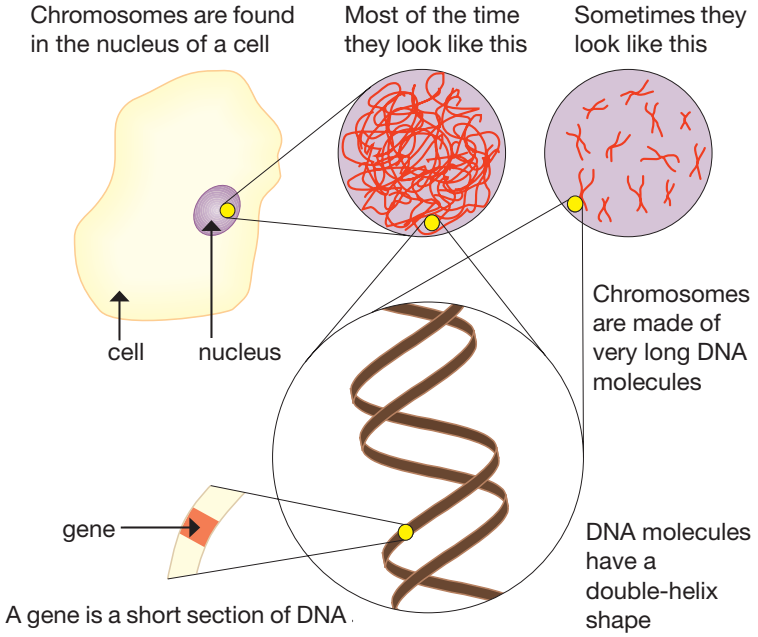

FIG. 9.1 Diagram showing magnification of chromosomes and DNA in cell nucleus

During the interphase, the chromosomes organize themselves as pairs in each cell and are called homologous chromosomes. One from each homologous pair is derived from each parent. The chromosomes which determine the sex of an individual are termed as sex chromosomes or allosomes. The chromosomes other than sex chromosomes are called autosomes.

For example, human cell consists of 23 pairs of homologous chromosomes, that is, 22 pairs of autosomes and one pair of allosomes or sex chromosomes (XY in males and XX in females).

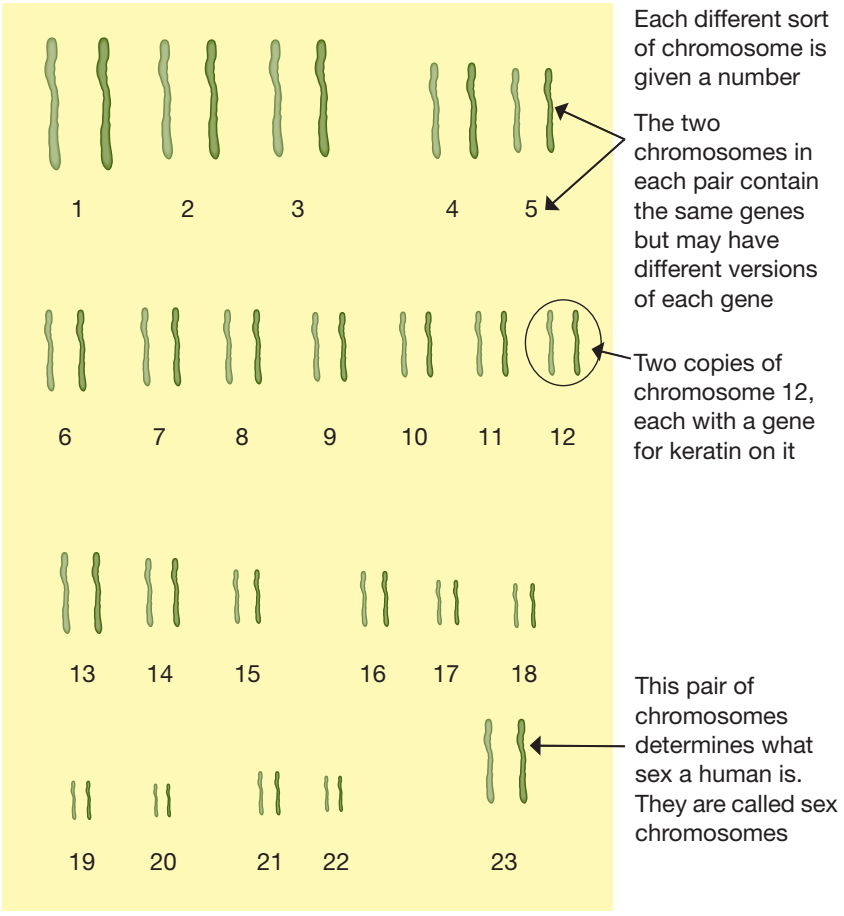

FIG. 9.2 Illustration of homologous chromosome in humans

Allele

The two chromosomes in a homologous pair possess genes representing the same characters. Each character is present as a pair of contrasting traits called alleles.

For example, a particular gene represents height of the organism. There are three possible combinations of the pair of genes representing the height.

1. The genes on both the chromosomes may correspond to tallness.
2. The gene on one chromosome may correspond to tallness and that on other chromosome may correspond to dwarfness.
3. The genes on both the chromosomes may correspond to dwarfness.

In this case, height of the organism is the character under consideration. The tallness and dwarfness are the contrasting traits. The genes that represent the two contrasting traits are called alleles. The distribution of chromosomes containing genes during cell division is the basis of the phenomenon of heredity.

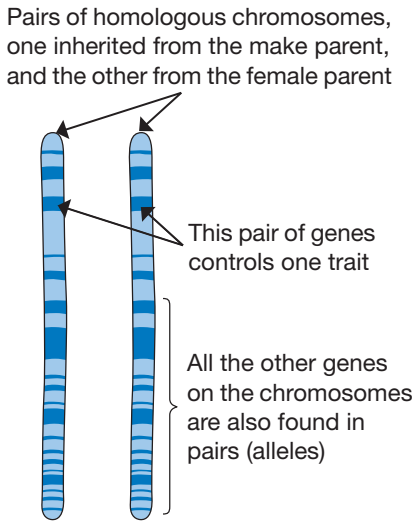

FIG. 9.3 Illustration of a pair of homologous chromosome

Cell Division and Heredity

During the process of cell division, the homologous chromosomes get separated and are distributed into the daughter cells. Somatic cells in the body of an organism undergo mitotic cell divisions in order to produce daughter cells with exactly same DNA sequence. This results in the growth and development of the organism. Germ cells in the body of an organism undergo meiotic cell divisions to produce male or female gametes as the case may be. Meiosis cell division involves the distribution of homologous chromosomes of the germ cells to the gametes. This distribution of homologous chromosomes in meiotic cell division takes a different course from that in mitotic cell division. This is the basis of mechanism of heredity.

Mechanism of heredity

The mechanism of heredity basically involves three important phenomena taking place in meiotic cell division.

1. DNA replication
2. Linkage and crossing over
3. Genetic recombination

DNA Replication

This takes place in the same way as in case of mitotic cell division during the interphase, which is the preparatory phase. The homologous chromosomes paired up during this phase possess the genes of both the parents. This leads to the transmission of characteristics of the previous generations.

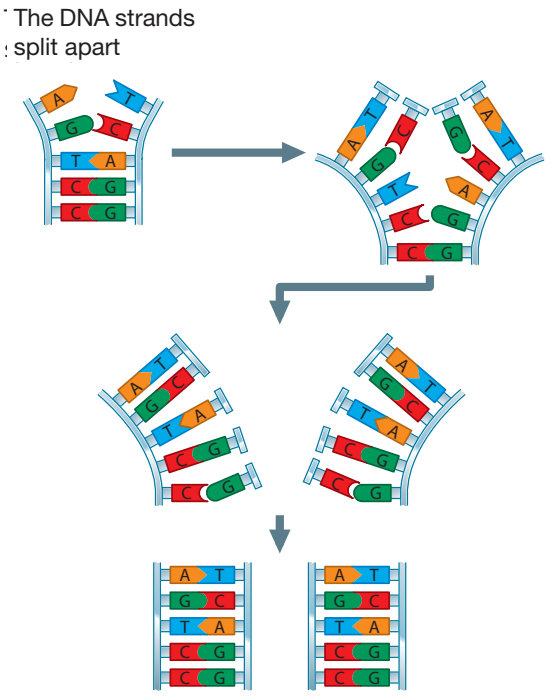

FIG. 9.4 Illustration of DNA cell replication

Linkage and Crossing Over

These processes are exclusive to meiotic cell division. These are responsible for inheritance of some characteristics and variation in some other characteristics. In meiotic cell division, all the genes on the homologous chromosomes are not transmitted to the daughter cells. Some groups of genes are together passed on to the daughter cell. Such group of genes that is normally inherited together is termed as linkage group and the genes are called linked genes. The phenomenon in which certain genes present on a chromosome are together carried on to daughter cell during meiotic cell division is called linkage. If all the genes on a chromosome are transferred, it is called complete linkage. If some of the genes are transferred, it is called incomplete linkage. Linkage is responsible for the inheritance of characters to the successive generations. Crossing over is a somewhat opposite phenomenon of linkage. The phenomenon of the exchange of segments of non-sister chromatids (genes) of homologous chromosomes is known as crossing over. Although linkage serves to preserve the characters of parental generation, crossing over gives scope for producing new characters in the successive generations.

Recombination

Due to the phenomenon of linkage and crossing over, two probable combinations of chromosomes can be visualized:

1. Parental combination
2. Recombination

Linkage of all the genes on the chromosomes results in parental combination, which usually does not happen during meiotic cell division. Crossing over is generally followed by recombination. This involves exchange of genetic material between the chromosomes and formation of chromosomes with some new sets of genes in the daughter cells. The daughter cells are the haploid gametes. These gametes undergo gametic fusion to form zygote with unique DNA sequence. This zygote undergoes repeated mitotic cell divisions in which the DNA sequence of the zygote is preserved. Consequently, the new organism (offspring) formed has a DNA sequence unique to itself and is considerably different from the parents. The new characters that are acquired by the offspring due to the new genetic combination are called variations. Therefore, linkage is responsible for parental combination.

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Crossing over followed by recombination is responsible for recombination.

For example, Father with dark complexion and long stature.
Mother with fair complexion and short stature.
Offspring with dark complexion and short stature is one recombination.
Offspring with fair complexion and long stature is another recombination.
Offspring with medium complexion and medium stature shows variations.

The differences existing among the individuals of a species from one generation to the next and also among the offspring of the same parents are called variations.

MENDELIAN INHERITANCE

Gregor Johann Mendel, an Augustinian priest and scientist, made a lot of progress in the study of the inheritance of various traits from one generation to the next. For this reason, he is regarded as the 'father of genetics.' Mendel conducted various experiments to study the mechanism of inheritance from parents to offspring by choosing pea plant (*Pisum sativum*) as the experimental material.

FIG. 9.5 Gregor—Johann Mendel 'Father of Genetics'

Reasons for Choosing Pea Plants for Mendel's Experiments

Mendel had selected pea plant for various reasons listed below.

- Easy cultivation of pea plants
- Pea is an annual plant that gives results within a year
- Pea plant is normally self-pollinated, however, it could be cross-pollinated by removing male and female reproductive parts.
- Large number of varieties with observable alternative forms for a trait in pea plants are available with a number of contrasting traits. This makes it convenient for taking plants with various contrasting features and studying the transmission of those traits to the next generations.

Mendel's Experiments

Mendel selected pea plants having one or two of the contrasting traits and conducted the experiments. According to Mendel, one of the contrasting traits was considered to be dominant and the other one to be recessive. According to Mendel, the contrasting traits are carried by factors that are now called genes. The dominant trait of each character is represented by the first letter of that trait with capital letter. Recessive traits of each character are represented by indicating them with small letters of the corresponding letter for the dominant trait.

Table 9.3 Seven pairs of contrasting features of garden pea

	Dominant factor		Recessive factor	
Character	**Trait**	**Representation**	**Trait**	**Representation**
Stem height	Tall	T	Dwarf	t
Flower colour	Purple	P	White	p
Flower position	Axial	A	Terminal	a
Pod colour	Green	G	Yellow	g
Pod shape	Full	F	Constricted	f
Seed colour	Yellow	Y	Green	y
Seed shape	Round	R	Wrinkled	r

Table 9.4 Description of genes and alleles

Dominant allele		Recessive allele	
Definition	**Example**	**Definition**	**Example**
The gene corresponding to the trait that expresses itself even in the presence of the gene for the contrasting trait as it masks and hides the other trait	• Purple colour of flowers is the dominant trait • P represents dominant allele	The gene corresponding to the trait that fails to express itself in the presence of the gene representing its contrasting dominant trait	• White colour of flowers is the recessive trait • P represents the recessive allele
Homozygous		**Heterozygous**	
Definition	**Example**	**Definition**	**Example**
An organism having the same alleles that represent either both dominant traits or recessive traits	RR, rr	An organism having contrasting factors or alleles representing contrasting traits (one dominant allele and one recessive allele)	Rr

Terminology Related to Genetics in Mendel's Experiments

Alleles: The genes (factors) representing a particular character have their contrasting forms that are called alleles. The alternative forms of a character are called contrasting traits.
Pure breed: The variety of individuals in which a particular character remained unchanged in large number of successive generations
Cross: A mating between two individuals leading to the fusion of gametes

Table 9.5 Description of cross

Monohybrid cross		Dihybrid cross	
Definition	**Example**	**Definition**	**Example**
The crossing of plants considering one pair of contrasting characters	**TT × tt**	The crossing of plants with two pairs of contrasting characters	**TTRR × ttrr**

Phenotype: The expressed or observable characteristics of an organism. For example, plant with purple flowers with both genes for purple; one gene with purple and one gene for white flowers.

Genotype: The actual genetic constitution of an organism. For example, PP type purple flowers; Pp type purple flowers

Mendel performed experiments in three stages:

1. Selection of pure or true breeding parents (Parental [P1] generation) obtaining first generation plants (First filial or F1 generation)
2. Self-pollination of F1 plants to get subsequent generations (F2, F3, etc.)
3. Hybridization to produce hybrid which is the offspring produced by a cross between the individuals of same species that differ from each other at least in one character.

Mendel's Monohybrid Cross

Pea plants with one pair of contrasting features are chosen and allowed to undergo self-pollination between them. This constitutes P1 parental generation). Since the plants of P1 generation are produced by self-pollination, they possess factors (genes) for either dominant or recessive traits.

For example, **P1** generation—Plant with purple flowers (PP) and plant with white flowers (pp). Purple colour of flower is a dominant trait. White colour of flower is a recessive trait.

F1 generation—Cross-pollination between purple-flowered plant and white-flowered plant resulted in all purple-flowered plants. But these are hybrid plants with one factor for dominant trait and one factor for recessive trait (Pp).

F2 generation—Self-breeding of F1 hybrid plants resulted in some purple-flowered plants and some white-flowered plants.

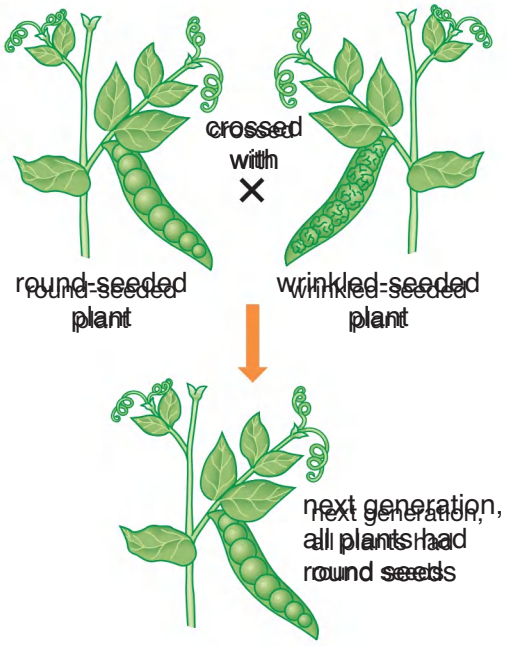


FIG. 9.6 Diagram showing investigation carried out with pea plants with round and wrinkled seeds

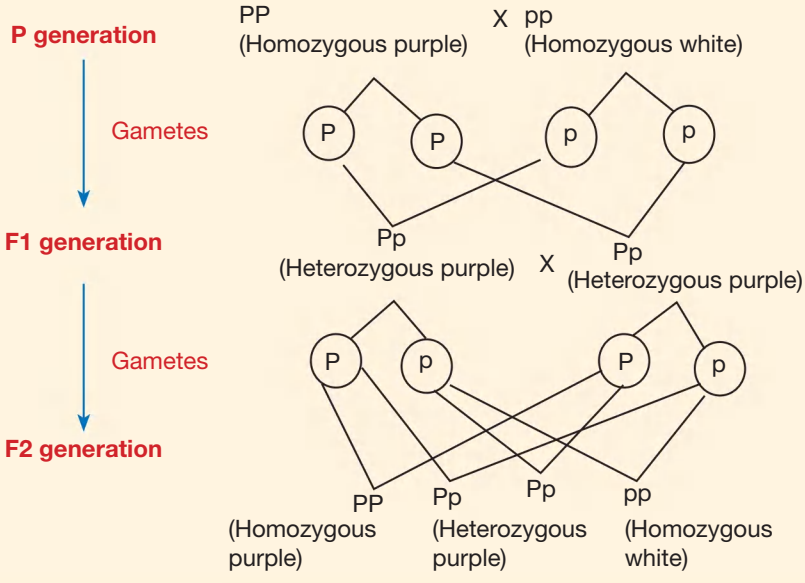


FIG. 9.7 Monohybrid crloss

Representation of Mendel's Monohybrid Cross by Checker Board (Punnett Square)

A Punnett square is a diagram that represents gametes of the concerned traits. Gametes of one parent are placed along one side and those of another parent are placed on the other side. In the sub-squares, possible combinations of genotypes are mentioned. the resulting phenotypes are mentioned under the respective genotype.

Table 9.6 Representation of a monohybrid cross

X	P (Dominant)	p (Recessive)
P	PP (Purple)	Pp (Purple)
P	Pp (Purple)	pp (White)

Phenotypic Ratio

Phenotypic ratio is the ratio of visible features. For example, for the Punnett square drawn above, phenotypic ratio is 3:1.

- 3 plants with purple flowers
- 1 plant with white flowers

Genotypic Ratio

Genotypic ratio is the ratio of genetic features. For example, for the Punnett square drawn above, genotypic ratio is 1:2:1.

- 1 homozygous purple-flowered plant
- 2 heterozygous purple-flowered plants
- 1 homozygous white-flowered plant

In order to test the homozygous or heterozygous nature of F1 generation progenies, Mendel performed experiments involving the crossing of the progeny (F1) with those of the parental generation.

Verification of Genetic Constitution of Progenies of F1 Generation

Back cross: The cross between the individual of F1 generation with either of the parents is known as back cross.

Test cross: The cross between the individual of F1 generation with the recessive parent is known as test cross.

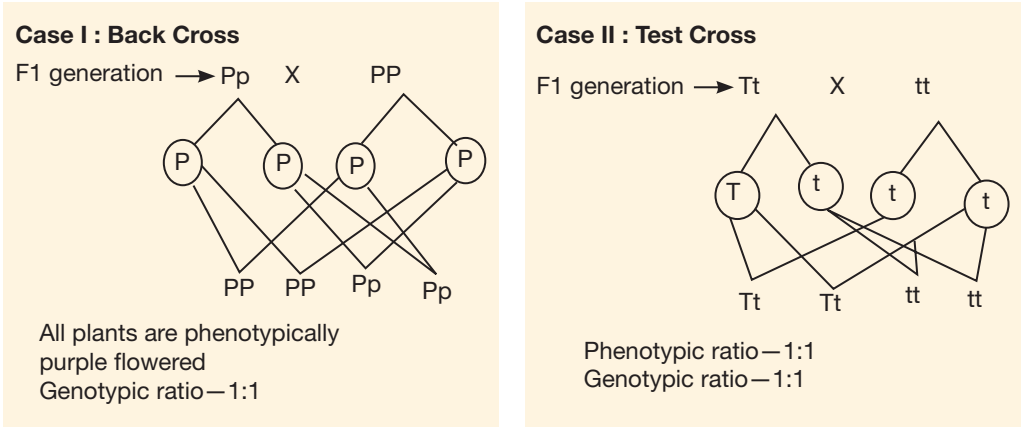


FIG. 9.8 Back cross and Test cross

Back Cross

F1 Gametes	Parental gametes: P (Dominant)	Parental gametes: P
P	PP (Purple)	PP (Purple)
p	Pp (Purple)	Pp (Purple)

Test Cross

F1 Gametes	Parental gametes: t (Recessive)	Parental gametes: t (Recessive)
T	Tt (Tall)	Tt (Tall)
t	tt (Dwarf)	tt (Dwarf)

Reciprocal Cross

A second cross of the same genotypes in which the sex of the individuals is reversed is known as a reciprocal cross.

	Male	Female	Offspring
Original cross	Tall (TT)	Dwarf (tt)	Tt
Reciprocal cross	Dwarf (tt)	Tall (TT)	Tt

Conclusions Drawn by Mendel on the Basis of Monohybrid Cross

On the basis of the results obtained for monohybrid cross, Mendel proposed three postulates. The latter two postulates are popularly known as Mendel's laws of inheritance.

Principle of paired factors: The inheritance of each character in an organism is controlled by two factors (genes) that lie on two homologous chromosomes and represent the same or alternative form of the same character (alleles)

Principle of dominance: When two contrasting factors are present in an organism, one of them may mask the presence of the other. The factor that expresses itself in the hybrid is called dominant allele and the factor that fails to express itself in the presence of dominant factor is called recessive allel.

Principle of segregation or purity of gametes: The two alternative factors of pair representing a character are separated from each other at the time of gamete formation, and hence, a gamete contains only one factor for the character.

Illustration

- P generation plants have same factors, and hence produce gametes with same factors (P or p).
- All F1 generation plants have purple flowers even though one of the parent plants has white flowers. That means, the factor or gene for the dominant trait is expressed out, and hence white colour is masked. However, all the plants possess the gene or factor for the recessive trait.

Mendel's Dihybrid Cross

A dihybrid cross involves breeding two pairs of contrasting characters.

- **P generation**: Plant with round and yellow seeds (RRYY) and plant with wrinkled and green seeds (rryy). Round shape of seed is a dominant trait. Yellow colour of seed is also a dominant trait. Wrinkled shape of seed and green colour of seed are the recessive traits.
- **F1 generation**: Cross-pollination between plant with round and yellow seeds and plant with wrinkled and green seeds resulted in all plants with round and yellow seeds. But these are hybrid plants with one factor for dominant trait and one factor for recessive trait for both the characters.
- **F2 generation**: Self-breeding of F1 hybrid plants resulted in plants having seeds with different combinations of traits.

Representation of Mendel's dihybrid cross by Checker Board (Punnett Square)

Phenotypic Ratio

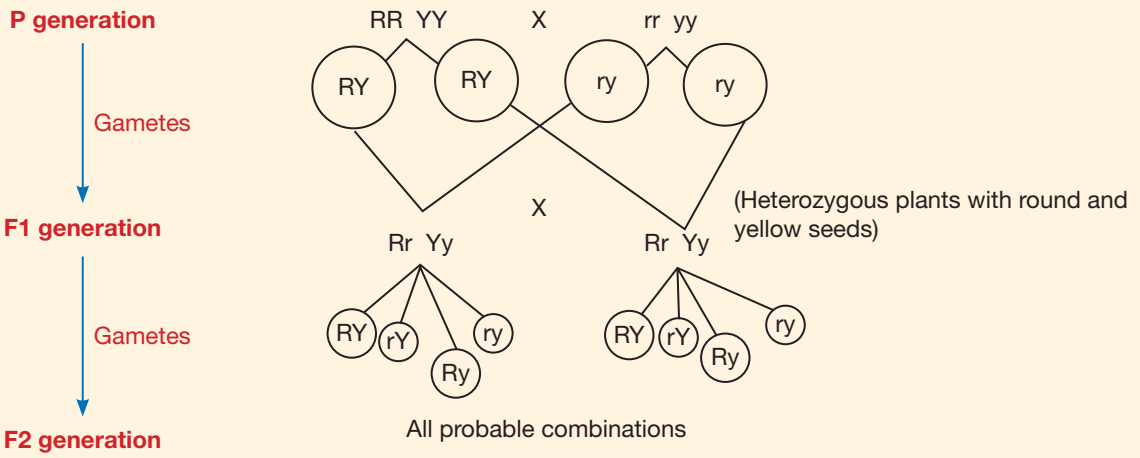
Phenotypic ratio for dihybrid cross in F2 generation—9:3:3:1

9 Plants	Round and Yellow seeds
3 Plants	Round and Green seeds
3 Plants	Wrinkled and Yellow seeds
1 Plant	Wrinkled and Green seeds

Genotypic Ratio

Genotypic ratio for dihybrid cross in F2 generation—1:2:1:2:4:2:1:2:1

1 Plant	Homozygous Round and Homozygous Yellow
2 Plants	Homozygous Round and Heterozygous Yellow
2 Plants	Heterozygous Round and Homozygous Yellow
4 Plants	Heterozygous Round and Heterozygous Yellow
1 Plant	Homozygous Round and Homozygous Green
2 Plants	Heterozygous Round and Homozygous Green
1 Plant	Homozygous Wrinkled and Homozygous Yellow
2 Plants	Homozygous Wrinkled and Heterozygous Yellow
1 Plant	Homozygous Wrinkled and Homozygous Green



RY	X	RY	→	RRYY	Ry	X	RY	→	RRYy
RY	X	rY	→	RrYY	Ry	X	rY	→	RrYy
RY	X	Ry	→	RRYy	Ry	X	Ry	→	RRyy
RY	X	ry	→	RrYy	Ry	X	ry	→	Rryy
rY	X	RY	→	RrYY	ry	X	RY	→	RyYy
rY	X	rY	→	rrYY	ry	X	rY	→	rrYy
rY	X	Ry	→	RrYy	ry	X	Ry	→	RrYy
rY	X	rY	→	rrYy	ry	X	ry	→	rryy

		Female gametes			
	X	**RY**	**rY**	**Ry**	**ry**
Male gametes	RY	RR YY	Rr YY	RR Yy	Rr Yy
	rY	Rr YY	rrYY	Rr Yy	rr Yy
	Ry	RR Yy	RrYy	RR yy	Rr yy
	ry	Rr Yy	rr Yy	Rr yy	rr yy

FIG. 9.9 Dihybrid cross

Conclusion Drawn by Mendel on the Basis of Dihybrid cross

Principle of independent assortment: If inheritance of two or more allele pairs at a time considered, their distribution in the gametes and in the subsequent generations takes place independent of each other.

Illustration

- All the plants of F1 generation produced Round and Yellow seeds since they are the dominant traits. But they were hybrid plants and possessed factors or genes for recessive traits also.
- In F2 generation, the plants obtained on self-pollination have different combinations, which proved that both the traits of the two characters are assorted independently, and hence plants with different kinds of seeds are produced.

Applications of Mendel's Laws of Inheritance

Mendel's Laws of Inheritance find applications in understanding various concepts of genetics. Some of them are listed below.

- New ideas regarding the new combination of hybrids could be generated
- Innovation of techniques of plant breeding and animal breeding for producing new breeds with desired traits
- Production of new varieties of plants with new combinations

Deviations from Mendel's Laws

Following are some of the demerits of Mendel's Laws of Inheritance.

- Mendel's laws were postulated much before the discovery of gene and DNA as the basis of inheritance. However, they were rediscovered by some scientists such as Hugo de Vries and others in the later period and then republished in a book called Flora.
- Scientist by name Thomas Morgan also conducted similar kind of experiments by taking fruit fly (*D. melanogaster*) instead of Mendel's garden pea and arrived at the same kind of conclusions as Mendel.
- At the same time, in some cases, it was found that the inheritance of characters did not exactly represent the Mendelian inheritance. They showed some deviations.
- The deviations are mainly of five kinds. They are as follows.

1. Incomplete dominance
2. Codominance
3. Multiple allele series
4. Pleiotropy
5. Polygenic traits

Incomplete Dominance

According to Mendelian inheritance, a cross of pure breeds (dominant and recessive) produces heterozygous individuals (F1 generation) that are phenotypically same as the dominant parent. However, in some cases, progenies produced in F1 generation do not resemble either of the parents in that character. That means, an intermediate expression between the dominant and recessive traits is expressed out in the heterozygous individuals.

For example, Snapdragon and 4 o'clock plant : Two types of pure breeds – Red flowers
White flowers

P generation	Red flowered		X	White flowered (Cross-pollination)
	RR			rr
F1 generation	Pink flowered		X	Pink flowered (self-pollination)
	Rr			Rr
F2 generation	RR	Rr		rr
	Red flowered	Pink flowered		White flowered

FIG. 9.10 Cross for incomplete dominance

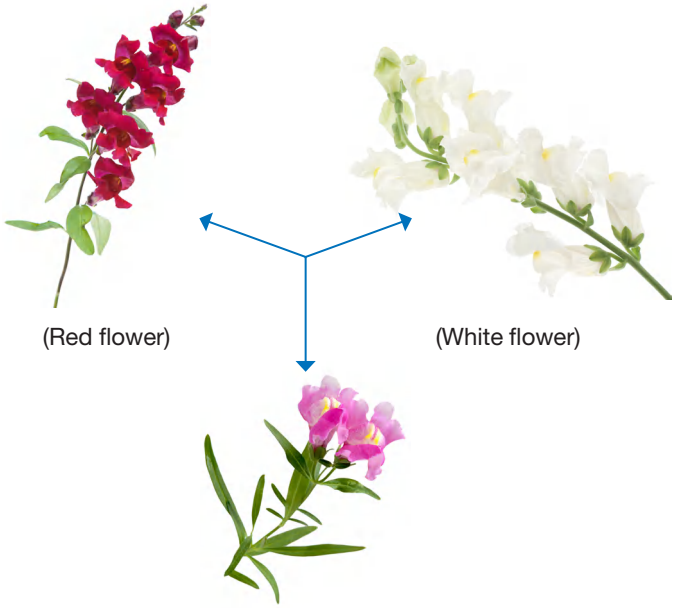


FIG. 9.11 Illustration of incomplete dominance

A cross of pure breeds of red-flowered (dominant) and white-flowered (recessive) plants produces all heterozygous progenies with pink-coloured flowers. Pink colour of flower is an intermediate expression between red colour and white colour of flower. The white colour of flower, which is a recessive trait, is expressed out in F2 generation just as in case of Mendelian monohybrid cross. However, the phenotypic ratio in this case is same as the genotypic ratio, that is, 1:2:1. The homozygous individuals possess red colour flowers as in P1 generation. But the heterozygous individual possesses pink colour as in F1 generation hybrids. The above results indicate that the dominant gene corresponding to red colour of flower shows incomplete dominance resulting in an intermediate expression of character.

Codominance

According to Mendelian inheritance, F1 generation individual resembles the dominant parent phenotypically and contains a recessive gene that is not expressed out. However, in some cases, the F1 generation individual resembles both the parents. That means, in heterozygous condition, the recessive gene also expresses itself along with the dominant gene. Such a phenomenon is called codominance. For example, presence of A; B; O blood groups in humans. The type of blood group is controlled by the gene 'I' that has three allelic forms, namely I^A; I^B; i. Alleles I^A and I^B produce different types of sugars and allele 'i' does not produce any sugar. I^A and I^B are completely dominant and 'i' is totally recessive and does not express itself in the presence of either I^A or I^B. When both I^A and I^B are present together, they both express their own type of sugars. Since three different alleles are there, six types of genotypes and four types of phenotypes are possible.

Table 9.7 Codominance in ABO blood groups

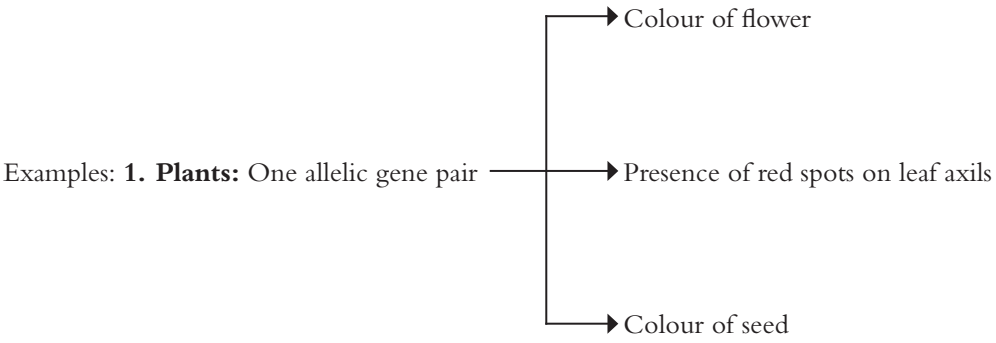
Allelic combination from parents	Genetic constitution of offspring (genoptype)	Blood group of offspring
I^A; I^A	I^AI^A	A
I^A; I^B	I^AI^B	AB
I^A; i	I^Ai	A
I^B; I^A	I^AI^B	AB
I^B; I^B	I^BI^B	B
I^B; i	I^Bi	B
i; i	ii	O

Multiple Allele Series

A single character is usually controlled by a pair of alleles that are contrasting traits. However, if a single character is controlled by more than two allelic genes, such a condition is known as multiple alleles.

Pleiotropy

According to Mendel, one allelic gene pair controls one specific character. The gene pair that controls height of the plant controls only that character. However, in some cases, one allelic gene pair may influence more than one character. Such phenomenon is called pleiotropy.



2. **Animals:** *Drosophila*—Gene responsible for white eye colour causes depigmentation in many parts of the body.
3. **Humans:** Sickle cell anaemia—Gene causes anemia and other problems such as muscle cramps, jaundice and kidney failure.

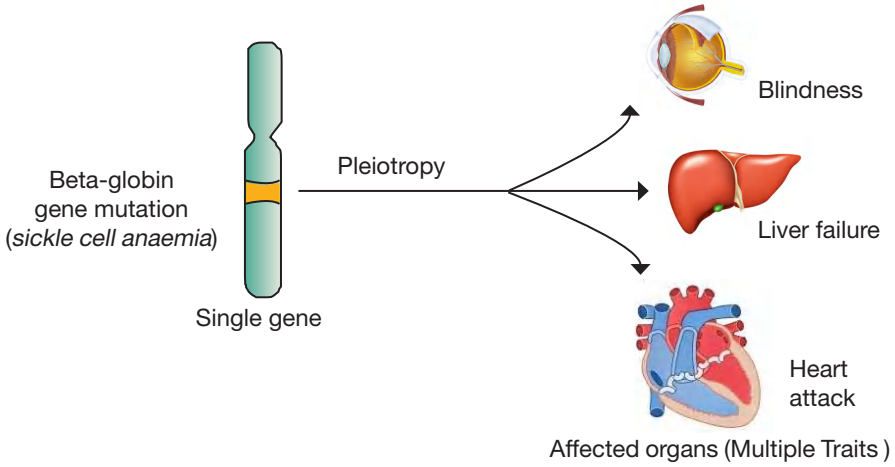


FIG. 9.12 Pleiotropic effects in sickle cell anaemia

Polygenic Traits

According to Mendel, the alternative forms of a single character are controlled by a single gene pair. The intermediate forms of the dominant and recessive traits were not considered. Such kind of traits are called monogenic traits or qualitative traits. However, on observation of different characters in various organisms, it is found that there are several traits that exhibit continuous gradation of the two alternative forms of the character under consideration. Such kind of traits are called polygenic traits or quantitative traits. These polygenic traits are controlled by more than one pair of genes and their additive effect results in individuals with a spectrum of phenotypes in contrast to monogenic traits that show clear and distinct phenotypes with contrasting traits.

Table 9.8 Some examples of polygenic traits

Plants	Animals
• Height of plant	• Skin colour in humans
• Seed size of beans	• Controlled by three pairs of genes
• Grain colour in wheat	• Human skin colour has number of intermediate shades from dark black to white.

APPLICATIONS OF GENETICS

Genetics not only deals with the transmission of characters and inheritance but also it has lot of practical applications in various fields such as plant breading and animal breading. Human genetics is a vast area that makes us understand the various genetic traits and diseases. The first and foremost application of genetics is in the determination of sex in different organisms.

Sex Determination

The sex of the progenies is determined on the basis of the inheritance or transmission of sex chromosomes to the progenies during gametic fusion. Different organisms show different kinds of sex determination mechanism depending on the nature of sex chromosomes in male and female organisms.

Types of Sex Determination

There are different types of sex determination methods depending upon the kind of organisms. Some of them are discussed below.

XX–XY Type Sex Determination

Most of the mammals and insects such as *Drosophila* show XX–XY type of sex determination. This mechanism of sex determination is called male heterogamety. The nucleus in human cell contains 46 chromosomes (23 pairs). Among these, 44 (22 pairs) are called autosomes. One pair of chromosomes is called sex chromosomes. Males possess one longer X chromosome and one shorter Y chromosome. Females possess two longer X chromosomes. During the formation of gametes, meiotic cell division takes place, and hence, the gametes formed possess 23 chromosomes (half the original number). Two types of gametes are formed in males, one carrying X chromosome and the other carrying Y chromosome. In females, only same type of gametes are formed that carry only one X chromosome each. During the process of fertilization, male and female gametes fuse to form a zygote that is diploid in nature. This restores the chromosome number. If male gamete with X chromosome fuses with any one of the female gametes, the zygote formed possesses both X chromosomes. If a male gamete with Y chromosome fuses with any one of the female gametes, the resultant zygote formed possesses one X and one Y chromosomes. The zygote with two X chromosomes (XX) develops into female, whereas the zygote with one X and one Y chromosome develops into a male. All the offspring invariably receive X chromosome from their mother. The offspring that inherits X chromosome from father will be a daughter. The offspring that inherits Y chromosome from father will be a son.

The characters that are linked to X chromosome or Y chromosome are transmitted to the next generation. Consequently, certain abnormal features are found in some persons. To a large extent, these are present in pairs.

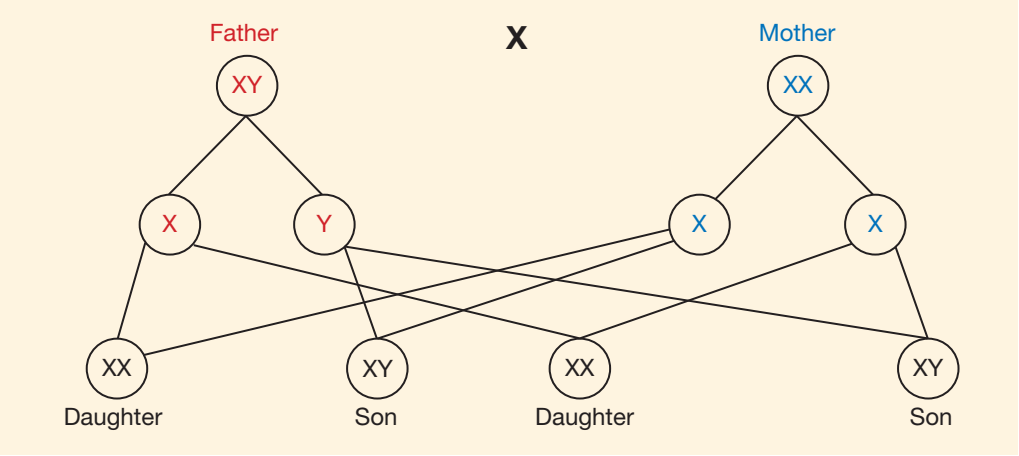


FIG. 9.13 Schematic representation—sex determination in man (XX–XY type)

Table 9.9 Example of certain abnormal traits found in humans

Trait	Dominant	Recessive
Hair	Presence	Absence
Ear lobes	Free	Attached
Tongue	Rolled	Not rolled
Tongue	Folding	Not folding

Info Box!

Organisms such as wasps and bees do not have sex chromosomes

XX–XO Type Sex Determination

Insects such as grasshopper have XO type mechanism of sex determination because the female has a pair of X chromosomes, whereas the male has a single X chromosome, and hence is represented as XO.

ZW–ZZ Type Sex Determination

In certain classes of organisms such as birds, the females possess two different types of sex chromosomes (ZW) and males possess two same types of sex chromosomes (ZZ). In these cases, the mechanism of sex determination is based on female heterogamety.

Aspects of Genetics Related to Gender/Sex of an Organism

Though most of the characters of an individual are passed on to the next generations without any relevance to the sex of the organism, there are certain traits that are exclusive to either male or female sex of an individual. Such traits that are related to the sex of an individual organism are categorized into three types which are given in the table.

Table 9.10 Traits related to the gender of an individual organism

Sex-limited traits	Sex-linked traits	Sex-influenced traits
These characters are not directly due to particular genes but are due to the sex hormones specific to male or female. For example: • Secretion of milk in female mammals • Development of beard and mustache in human males • Low-pitch voice in males	These are the traits that are determined by the genes present on the sex chromosomes For example: • Red-green colour blindness, haemophilia	These genes occur in both the sexes. But they are expressed in a particular sex For example: • Baldness in males

Sex-Linked Inheritance

Generally, according to the Mendelian inheritance, each gene has an allele for the same character on the homologous chromosome. But, this is not always the case with sex chromosomes. There are some genes that are located exclusively on X chromosome or Y chromosome in humans. These are called X-linked genes or Y-linked genes. In birds and other such species that possess Z chromosome, such genes are called Z-linked genes. The inheritance that exclusively involves X-linked or Y-linked or Z-linked genes from one generation to the next is called sex-linked inheritance.

Types of sex-linked inheritance are tabulated below.

Table 9.11 Types of X-lined inheritance

X-linked inheritance	Y-linked inheritance	XY-linked inheritance
Localized on non-homologous sections of X chromosome.	Localized on Y chromosome.	The genes that are located on the homologous part of X or Y chromosome are XY-linked genes.
No corresponding allele on Y chromosome.	Since males are heterozygous, no allele on the other chromosome. Such condition is called hemizygous.	These behave like autosomal genes.
X chromosome is common to male and female, and hence, the genes are called sex-linked genes.	Y-linked traits are less common and are transmitted to only male children.	
The characters controlled by these genes are known as sex-linked characters.	These genes are called holandric genes.	
The transmission of these sex-linked traits is known as sex-linked inheritance	This inheritance is known as holandric inheritance.	

Sex-Linked Inheritance in *Drosophila*

A mutant gene for producing white eye is found to be located on the X chromosome. This is recessive to dominant gene for red eye. The mutant recessive gene for white eye is represented as w and the dominant gene for red eye is represented as w^+. If both the X chromosomes in female possess alleles for white eye or red eye as the case may be, such a female is said to be homozygous for that trait (eye colour). If one X chromosome possesses allele for white eye and another X chromosome possesses allele for red eye, such a female is said to be hemizygous. However, in this condition, female does not phenotypically exhibit white eye as it is a recessive allele. Mother therefore acts as a carrier for that trait. Male possesses only one X chromosome and if it possesses allele for white eye, it is hemizygous and phenotypically exhibits white eye.

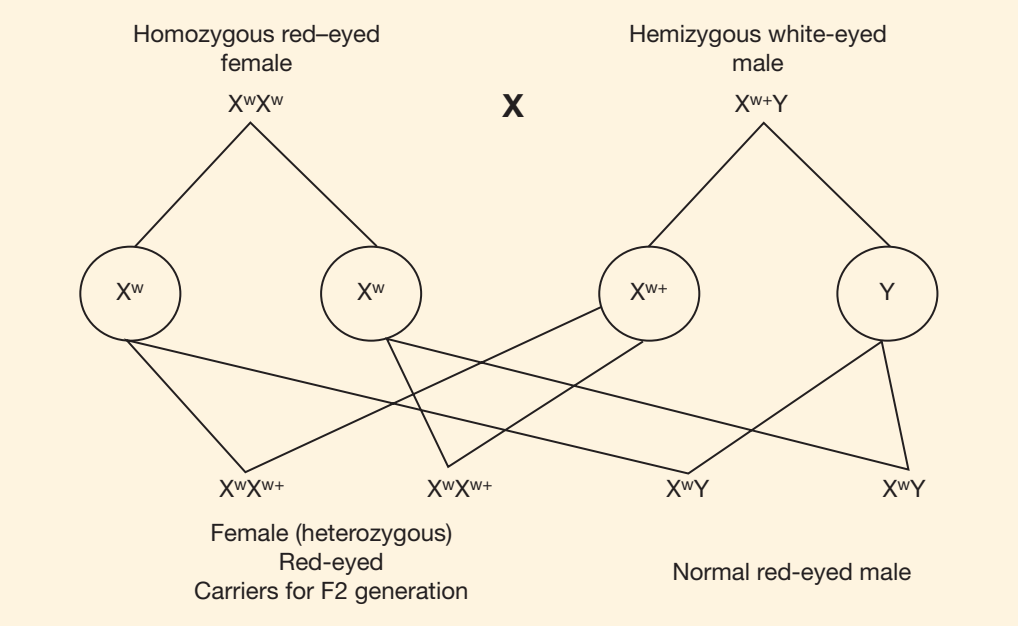


FIG. 9.14 Illustration of sex-lined inheritance

Characteristics of Sex-Linked Inheritance

Major features of sex-linked inheritance are listed below.

- The Y chromosome being confined only to one sex and being smaller with less number of genes, generally the inheritance of traits controlled by the genes located on the X chromosome of either male or female is only considered as sex-linked inheritance.
- This is a criss-cross inheritance. That means, fathers pass the trait to their grandsons through their daughters, whereas mothers pass the trait to both sons and daughters. That means, sons receive these genes only from mother, whereas daughters receive these genes from both mother and father.
- The phenotypic expression of the respective trait depends on whether it is X-linked dominant gene or X-linked recessive gene in case of females. In case of males, the presence of allele on X chromosome itself is sufficient to express the trait phenotypically irrespective of whether the gene is dominant or recessive. In case of females, if the gene is recessive, they act as carriers for that trait or disease.

Genetic Disorders

Certain diseases are passed on from one generation to the next generation because of the sex-linked inheritance. These are called sex-linked diseases or disorders. The presence of a gene with the disease on the homologous chromosome of mother or father may lead to an abnormality or disorder that can be passed on to the offspring. Such kind of disorders are called genetic disorders.

Types of Genetic Disorders in Humans

Depending on the nature, genetic disorders are broadly classified into two types which are listed below.

1. Chromosomal disorders
2. Mendelian disorders

Chromosomal Disorders

- These disorders are caused by the change in the structure or number of chromosomes.
- The chromosomes involved can be autosomes or allosomes (sex chromosomes).

Mendelian Disorders

- These disorders can be caused by the changes in the structure of gene.
- Such genes responsible for the disorder may appear in any generation due to various reasons. The occurrence of mutations is the most common reason for the disorder.

Mutations

Variations occur due to the recombination resulting due to crossing over during meiotic cell division. These are transmitted to the next generation during the formation of zygote (gametic fusion). However, some sudden variations may take place in a particular individual due to the change in the structure of a gene (DNA sequence) or chromosome or in the number of chromosomes. Such sudden changes are called mutations. These mutations may bring about a change in the genetic message of a cell thereby resulting in the consequent change in the phenotype of the respective organisms. Mutation is a random phenomenon that takes place spontaneously at a low frequency. Some external or environmental factors may induce mutations artificially in certain organisms. Such agents that cause mutations are called mutagens. Radiations, chemicals, and temperature are the important mutagens. The gene that undergoes change in structure is called mutant gene or mutant allele. These mutations may occur either on autosomes or on sex chromosomes. Depending on the type of chromosome with the mutant gene, Mendelian disorders can be classified into two types as listed below.

1. Autosomal disorders
2. Sex-linked disorders

If the mutant gene is present on the autosomes, it is called autosomal disorder. It can be autosomal dominant or autosomal recessive depending on whether the gene is dominant or recessive. Autosomal dominant or recessive disorders are heritable in case of both the sexes in equal frequency. That means, they are not sex specific.

If the mutant gene is present on the sex chromosomes, it is called sex-linked disorder. It is highly sex specific. If the mutant gene is present on X chromosome, it is called X-linked disorder. Depending on whether the gene is present on both X chromosomes or not, the disorder can be dominant or recessive. If the mutant gene is present on Y chromosome, it is called Y-linked disorder. Since only one Y chromosome is present, it is always dominant. X-linked recessive inheritance is much more common than X-linked dominant inheritance. Y-linked disorders are very few and are inherited only by sons. Females are not at all affected by such disorders.

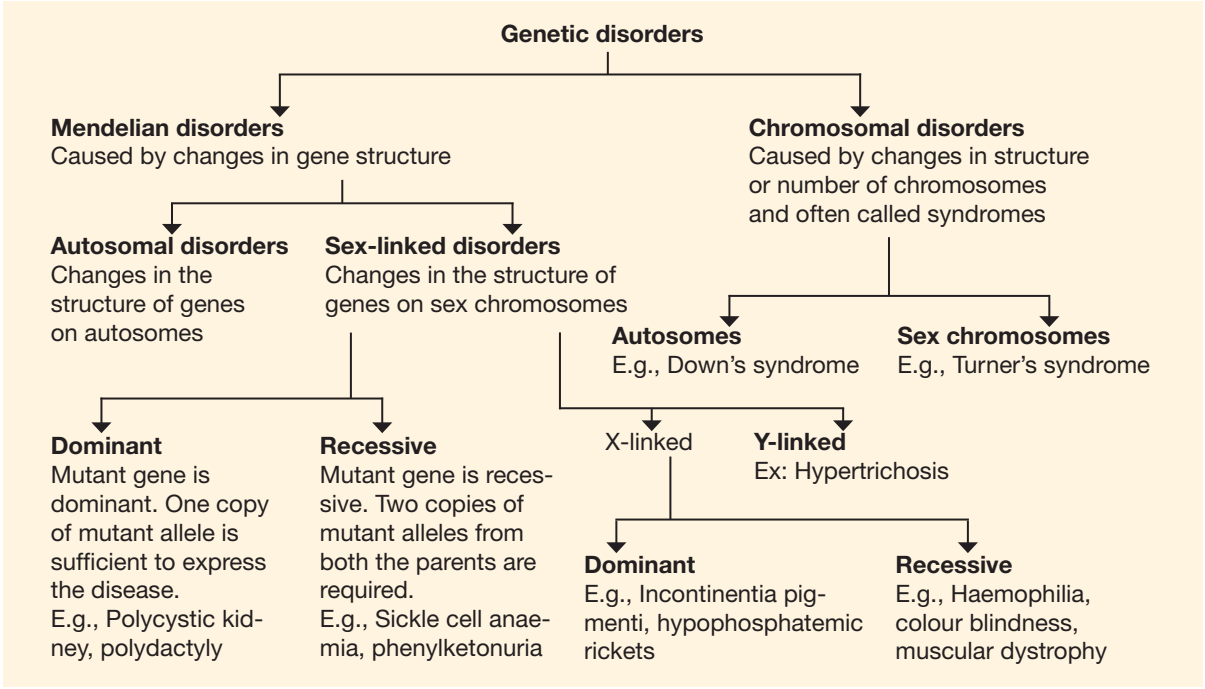


FIG. 9.15 Classification of genetic disorders

Illustrations of X-linked disorders

Case I : Mother with X-linked dominant disorder (consider normal father)

1. Daughters with gene on X (not affected by disorder)
2. Sons with gene on X (not affected by disorder 50%)
3. Daughters with gene on X (affected by disorder)
4. Sons with gene on X^+ (affected by disorder 50%)

Case II: Mother with X-linked recessive disorder (XX^+) (consider normal father)

1. Daughters with gene on X (no gene for disorder 25%)
2. Sons with gene on X (no gene for disorder 25%)
3. Daughters with gene on X^+ (recessive gene for disorder—carrier)
4. Sons with gene on X^+ (gene for disorder—affected by disease 25%)

Case III: Father with X-linked disoreder (X^+Y) (consider normal mother)

1. Daughter with X+ gene (affected by disorder 100%)
2. Sons with gene on Y chromosome (not affected by disorder 100%)

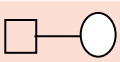
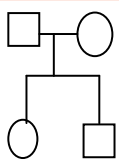
Pedigree Analysis

Pedigree is a family tree that describes the mode of inheritance of certain characteristics or diseases across the generations in that family. This analysis helps us to understand the family history and also predict the future of the family for that abnormal trait or disease as the case may be.

Symbols Used in Human Pedigree Analysis

Common symbols used in Pedigree analysis are given in the table below.

Table 9.12 Symbols used in pedigree analysis

Symbol	Representation
	Normal male
	Affected male
	Normal female
	Affected female
	Carrier female
	Normal offspring (male/female)
	Affected offspring (male/female)
	Mating
 1st 2nd	Parents Offspring

Info Box!

Haemophilia is referred to as the royal disease since it has been prevalent in European royal families

For example, three generation pedigree chart for the trait-tongue rolling.

Tongue rolling is an autosomal dominant disorder. Affected male or female is a non-roller, whereas normal male or female is a roller. There are no carriers for this trait. This is because presence of single mutant gene is sufficient to express this trait in an individual.

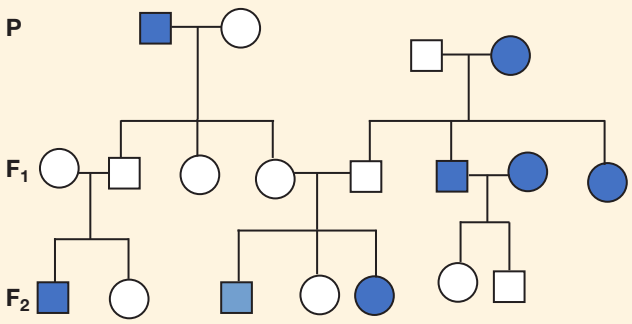


FIG. 9.16 Pedigree analysis of trait-tongue rolling

Illustrative examples of some commonly occurring sex-linked diseases in humans are given below.

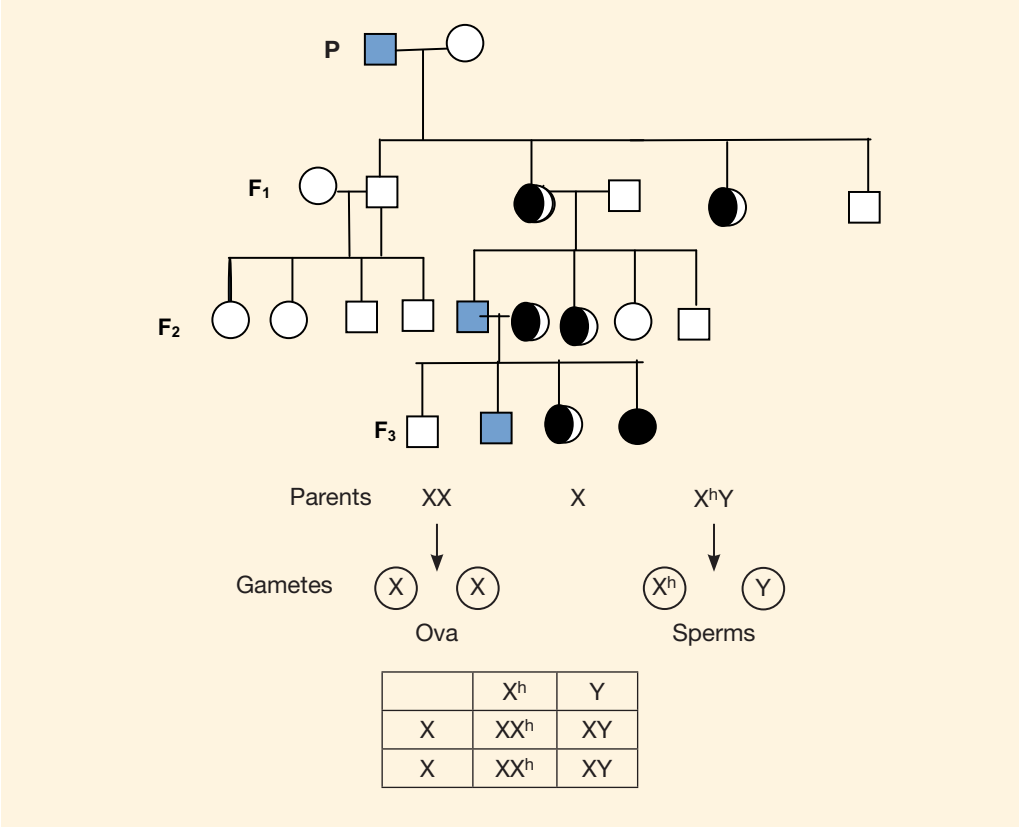
Haemophilia (X-linked Recessive Disorder)

This disease occurs due to the absence of a factor (gene) for clotting of blood and is called bleeder's disease. The patient continues to bleed even from a minor injury as the factor responsible for the synthesis of the substance required for clotting of blood is absent. This is caused by the presence of a recessive gene on the X chromosome. A woman is affected by the disease only in homozygous condition where both the X chromosomes possess gene for haemophilia. Generally, this condition does not sustain life and likely to end up in foetal stage. (in heterozygous condition, the affected gene is masked and the woman acts as a carrier [XXh]) A man is affected by the disease if he possesses X chromosome with gene for haemophilia (XhY). Otherwise, he is normal (XY).

F1 generation—Carrier daughters; Normal sons
Pedigree analysis/chart for haemophilia:
For example, cross between normal female and haemophilic male

Colour Blindness (X-Linked Recessive Disorder)

In this disease, the eyes fail to distinguish red and green colours. This condition is caused by a recessive gene present on the X chromosome. Females are affected by the disease only in homozygous condition when the mutant genes are present in both the X chromosomes. If they are in heterozygous condition, they act as carriers and are not affected by the disease. Males are affected by the disease if the lone X chromosome possesses the mutant gene for colour blindness.



	X^h	Y
X	XX^h	XY
X	XX^h	XY

FIG. 9.17

For example, cross between a normal man and a carrier woman

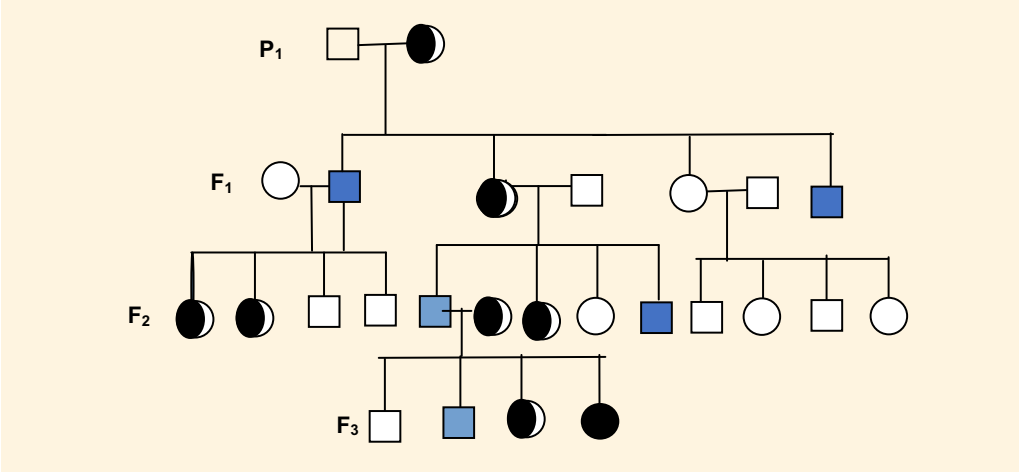


FIG. 9.18 Pedigree analysis of colour blindness

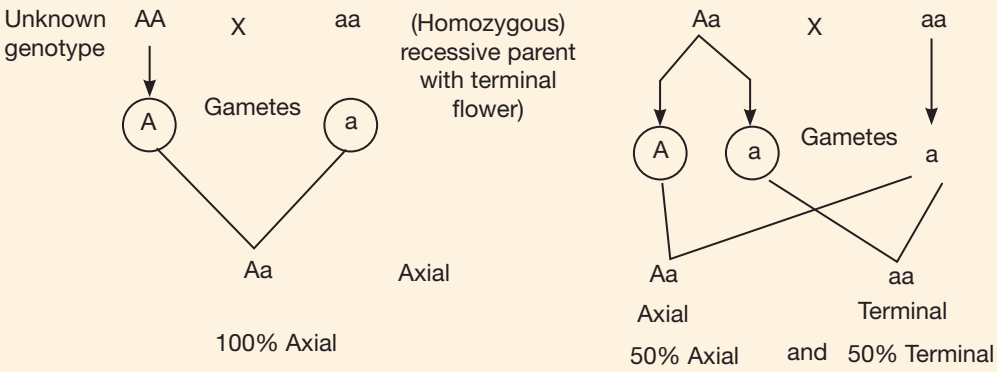
QUICK RECAP

1. Variations are produced in each generation in sexual mode of reproduction. Give reasons.

- Meiotic cell division involves exchange of genetic material between the homologous chromosomes.
- The variations occurring during meiotic cell division are transmitted to the next generation through gametic fusion.
- The variations produced during mitotic cell division may not be transmitted to the next generation.
- Consequently, progenies of each generation show some resemblances and some heritable and some non-heritable variations with those of next generation as well as with the individuals of the same generation.

2. A garden pea plant produces only axial flowers. How will you determine whether it is homozygous or heterozygous dominant? Explain with the help of crosses. How is codominance different from the above?

Usually, the unknown genotype of an individual is determined with the help of test cross. Test cross is the cross between individual with unknown genotype and its homozygous recessive parent. Genotype of a pea plant with axial flower can be either AA (homozygous dominant) or Aa (heterozygous dominant). There are two possibilities for a test cross as given below:



So from the above cross, it is clear that if pea plant with axial flower produces only axial flower, then the plant is homozygous dominant.

When the two alleles neither show dominant recessive relationship nor show intermediate condition but both of them express themselves simultaneously, this condition is known as codominance.

3. Heterozygous recessive progenies are not found in Mendelian inheritance. Justify.

A heterozygous progeny has a pair of dominant and recessive genes for the contrasting traits. A dominant gene always is expressed out and the recessive trait is masked. Hence, all heterozygous progenies exhibit the dominant traits only. They can never exhibit the recessive trait.

QUICK RECAP

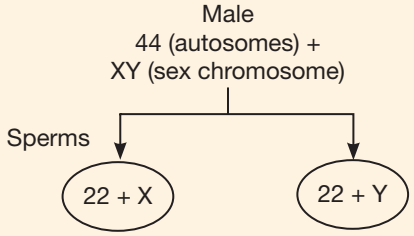
4. What is meant by mutation? How do mutations differ from variations? What is the significance of mutations?

Sudden change in the structure of chromosome that takes place in an individual organism is called mutation. Variations occur due to the recombination resulting due to crossing over during meiotic cell division. These are transmitted to the next generation during the formation of zygote (gametic fusion). These are gradual changes and some variations necessarily take place in every successive generation. Variations are necessary for evolution and are an integral part of sexual reproduction. Mutations are sudden changes taking place in a particular individual of any generation due to the changes in the structure or number of chromosomes or genes. They can occur either in autosomes or in sex chromosomes. The mutations occurring in chromosomes may or may not be expressed.

5. Human males are heterogametic. Justify.

In human male, cells contain one X and one Y as their sex chromosomes, that is, their cells have XY pair of sex chromosomes.

During gamete formation, male parent produces two types of sperms, that is, sperm having either 'X' as the sex chromosome or 'Y' as the sex chromosome. Hence, male is said to be heterogametic in nature (producing different types of gamete).

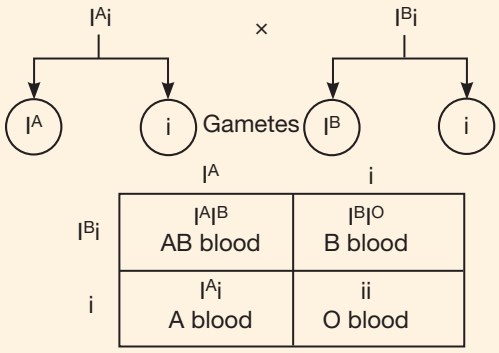


6. Explain how it is possible to have four children with different blood groups in one family.

Genotype of father can be either I^Ai or $I^A\ I^A$

Genotype of mother—I^Bi

Cross can be represented as below:



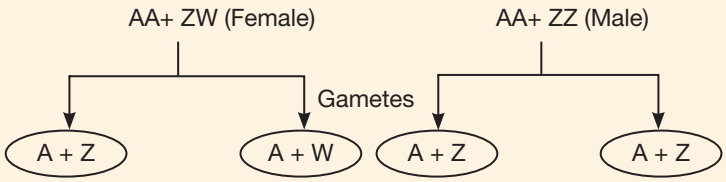
Blood groups of offspring can be A, B, AB, or O

QUICK RECAP

7. Explain mechanism of sex determination in birds

Sex determination in birds can be explained by ZW–ZZ mechanism. Here females are heterogametic (zw) and males are homogametic. Females produce two types of gametes; one with sex chromosome 'Z' and one with sex chromosome 'W.' Males produce single type of gamete with Z as sex chromosome.

Gamete formation can be represented as below:



8. Women are often blamed for giving birth to female babies. What is your opinion and how will you justify your statement?

Blaming women for giving birth to female children is wrong as in humans, sex of the baby is determined by father. More specifically, the sex of the baby is decided by the type of male gamete (sperm) fusing with female gamete (ovum). In human, males are heterogametic producing two types of gametes, that is, gamete with X chromosome and gamete with Y chromosome. Females are homogametic producing only one type of gamete. During fertilization, there is equal chance for the female gamete to fuse with either of the male gametes. According to this, sex of the baby is decided.

The two possible scenarios are given below:

(a) A + X (male) fuses with A + X (female) to give zygotes with AA + XX configuration. The individual with AA + XX chromosome will develop as female.

(b) A + Y (male) fuses with A + X (female) to give zygote with AA + XY configuration. The individual with AA + XY chromosome configuration will develop as male.

From the above scenarios, it is clear that in human beings, sex of the baby is determined by father.

9. A normal man and a normal woman got married. One female child was found to be colour blind and did not survive long, while all the rest of the children were normal.

(a) What conclusion can you draw from the above observation?

(b) Give justification for the conclusion.

(c) In F2 generation, if the colour blind woman marries a normal man, what is the probability of her daughters and sons being colour blind?

(a) The colour blindness of the female child may be because of mutation.

(b) Colour blindness is an X-linked disorder. Females exhibit the disease or trait only in homozygous condition. Even if the mother is a carrier for the gene, daughter should get only one X chromosome with the gene for the disease. In that case, she can be a carrier. She cannot be affected by the disease. Since one daughter is affected by the disease, it can be presumed that the condition would have been due to mutation in that particular individual.

QUICK RECAP

(c) The colour blind woman should have mutant genes on both the X chromosomes. All sons would be colour blind as they get any one of the X chromosomes from mother. Since the father has normal genes on X chromosome, the daughters get only one X chromosome with mutant gene from mother. As a result, all the daughters will be carriers. None of the daughters are colour blind or normal.

10. Explain haplodiploid type of sex determination

It is a type of sex determination in which the male is haploid whereas the female is diploid. Haplodiploidy occurs in some insects such as bees and ants. In honeybee, males are haploid as they develop parthenogenetically from eggs without fertilization. Females grow from fertilized egg and are hence diploid.

POINTS TO REMEMBER

- The branch of biology that deals with the study of heredity and variations is called genetics.
- This transmission of characters and variations along the forthcoming generations is known as inheritance.
- The inheritance of characters from the parents to the offspring is known as heredity.
- The chromosomes present in the nucleus of living cells serve as the vehicles of transmission of characters.
- Genes are the physical units of heredity and DNA is the molecular or chemical unit of heredity.
- The genes (factors) representing a particular character have their contrasting forms that are called alleles.
- Phenotypic ratio is the ratio of visible features.
- Genotypic ratio is the ratio of genetic features.
- The cross between the individual of F1 generation with the recessive parent is known as test cross.
- One allelic gene pair may influence more than one character and this phenomenon is called pleiotropy.
- Chromosomal disorders are caused by the change in the structure or number of chromosomes.
- Mendelian disorders are caused by the changes in the structure of gene.

PRACTICE QUESTIONS

TEST YOUR CONCEPTS

Directions for questions from 1 to 28: Fill in the blanks in each question.

1. The chemical unit of heredity is _____.
2. Chromosomes are visible in the nucleus only during the process of _____.
3. _____ division is also called reduction division.
4. A fragment of DNA that provides complete information about one protein is referred as _____ for that protein.
5. _____ is the degree of differences between parents and offspring.
6. _____ are different forms of a particular gene that control the same character..
7. The study of heredity and variations along the generations is known as _____.
8. _____ are the molecular units of heredity.
9. Self-pollination of homozygous plants gives _____ plants in F1 generation.
10. Genes responsible for certain traits were called _____ by Mendel.
11. Mendel's monohybrid test cross ratio was _____.
12. Phenotypic ratio for incomplete dominance is _____.
13. Gene controlling starch synthesis in pea plant is an example for _____.
14. The scientific name of the plant on which Mendel worked is _____.
15. In human cell, there are _____ autosomes and _____ sex chromosomes.
16. The cross of an F1 plant with any one of its parents is called _____.
17. Mendel's monohybrid test cross ratio was _____.
18. In ABO blood group system allele I^A is _____with I^B allele.
19. The type of inheritance associated with the homologous part of the sex chromosomes is called _____.
20. The genes located on Y chromosomes are called _____.
21. Genetic disorders occurring due to the change in the structure or number of chromosomes are called _____.
22. _____ gives the analysis of future of the family with respect to the particular sex-linked trait.
23. Muscular dystrophy is caused due to the mutant _____ gene on _____ chromosome.
24. The traits that are not due to specific genes, but are affected by sex hormones are called _____traits.
25. Males are said to be _____ for genes on the X chromosome.
26. In colour blindness, the affected person fails to distinguish between _____ and _____ colours.
27. *Drosophila* shows _____ type of sex determination.
28. Non-stop bleeding is the characteristic symptom for the disease _____.

Directions for questions from 29 to 47: For each of the following questions, for choices have been provided. Select the correct alternatives.

29. Identify the phenomenon that serves to preserve the parental characteristics in the next generation.
    (a) Crossing over (b) Recombination
    (c) Linkage (d) DNA replication
30. The ratio of dihybrid test cross is
    (a) 3 : 1 (b) 1 : 2 : 1
    (c) 1 : 1 (d) 1 : 1 : 1 : 1

31. In the phenotypic ratio 9:3:3:1, 9 stands for
(a) Homozygous dominant
(b) Heterozygous dominant
(c) Homozygous recessive
(d) Heterozygous recessive

32. Identify the combination that is not possible.
(a) Homozygous dominant
(b) Heterozygous dominant
(c) Homozygous recessive
(d) Heterozygous recessive

33. Which of the following represents recombination with respect to both the traits?
(a) RRYY (b) rrYy
(c) RrYy (d) RrYY

34. Which of the following is the characteristic of a pleiotropic gene?
(a) Controls sexual characters
(b) Presents only in prokaryotes
(c) Controls one character in association with the other
(d) Control more than one character

35. How many autosomes are found in human ovum?
(a) 44 (b) 22
(c) 23 (d) 46

36. Which among the combinations represents phenotypically similar, but genotypically different plants?
(a) RRYY and RrYy
(b) Rryy and rrYY
(c) rrYY and rryy
(d) RrYY and rrYy

37. When 480 plants are produced in the F2 generation of dihybrid cross made with pure breeding plants, the total number of plants with parental combinations obtained is
(a) 300 (b) 270
(c) 30 (d) 180

38. Red (RR) flower plant is crossed with white (rr), all the offspring were pink, this indicates that R gene is
(a) Dominant
(b) Recessive
(c) Incomplete dominant
(d) Codominant

39. Two snapdragon plants with pink flower were hybridized. The F1 plants produced red, pink, and white flowers in the proportion of 1 red, 2 pink, and 1 white. What could be the genotype of the two plants used for hybridization? (Red flower colour–RR white–rr)
(a) rr (b) Rr
(c) RR (d) RRRR

40. Predict the number of squares in the Punnett square of a trihybrid cross.
(a) 16 (b) 64
(c) 27 (d) 81

41. In honeybee, males are
(a) Haploid (b) Diploid
(c) Triploid (d) None of the above

42. In grasshopper, total number of chromosomes in male is
(a) Equal to female
(b) Less than female
(c) Double than female
(d) More than female

43. In which of the following organisms, female heterogamety is observed?
(A) Peacock (B) Crocodile
(C) Butterfly (D) Rabbit
(E) Fish
(a) A, B, C (b) B, C, D
(c) A, B, E (d) B, C, E

44. Identify the feature for which genes are present in both the sexes, but is predominantly sex specific in humans.
(A) Bald head
(B) Secretion of milk

PRACTICE QUESTIONS

(C) Skin colour
(D) Colour blindness
(E) Development of mustache
(a) A, B (b) A, D
(c) A, B, E (d) B, C, E

45. Identify the diseases that are not sex specific.
(A) Hypertrichiosis (B) Haemophilia
(C) Phenylketonuria (D) Pentadactyly
(E) Muscular dystrophy
(a) A, B, E (b) C, D
(c) C, D, E (d) A, E

46. Which of the following type of sex determination involves organisms with different number of chromosomes in males and females?
(a) XX–XY (b) XX–XO
(c) ZZ–ZW (d) None of these

47. Albinism is a genetic disorder. But it affects both eye sight as well as hearing. Which of the following term can describe this condition?
(a) Codominance
(b) Polygenic traits
(c) Incomplete dominance
(d) Pleiotropy

Directions for questions 48 and 49: Match the entries of Column 1 with those of Column 2.

48.

Column 1	Column 2
A. A single gene affecting number of characters	(i) Polygenic inheritance
B. Spectrum of phenotypes	(ii) Codominance
C. Intermediate phenotypic expression	(iii) Pleiotropy
D. ABO blood groups	(iv) Incomplete dominance

(a) A→ (iii), B→ (i), C→ (iv), D→ (ii)
(a) A→ (ii), B→ (i), C→ (iv), D→ (iii)
(a) A→ (i), B→ (iii), C→ (iv), D→ (ii)
(a) A→ (iv), B→ (i), C→ (iii), D→ (ii)

49.

Column 1	Column 2
A. Birds	(i) One gene controls more than one character
B. True bugs	(ii) ZW–ZZ sex determination
C. Pleiotropy	(iii) More than two alleles for a gene
D. Multiple allelism	(iv) XX–XO type

(a) A→ (iii), B→ (i), C→ (iv), D→ (ii)
(a) A→ (ii), B→ (i), C→ (iv), D→ (iii)
(a) A→ (i), B→ (iii), C→ (iv), D→ (ii)
(a) A→ (iv), B→ (i), C→ (iii), D→ (ii)

PRACTICE QUESTIONS

MASTERING THE CONCEPTS

Knowledge and Understanding

1. What are homologous chromosomes alleles? Give examples.

2. Define the following terms:
(a) Inheritance
(b) Heredity
(c) Trait
(d) Gene
(e) Variations

3. Distinguish between linkage and crossing over.

4. What is meant by recombination? Explain its significance.

5. Distinguish between dominant trait and recessive trait.
6. Distinguish between homozygous and heterozygous.
7. Distinguish between monohybrid cross and dihybrid cross.
8. Explain F_1 and F_2 generations.
9. What do you mean by a true breeding plant?
10. Define multiple alleles.
11. Define linkage.
12. Mention the pairs of contrasting characters chosen by Mendel related to seed in pea plant.
13. Mention the pairs of contrasting characters chosen by Mendel related to flower in pea plant.
14. What is test cross and what is its significance?
15. Differentiate between dominance and codominance.
16. RRYY with rryy plants in P generation are crossed and the plants produced in F1 generation are allowed to self-pollinate.
    (a) Mention the gametes formed by each parent in both the generations?
    (b) What do they represent?
    (c) Mention the two types of gametic combinations possible.
17. In pea plants, axial position of flowers is considered as dominant trait and terminal position of flower is considered as recessive trait. Heterozygous plant with terminal flower is not possible.
18. Taking seed colour of pea plant as the trait (yellow as dominant and green as recessive), design a test cross to show the nature of P1 generation plants.
19. Distinguish between autosomal genetic disorders and allosomal genetic disorders.
20. What is meant by criss-cross inheritance? Justify the mode of inheritance by taking example of any trait.
21. The sex of a child is determined by the father and not mother. Give reason.
22. Y-linked traits are very less common than X-linked traits. Give reason.

Application and Analysis

23. Show that in incomplete dominance, the phenotypic and genotypic ratio is same.
24. What would be the possible blood groups of the progeny, if mother is O blood group and father is AB blood group?
25. What would be the blood groups of the progeny of father with blood group 'A' and mother heterozygous B? Explain the cross.
26. With the help of a Punnett square, explain Mendelian experiment where a plant bearing green pods (dominant) is crossed with a plant bearing yellow pods. Write both the phenotypic and genotypic ratios for F2 generation.
27. An experiment is conducted with two sets of plants with red flowers. One set of plants gave rise to progenies with only red flowers in all successive generations. Another set of plants when crossed gave rise to few plants with white flowers also in F2 generation. What conclusions can you draw regarding the plants of P1 generation?
28. Tallness of plant and purple colour of flower are the dominant traits. Dwarfness of plant and white colour of flower are the recessive characters. These two plants are crossed.
    (a) What percentage of dwarf plants are produced with purple flowers and white flowers in F2 generation?

PRACTICE QUESTIONS

(b) Which type of plants are produced in maximum numbers? Predict their percentage.

(c) Subdivide the above category of plants on the basis of their genotypes.

(d) Explain by using Punnett square.

29. Plant with round seeds is crossed with plant with wrinkled seeds. All F1 generation progenies are with round seeds.

(a) What is the percentage of plants with wrinkled seeds in F2 generation?

(b) What is the percentage of hybrid plants in F2 generation?

(c) Justify the above results by drawing Punnett square.

30. Plant with full green pod is said to be homozygous dominant. Plant with constricted yellow pod is said to be homozygous recessive. Crossing of these two plants can also give rise to plants with full yellow pod. Plants with constricted green pod are also produced.

(a) What conclusion could Mendel draw from this observation?

(b) Explain with the help of appropriate diagram.

31. Mendel produced 556 pea plants in his dihybrid cross. What are the various types of plants produced in F2 generation? What are their respective numbers? Justify.

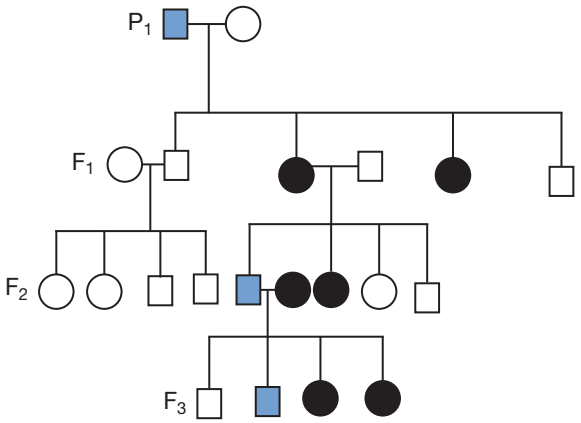
32. Mendel produced 929 pea plants in his experiment for monohybrid cross considering red and white colours. How many red-coloured plants are produced in F2 generation? Are all of them same? Justify.

33. Red-flowered plants of F1 generation plants are different from the red-flowered plants of P1 generation. Give reason.

34. Human beings are found to possess variable skin complexion. Give reason.

35. A person affected by sickle cell anemia may experience problems in multiple organs. Give reason.

36. Study the following pedigree chart and answer the following questions:



(a) If the disease is expressed in mother, predict the type of genetic disorder represented by the given pedigree chart.

(b) What is the status of sons in F1 generation with respect to the disease? Justify.

(c) In which cases, the successive generations will be totally free of the diseased genes?

(d) Continue the given pedigree chart for F2 generation of B.

(e) Which sex is more affected by the disease? Give justification.

37. How is it possible to conclude whether a particular trait is X-linked or Y-linked?

38. A man suffers from muscular dystrophy. The woman is normal and had no family history for the disease.

(a) Can all her daughters be normal?

(b) Will the successive generations be free of the disease? Discuss different cases considering mating with normal men or women in the later generations.

(c) Draw pedigree chart for three generations and justify the type of inheritance of the disease.

39. Distinguish between Down's syndrome and Turner's syndrome and explain the nature of these diseases with respect to the concept of genetics.

40. Father has blood group B and mother has blood group A. Predict the probable blood group of the child. Justify.

PRACTICE QUESTIONS

41. Sickle cell anemia and haemophilia are disorders of blood and are found to be genetically transmitted. However, sickle cell anemia is found in both the sexes, whereas haemophilia is usually rare in females. Justify by using pedigree analysis.

42. A child possesses some characteristic resemblances with both father and mother. However, the child is not an exact copy of either of the parents. Give reason.

43. Secretion of milk is the characteristic feature of only female mammals. Give reason.

44. In *Drosophila*, presence of white eye is more predominant in males than in females. Justify with respect to the concept of genetics.

45. Consanguineous marriages are not suggestible from the point of view of genetics. Justify by using pedigree analysis.

Assertions and Reasons

Directions for questions from 46 to 58: Choose the correct option.

(a) Both A and R are true and R is the correct explanation for A.
(b) Both A and R are true, but R is not the correct explanation for A.
(c) A is true and R is false.
(d) A is false and R is true.

46. **Assertion (A):** Sexual reproduction brings about appreciable variations in the offspring
Reason (R): Sexual reproduction involves cell division in which DNA replication takes place.

47. **Assertion (A):** The DNA of the child will be the same as the DNA of the parent.
Reason (R): The two DNA strands are complementary to each other.

48. **Assertion (A):** Heterozygous recessive progenies are not found in Mendelian inheritance.
Reason (R): Formation of gametes is associated with the segregation of alleles.

49. **Assertion (A):** Red-flowered plants of F1 generation are same as red-flowered plants of P generation.
Reason (R): All the offspring obtained in F1 generation are phenotypically and genotypically same.

50. **Assertion (A):** F1 generation progenies can show an intermediate expression between dominant and recessive characters.
Reason (R): The cross involves the phenomenon of codominance.

51. **Assertion (A):** Children necessarily possess the same blood group as either of the parents.
Reason (R): The gene corresponding to the blood group has three alleles.

52. **Assertion (A):** A cross between tall plants with round seeds can also produce dwarf plants with wrinkled seeds.
Reason (R): Each progeny inherits one homologous chromosome each from male and female.

53. **Assertion (A):** In grasshopper, male individual determines the sex of the offspring.
Reason (R): All organisms show male heterogamety.

54. **Assertion (A):** Genes on the X chromosome are called sex-linked genes.
Reason (R): X chromosome is present in both the sexes.

55. **Assertion (A):** Males cannot act as carriers for genetic disorders.
Reason (R): Males cannot transmit the X-linked genes to their sons.

56. **Assertion (A):** Hypertrichosis is a disease found only in males.
Reason (R): Y-linked traits cannot be transmitted to daughters.

57. **Assertion (A):** For sickle cell anemia, males also can be carriers.
Reason (R): Autosomal recessive traits are not sex specific.

58. **Assertion (A):** Sex of a child is determined by father and not mother.
Reason (R): Father is heterogametic.

ASSESSMENT TESTS

PRACTICE QUESTIONS

Directions for questions from 1 to 4: For each of the following questions four choices have been provided. Select the correct alternative.

1. Identify the plant that exhibits the same characters as represented by TTAA.

(a) Ttaa (b) Ttaa

(c) TtAa (d) ttAA

2. The percentage of heterozygous plants obtained in the F2 generation of monohybrid cross is

(a) 25 per cent (b) 50 per cent

(c) 75 per cent (d) 100 per cent

3. When 500 plants are produced in the F2 generation of monohybrid cross, the number of homozygous recessive plants obtained is

(a) 500 (b) 375

(c) 250 (d) 125

4. Which of the following is an abnormal trait in human beings?

(a) Attached ear lobes

(b) Tongue rolling

(c) Presence of hair

(d) Folding tongue

Directions for questions from 5 to 9: Correct the following statements.

5. The number of chromosomes in a particular species is variable.

6. The proportion of plants heterozygous with respect to both the traits in F2 generation of Mendel's dihybrid cross is 3/16.

7. The probability of birth of male child in humans is 25 per cent.

8. Human Y chromosome is longer than X chromosome.

9. Autosomes are responsible for the transmission of characters from parents to the offspring.

Directions for questions from 10 to 17: Write the missing correlated terms.

10. Monohybrid cross: 3:1::Dihybrid cross: _____

11. Tall : dominant :: _____ : recessive

12. RR : homozygous :: _____ : heterozygous

13. _____ : Dominant :: Wrinkled seed : recessive

14. Phenotypic ratio : 3 : 1 :: Genotypic ratio : _____

15. XX : female :: _____ : male

16. Homozygous dominant : _____ :: Homozygous recessive : 1

17. TTPP : Parental combination :: _____ : Recombination

Directions for questions from 18 to 23: Find the Odd one.

18. Chromosome; DNA; Gene; Nucleus

19. PR; pr; RY; Ry

20. R; r; Yy; T

21. RRyy; RrYy; RrYY; RRYY

22. Polycystic kidney; Polydactyly; Phenylketonuria; Haemophilia

23. Crow; *Drosophila*; Whale; Man

Directions for questions from 24 to 35: Answer the following questions.

24. What is a reciprocal cross?

25. What is law of segregation?

26. Explain law of dominance with an example.

27. Explain test cross and back cross.

28. What is pleiotropy? Give one example.

29. When a cross is made between tall plant with green pod (TtGg) and a tall plant with yellow pod (Ttgg), what proportion of phenotype in an offspring could be

(a) Tall green

(b) Dwarf green

30. Analyze the progenies of F1 generation in the following cases:

(a) Homozygous tall and homozygous dwarfs

(b) Homozygous tall (dominant) and heterozygous tall

(c) Heterozygous tall and heterozygous tall

(d) Heterozygous tall and homozygous dwarf

31. Distinguish between allosomes and autosomes.

32. What are mutagens? Name some mutagens.

33. Give the suitable term for the following:

(a) Condition in which one X chromosome has a gene for particular trait and the other one has no allele for the trait.

(b) A fragment of DNA helix that determines a particular character of an individual.

(c) The gene present in an individual but not expressed out.

(d) A pair of chromosomes having contrasting alleles on each.

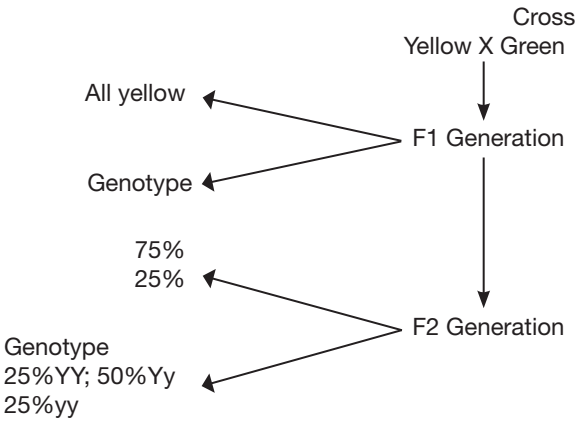
(e) The condition in which one gene affects more than one trait.

(f) The traits that are not sex specific.

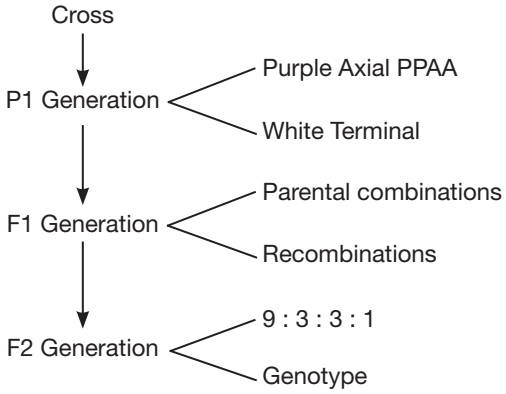
(g) The condition in which both traits are expressed in heterozygous condition.

(h) The progenies formed due to asexual reproduction.

34. Fill in the blanks in the following flow chart with appropriate terms. (yellow—dominant; green—recessive)



35. A plant with purple-coloured axial flowers is crossed with a plant with white-coloured terminal flowers. Purple colour and axial position of flowers are dominant traits, whereas white colour and terminal position of flowers are recessive traits. Fill in the blanks in the following flow chart with the appropriate terms for the above cross.



PRACTICE QUESTIONS

HINTS AND EXPLANATION

TEST YOUR CONCEPTS

1. DNA
2. Cell division
3. Meiotic
4. Gene
5. Variation
6. Alleles
7. Genetics
8. Genes
9. Hybrid
10. Factors
11. 1 : 1
12. 1 : 2 : 1.
13. Pleiotropy
14. Pisum sativum
15. 22 pairs of, One pair of
16. Back cross
17. 1 : 1
18. Codominant
19. XY-linked inheritance
20. Holandric genes
21. Syndromes
22. Pedigree analysis
23. Recessive, X
24. Sex-influenced
25. Hemizygous
26. Red, Green
27. XX–XY
28. Haemophilia
29. (c)
30. (d)
31. (b)
32. (d)
33. (c)
34. (d)
35. (b)
36. (a)
37. (a)
38. (c)
39. (b)
40. (c)
41. (a)
42. (b)
43. (c)
44. (a)
45. (b)
46. (b)
47. (d)
48. A → (iii), B → (i), C → (iv), D → (ii)
49. A → (ii), B → (iii), C → (i), D → (iv)

MASTERING THE CONCEPTS

Knowledge and Understanding

1. The chromosomes occur in pairs in each cell. The two chromosomes of the pair are similar in structure (gene position) and are derived one from each parent. Such pairs of chromosomes are known as homologous chromosomes.

Alleles: The genes (factors) representing a particular character as contrasting traits are called alleles. The alternative forms of a character are called contrasting traits.

2. (a) **Inheritance:** The transmission of characters and variations along the forthcoming generations.

(b) **Heredity:** The inheritance of characters from the parents to the offspring.

(c) **Gene:** The fragment or region of DNA that is made up of specific sequence of nucleotides is called gene, which is the molecular unit of heredity.

(d) **Trait:** The character or feature that is carried by a gene such as height of plant.

(e) **Variations:** The differences existing among the individuals of a species and also among the offspring of the same parents.

3.

Linkage	Crossing over
Tendency of genes on a chromosome to remain together and pass as such to the next generation.	Exchange of genes to establish a new linkage.
Helps to preserve the parental gene combinations.	Helps to bring about recombinations.
Generally more favorable among the genes that are closely placed on a chromosome.	Generally favorable among the genes that are distantly placed on the chromosomes.
The probability of linkage increases with increase in age.	The probability of crossing over decreases with increase in age.

4. The phenomenon of the exchange of segments of non-sister chromatids (genes) of homologous chromosomes is known as crossing over. Crossing over is followed by recombination during the meiotic cell division. The reproductive cells (diploid germ cells) undergo meiotic cell division producing haploid (n) male and female gametes. That means, meiotic cell division involves genetic combination that results in the exchange of some genes and forms the main basis for the variations among the organisms of same species. Due to genetic recombination taking place in meiotic cell division, the exchange of genetic material takes place between the chromosomes. This results in variations that are inherited because the union of male and female gametes results in the formation of diploid zygote.

5.

Dominant Trait	Recessive Trait
The trait that expresses itself even in the presence of its contrasting trait as it masks and hides the other trait. For example: Purple colour of flowers	The trait that fails to express itself in the presence of its contrasting dominant trait. For example: White colour of flowers

6.

Homozygous	Heterozygous
An organism having the traits that carry either both dominant or recessive factors. For example: RR	An organism having contrasting factors (one dominant factor and one recessive factor). For example: Rr

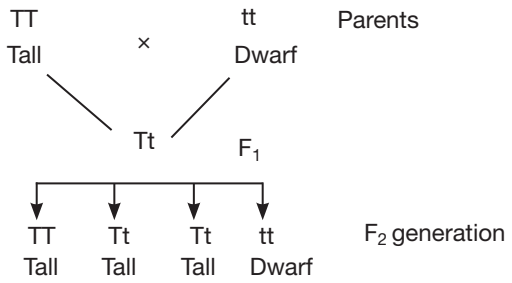
HINTS AND EXPLANATION

7.

Monohybrid cross	Dihybrid cross
The crossing of plants with one pair of contrasting characters. For example: TT tt	The crossing of plants with two pairs of contrasting characters. For example: TTRR ttrr

8. F_1 stands for first-filial generation. It is the generation of hybrids produced from a cross between genetically different parents. For example, cross-fertilization of homozygous tall plants (TT) with homozygous dwarf plant (tt) gives F1 generation heterozygous tall plants with Tt genotype.

F_2 generation or second-filial generation is the generation of individuals that arises as a result of F1 hybrid self-fertilization.



9. A true breeding plant is the one that when self-fertilized, produces offspring with the same traits. They will be either homozygous dominant or homozygous recessive.

10. Multiple allele is a type of non-Mendelian inheritance pattern that involves more than two alternative forms (alleles) of a gene in a population; for example, gene controlling blood group has three allelic forms of the gene I^A, I^B, I^0.

11. Linkage states that if genes are located very close to one another on the same chromosome, then during gamete formation they segregate together as one unit.

12.

Seed colour	Yellow	Y	Green	y
Seed shape	Round	R	Wrinkled	r

13. Contrasting characters chosen by Mendel

(a) Flower colour	Purple	P	White	p
(b) Flower position	Axial	A	Terminal	a

14. The cross between the individual of F1 generation with the recessive parent is known as test cross.

Taking seed colour of pea plant as the trait (yellow as dominant and green as recessive), design a test cross to show the nature of P1 generation plants.

15. According to Mendelian inheritance, F1 generation individual resembles the dominant parent phenotypically and contains a recessive gene that is not expressed out.

However, in some cases, the F1 generation individual resembles both the parents. That means, in heterozygous condition, the recessive gene also expresses itself along with the dominant gene. Such a phenomenon is called codominance.

16. (a) The gametes formed by one parent are RY and RY and those formed by the other parent are ry and ry in P1 generation.

(b) The gametes formed by all F1 generation progenies are Ry type. During the formation of gametes, the characteristics get segregated out.

(c) Mention the two types of gametic combinations possible.

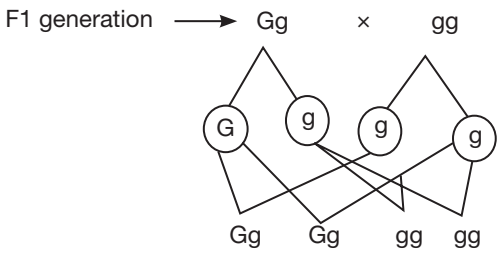
Parental combination–For example, Ry × Ry

Recombination–For example, Ry × RY; rY × RY

17. Heterozygous condition represents the presence of one gene for axial position and one gene for terminal position. Since axial position of flower is a dominant trait, the gene for terminal position of flower is not expressed out. Therefore, heterozygous recessive condition does not exist.

HINTS AND EXPLANATION

18. The cross between the individual of F1 generation with the recessive parent is known as test cross.



Phenotypic ratio: 1:1

Genotypic ratio: 1:1

F1 Gametes		g (recessive)	g (recessive)
	G	Gg (Tall)	Gg (Tall)

19. Autosomal genetic disorders are those diseases arising due to the mutations occurring in the structure or number of genes or non-sex chromosomes. Allosomal genetic disorders arise due to the mutations occurring in the structure or number of genes in sex chromosomes.

20. X-linked traits show criss-cross inheritance in which the gene corresponding to a trait or disease is passed on from father to daughter. This is because daughters get one X chromosome from father and one X chromosome from mother. Therefore, the genes corresponding to X-linked traits are passed on to daughters, and hence, they act as carriers if they are recessive traits. Sons are normal as they do not get any genes on the X chromosome of father as sons get only Y chromosome from father. Since sons get X chromosome from mother, in F2 generation, if the sons get X chromosome with the recessive gene for the trait or disease, they will be affected by the disease.

21. In human males, two dissimilar sex chromosomes are present, that is, one X chromosome and one Y chromosome. In human females, two X chromosomes are present.

- The males produce two types of gametes; one with X chromosome and the other one with Y chromosome. The females produce only one type of gametes both containing X chromosomes.
- If the female gamete fuses with the male gamete with X chromosome, the zygote formed has both X chromosomes. If the female gamete fuses with the male gamete with Y chromosome, the zygote has one X chromosome and one Y chromosome. The former develops into a female child whereas the latter develops into a male child.
- Since the child inherits different chromosomes from father and same chromosomes from mother, the chromosome inherited from father only determines the sex of the child.

22. Y chromosome is smaller in size and contains less number of genes when compared to X chromosome. Therefore, Y-linked traits are very less common than X-linked traits.

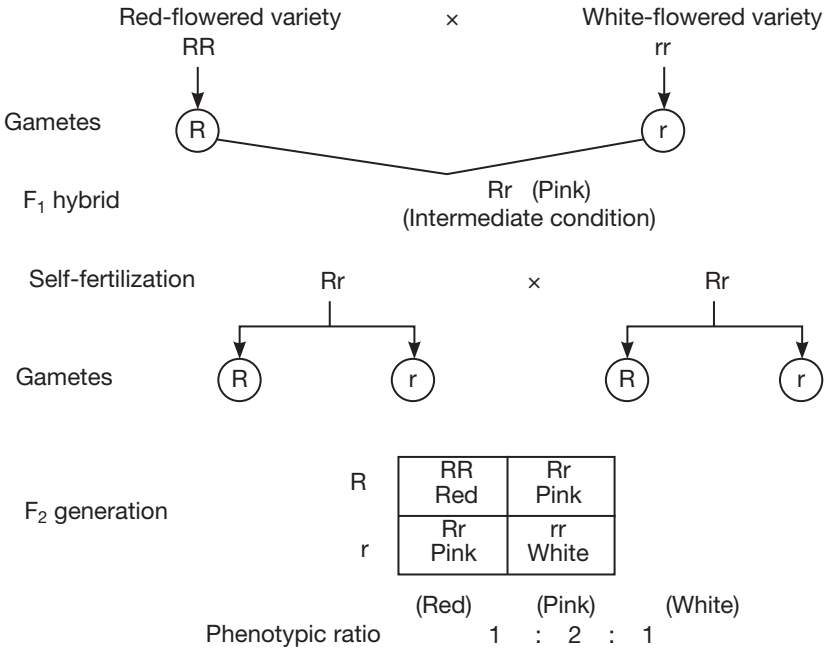
Application and Analysis

23. Incomplete dominance is the phenomenon where when two alleles of a gene are brought together, none of them is dominant over each other, instead a new phenotype is formed. New phenotype is intermediate between the independent expressions of the two alleles.

In monohybrid cross of snapdragon flower plant (dog flower) (*Antirrhinum majus*), F_1 generation plants showed a blend of parental characters.

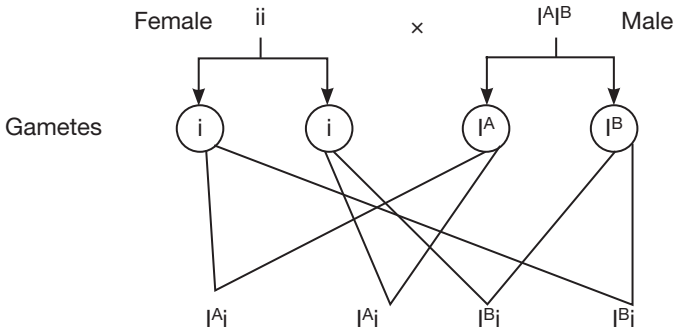
The cross can be summarized as follows.

HINTS AND EXPLANATION



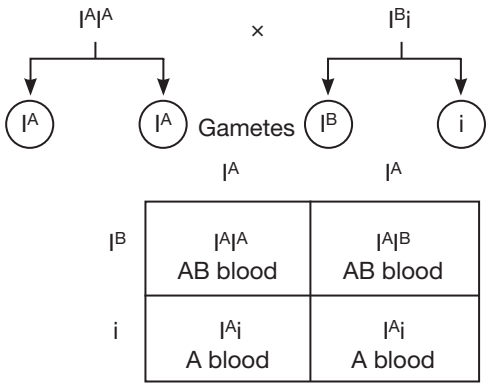
Genotypic ratio 1:2:1

24. Genotype of mother—ii
Genotype of father—I^AI^B



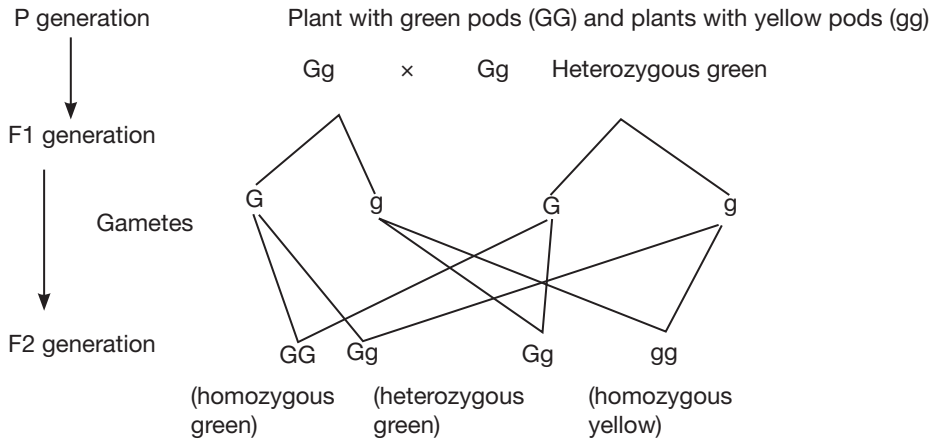
The possible blood groups of the progeny are blood group A and B.

25. Genotype of father can be either I^Ai or I^AI^A
Genotype of mother—I^Bi
Cross can be represented as below:



HINTS AND EXPLANATION

26.



Representation of Mendel's monohybrid cross by Checker Board (Punnett Square)

	G (dominant)	g (recessive)
G	GG (Green)	Gg (Green)
g	Gg (Green)	gg (Yellow)

Phenotypic ratio—3 : 1 (3 plants with green pods; 1 plant with yellow pods)

Genotypic ratio—1 : 2 : 1 (1 homozygous plant with green pods; 2 heterozygous plants with green pods;1 homozygous plant with yellow pods)

27. **Case I:** The plants of P generation in first case would have been homozygous dominant red plants. It would be a cross between two homozygous red plants. Since there is no gene for the recessive trait, all the plants of successive generations would be homozygous dominant red plants.

Case II: The plants of P generation would have been homozygous dominant red-flowered plant and homozygous recessive white-flowered plant. The cross between these two results in heterozygous red-flowered plants in F1 generation. Since they carry the gene for the recessive trait (white flower), one of the next F2 generation progenies would have white flowers.

28. (a) Around 19 per cent of plants are dwarf plants with red flowers.

3 plants out of 16 plants = 18.77 per cent

Around 6 per cent of plants are dwarf with white flowers.

1 plant out of 16 plants = 6.25 per cent

(b) Tall plants with purple flowers are produced in maximum numbers. Around 56 per cent plants are of this type.

(c) 1 plant is homozygous tall with red flowers.

2 plants are homozygous with respect to colour of flower.

(d)

Male gametes	Female gametes				
	X	TP	tP	Tp	tp
	TP	TT PP	Tt PP	TTPp	TtPp
	tP	Tt PP	ttPP	TtPp	ttPp
	Tp	TT Pp	TtPp	TTpp	Ttpp
	tp	Tt Pp	ttPp	Ttpp	ttpp

29. (a) 25 per cent of plants are with wrinkled seeds in F2 generation.

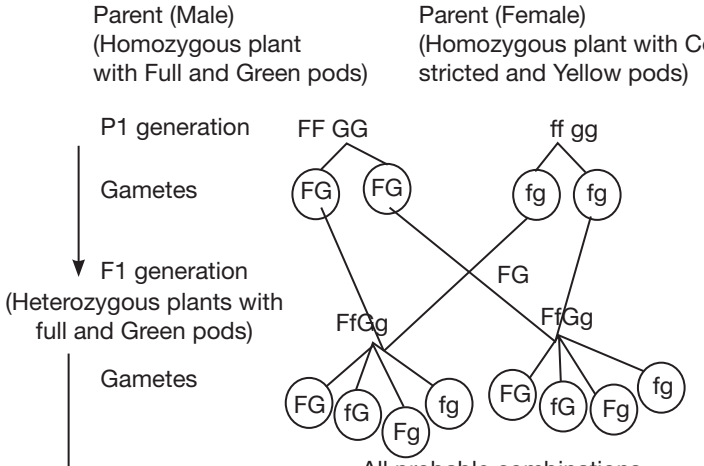
(b) 50 per cent of plants are hybrid plants (heterozygous) as they contain the genes for both round seed and wrinkled seed.

(c) Punnett square for the above experiment

X	R (dominant)	r (recessive)
R	RR (Round)	Rr (Round)
r	Rr (Round)	rr (Wrinkled seeds)

30. (a) The plants obtained on crossing the above plants can have different combination as gametic fusion can take place in any manner (both parental combination and recombination) are possible, which proved that both the traits of the two characters are assorted independently, and hence, plants with different kinds of pods are produced.

(b)



FG	×	FG	→	FFGG	Fg	×	FG	→	FfGg
FG	×	fG	→	FfGG	Fg	×	fG	→	FfGg
FG	×	Fg	→	FFGg	Fg	×	Fg	→	FFgg
FG	×	fg	→	FfGg	Fg	×	fg	→	Ffgg
fG	×	FG	→	FfGG	fg	×	FG	→	FfGg
fG	×	fG	→	ffGG	fg	×	fG	→	ffGg
fG	×	Fg	→	FfGg	fg	×	Fg	→	Ffgg
fG	×	fg	→	ffGg	fg	×	fg	→	ffgg

31. Homozygous and heterozygous dominant—556 × 9/16 = 313

Dominant with respect to only first trait—556 × 3/16 = 104

Dominant with respect to only second trait—556 × 3/16 = 104

Recessive with respect to both traits—556 × 1/16 = 35

32. Number of red-flowered plants in F2 generation—929 × 3/4 = 697

All red-flowered plants are not genotypically same. They may be homozygous red or heterozygous red. A total of 232 plants are homozygous red with both dominant genes; 465 plants are heterozygous red.

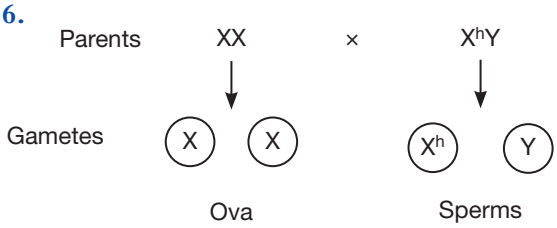
33. In Mendel's monohybrid cross, one plant of P1 generation is homozygous with both the genes for the red colour, which is a dominant trait. The other parent is homozygous recessive with both the genes for yellow colour.

- The gametes produced by dominant (RR) parent contain both R genes. The gametes produced by recessive (rr) parent contain both r genes.
- During recombination, one gene from each parent gets paired up, and hence, the plants produced have one R gene and one r gene. That means, the plants are heterozygous (Rr) with one dominant gene for red colour and one recessive gene for yellow colour. Since the dominant trait is expressed out, the plants bear red-coloured flowers.
- The plants of F1 generation are phenotypically similar to the homozygous dominant plants of P1 generation (red-coloured flowers). Bur they are genotypically dissimilar as they are heterozygous with a gene for the recessive trait.

34. Skin complexion is a character that is affected by more than one pair of genes. Such traits are called polygenic traits. These polygenic traits are controlled by more than one pair of genes and their additive effect results individuals with a spectrum of phenotypes in contrast to monogenic traits that show clear and distinct phenotypes with contrasting traits. Due to this reason, human beings are found to possess variable skin complexion.

35. Sickle cell anemia is an autosomal recessive disorder that arises due to a mutant allele. But this gene affects various other organs also. This comes under the condition of pleiotropy where a single gene has effect on various characters.

36.



	X^h	Y
X	XX^h	XY
X	XX^h	XY

(a) Since the presence of one X chromosome with the mutant allele is sufficient to express the disease, it should be X-linked dominant disorder.

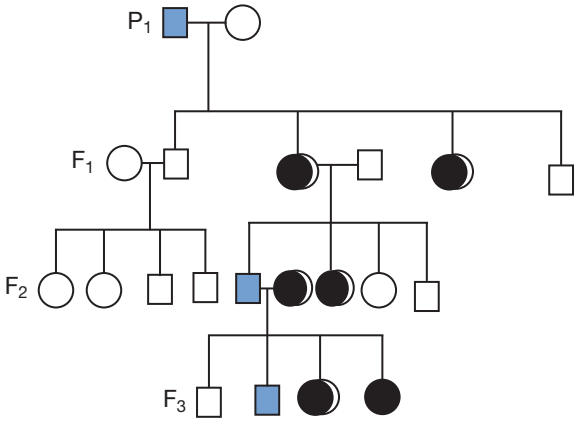
(b) Fifty per cent of sons are affected by the disease, whereas 50 per cent of sons are normal in F1 generation. 'A' is normal son and 'B' is affected son. The son who inherits X chromosome mutant gene from mother is affected by the disease. The son who inherits normal X chromosome from mother is not affected by the disease.

(c) Since A and C are son and daughter without any mutant genes, the successive generation are totally free of the diseased genes.

(d) As far as F_2 generation is concerned, E and F are normal sons as the mother is normal and sons do not inherit X chromosomes from father. G and H are affected daughters as they inherit the only X chromosome with mutant gene from their father.

(e) Females are more affected by the disease than males. If father is affected, 100 per cent daughters are affected by the diseases as they inherit the single X chromosome from father. If mother is affected, 50 per cent daughters are affected by the disease as they inherit at least one X chromosome from mother. Fifty percent of daughters are not affected by the disease. Therefore,

females are predominantly affected by the disease in case of X-linked dominant disorders.

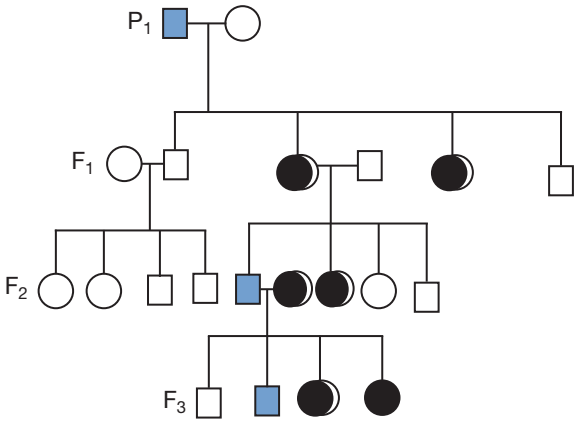


37. X-linked traits show criss-cross inheritance in which the gene corresponding to a trait or disease is passed on from father to daughter. This is because daughters get one X chromosome from father and one X chromosome from mother. Therefore, the genes corresponding to X-linked traits are passed on to daughters, and hence, they act as carriers if they are recessive traits. Sons are normal as they do not get any genes on the X chromosome of father as sons get only Y chromosome from father. Since sons get X chromosome from mother, in F2 generation, if the sons get X chromosome with the recessive gene for the trait or disease, they will be affected by the disease. Y-linked traits are inherited to sons from fathers. They do not show criss-cross inheritance.

38. (a) A man suffering from muscular dystrophy should have a mutant allele on the X chromosome. Since daughters necessarily get one X chromosome from father, they inherit the gene for the disease. Since it is a recessive gene, the daughters act as carriers. Since mother has no gene for the disease, they are not affected by the disease.

(b) The sons receive normal X chromosome from mother, and hence do not possess any mutant gene for the disease. If they marry normal women without any family history for muscular dystrophy, the disease is not at all passed on to the successive generations. If only sons are born in the first generation, the successive generations will be free of the disease. If daughters are born, they act as carriers and can pass on the genes to their sons as well as daughters. Since one X chromosome only possesses the mutant allele, the daughters who get normal X chromosome will be normal and they do not carry the gene. The sons who get normal X chromosome will be normal. In these two cases, the successive generations will be free of the disease.

(c) Pedigree chart for the disease:



39. Both Down's syndrome and Turner's syndrome are chromosomal disorders. Down's syndrome results due to the presence of extra chromosome. The extra copy of chromosome is autosome and not sex chromosome. Turner's syndrome results due to the absence of an X chromosome in females. In Down's syndrome, the affected individuals are characterized by abnormal physical features and impaired psychomotor functions coupled with mental retardation. In Turner's

syndrome, the females are characterized by sterility with rudimentary ovaries and also underdeveloped secondary sexual characters.

40. The blood groups A, B, AB, and O refer to the phenotypic expressions of the various allelic combinations corresponding to the different genotypic combinations. The type of blood group is controlled by the gene I that has three allelic forms, namely I^A; I^B; i. Alleles I^A and I^B produce different types of sugars (glycoproteins) and allele 'i' does not produce any sugar. I^A and I^B are completely dominant and 'i' is totally recessive and does not express itself in the presence of either I^A or I^B.

When both I^A and I^B are present together, they both express their own type of sugars. Since three different alleles are there, six types of genotypes and four types of phenotypes are possible. Blood group A (mother) corresponds to the presence of a dominant gene I^A that produces antigen A. Blood group B (father) corresponds to the presence of a dominant gene I^B that produces antigen B. Along with these genes when allele 'i' is present, it is not expressed out as it is a recessive allele.

The probable genetic combinations:

Mother	Father	Child
I^A; I^A	I^B; I^B	AB
I^A; i	I^B; I^B	AB; B
I^A; I^A	I^B; i	AB; A
I^A; i	I^B; i	AB; A; B; O

41. Sikle cell anemia is an autosomal recessive disorder whereas haemophilia is an X-linked disorder. Autosomal genetic disorders are not sex specific as they do not involve sex chromosomes. Haemophilia is an X-linked recessive disorder. For X-linked recessive traits, females generally act as carriers of the affected gene whereas males are affected by the disease. Females can be affected by the disease only when both the X chromosomes contain mutant genes for the disease. But this condition usually is not viable and the babies are susceptible to death in fetal stage itself. Hence, haemophilia is very rare in females.

42. Male and female gametes are formed by means of meiotic cell division. This involves the phenomena of linkage and crossing over of homologous chromosomes. A group of genes that is normally inherited together is termed as linkage group and the genes are called linked genes. This phenomenon in which certain genes present on a chromosome are together carried on to daughter cell is called linkage. Linkage is responsible for the inheritance of characters to the successive generations. If all the genes on a chromosome are transferred, it is called complete linkage. If some of the genes are transferred, it is called incomplete linkage. Generally, incomplete linkage takes place during meiotic cell division. As some genes are passed on to the daughter cell (gamete) together, the corresponding traits are passed on from the respective parent. The phenomenon of the exchange of segments of non-sister chromatids (genes) of homologous chromosomes is known as crossing over. Crossing over is followed by recombination. Due to the exchange of some genes between the homologous chromosomes, the pairing of chromosomes may give rise to a new combination of genes, which is called recombination.

This results in variations (new traits) that are not there in the parents. These are inherited to the next generation because the fusion of these gametes results in the formation of diploid zygote. Therefore, the variations occurred in the chromosomes are transmitted to the children. Therefore, a child possesses some characteristic resemblances with both father and mother. However, the child is not an exact copy of either of the parents.

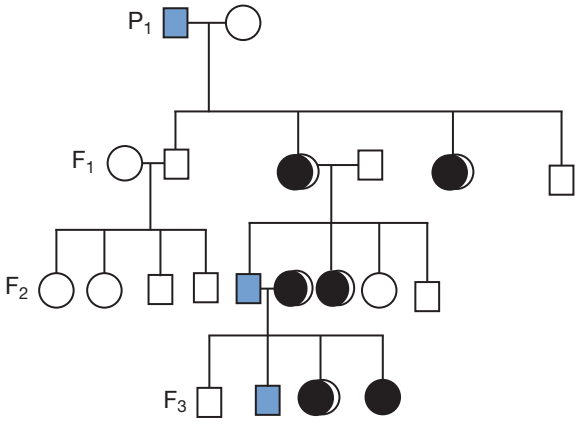
43. The genes controlling the secretion of milk are found in both males and females. But,

they are in recessive condition in males. Hence, the character is not expressed in males. It is expressed in females as the gene is in dominant condition. Such traits are called sex-limited traits.

44. In *Drosophila*, the gene controlling the colour of the eye is present on the X chromosome. The gene for red colour of eye is a dominant allele whereas the gene for the white colour of eye is a recessive allele. That means, white eye colour in *Drosophila* is an X-linked recessive trait. Since it is a recessive trait, it is phenotypically expressed out only in homozygous condition. In females, since two X chromosomes are present, both the chromosomes possessing the diseased genes is a very rare condition and may not be viable. In heterozygous condition, they express only red colour of eye and act as carriers for the gene for white eye. In males, since only one X chromosome is present, the disease is expressed out and the person suffers from the disease.

45. Consanguineous marriages are the marriages between close relatives; such marriages are not suggestible due to the following reasons:

- Any gene carrying a particular abnormality or disorder present in the homologous chromosomes is passed onto the next generation. All the offspring necessarily need not be affected by the disease. Sometimes, if the gene is recessive allele, it may be present in the offspring but the child is normal. However, such persons can act as carriers for the disease.
- If the carrier male or female marries a normal female or male, the probability of progenies suffering from the disease is very less. At the most, some of the progenies may become carriers.
- In case of mating between close relatives, there is a chance that both male and female may act as carriers due to their common ancestry. In such a case, the probability of the offspring suffering from the disease is much higher than in the previous case.
- A part from this, the closely related individuals have some part of DNA common, and hence, the variation is less. Variations lead to genetic diversity and heritance of good traits in the family. This scope also is limited in case of marriages between close relatives.



Assertions and Reasons

46. Sexual reproduction brings about variations in the offspring because it involves the fusion of gametes produced by meiotic cell division. This is because meiotic cell division involves exchange of genetic material due to crossing over of homologous chromosomes. Crossing over does not take place during mitotic cell division. That is why mitotic cell division does not result in variations in the offspring.

On the other hand, DNA replication is a part of both mitotic cell division and meiotic cell division. As the new strand formed is always complementary to the original strand, DNA replication serves to preserve the DNA

sequence across the generations. Therefore, assertion is correct and reason is wrong.
Hence, the correct option is (c)

47. The DNA of the child will not be same as that of either of the parents. The DNA replication results in the formation of DNA with the same nucleotide sequence as that of the parent cell because the two strands are complementary to each other. Thus, the nucleotide sequence is preserved in the daughter cell. However, gamete formation involves meiotic cell division, which involves the important phenomena of linkage and crossing over. Linkage passes on the same genes (DNA sequence) whereas crossing over results in the exchange of genetic material between the homologous chromosomes. As a result, the gametes formed will have some part of DNA same as the parent while some part of DNA different from the parent. During gametic fusion, the zygote formed will have a DNA sequence that is a combination of mother and father. Therefore, the DNA of the child resembles that of the parents, but not exactly same as the parent.
Hence, the correct option is (c)

48. According to Mendelian inheritance, every character is represented by two alternative traits.

Each character is controlled by a pair of genes called alleles that are located on the homologous chromosomes. One is a dominant allele and the other is a recessive allele. The trait controlled by the recessive allele is always masked by the dominant allele, and hence not expressed out in the presence of its dominant gene. Heterozygous condition represents one dominant and one recessive trait of the same character. Therefore, heterozygous recessive progenies are not possible in Mendelian inheritance. During the formation of gametes, the alleles get segregated independently, which explains the purity of gametes. But this is not the reason for the assertion given.
Hence, the correct option is (b).

49. Red-flowered plants of P generation are pure breeds (homozygous dominant and homozygous recessive white flowered plant). The cross between these two plants produces all red-flowered plants in F1 generation. But, these plants are heterozygous as they possess a recessive gene for white flower. Since all the plants of F1 generation are heterozygous red-flowered plants, the genotypic ratio and the phenotypic ratio are same.
Hence, the correct option is (d).

50. When the dominant gene shows incomplete dominance, the flowers of the plants of F1 generation show an intermediate expression between the dominant trait and the recessive trait.
Hence, the correct option is (c).

51. The gene corresponding to blood group has three alleles instead of usual three alleles. They are represented as I^A; I^B; i. Both I^A and I^B are codominant and hence both are expressed in heterozygous condition. The gene 'I' is recessive, and hence is not expressed in the presence of either of the dominant genes. It is expressed only in homozygous condition. When both the parents have same blood group, then the children necessarily possess the same blood group. But when the parents have different blood groups, the children need not possess the same blood group. For example, if one parent has blood group 'A' and another parent has blood group 'B,' the child will get 'AB' blood group.
Hence, the correct option is (d).

52. In case of a dihybrid cross, two different traits are considered. The two alternative forms of each trait are assorted independently during the formation of gametes. As a result, the trait for tallness of plant can be combined with that for wrinkled shape of the seeds and the trait for dwarfness of the plant can be combined with that for round shape of the seed. This is because of the independent assortment

of the traits. Each progeny gets a homologous chromosome from male and female. But only when these chromosomes possess contrasting alleles, the progenies may inherit the recessive genes. Parental generation plants should be hybrid varieties.
Hence, the correct option is (b).

53. In grasshopper, male individual determines the sex of the offspring as female has two X chromosomes and males have only one X chromosome. This is called XX–XO type of sex determination. But all organisms do not show male heterogamity. Some organisms such as birds show female heterogamity as females have one Z chromosome and one W chromosome whereas males have two Z chromosomes.
Hence, the correct option is (c)

54. Since X chromosome is present in both the sexes, genes on the X chromosome are called sex-linked genes.
Hence, the correct option is (a)

55. Males possess only one X chromosome. So, if any mutant allele is present on X chromosome, it is phenotypically expressed out. It is no more a recessive gene. Most of the genetic disorders are X-linked diseases. Even in Y-linked traits also, since males only possess Y chromosome, the traits are obviously expressed out. Therefore, males cannot act as carriers for sex-linked traits due to the heterozygous nature of sex chromosomes. Males cannot transmit the X-linked genes to their sons as sons get Y chromosome from father and X chromosome from mother. But this is not the reason for the given assertion.
Hence, the correct option is (b)

56. Hypertrichosis is a disease found only in males because it is a Y-linked trait. Since Y chromosome is transmitted only to sons from fathers, Y-linked traits are passed on to sons. Since females do not possess Y chromosome at all, all Y-linked traits are found only in males.
Hence, the correct option is (a)

57. Sickle cell anemia is a genetic disorder due to the presence of a recessive mutant allele on one of the chromosomes other than sex chromosomes. It is an autosomal recessive disorder. As it does not involve sex chromosomes, it is not sex specific. It can be found in either males or females in equal frequency.
Hence, the correct option is (a)

58. Sex of a child is determined by father and not mother because father is heterogametic with one X chromosome and one Y chromosome. Mother has only two X chromosomes. Mother contributes only X chromosome to either son or daughter. If sperm with X chromosome of father fuses with the egg of the mother, the fusion results in the birth of daughter. If sperm with Y chromosome fuses with the egg of mother, the fusion results in the birth of son.
Hence, the correct option is (a)

ASSESSMENT TESTS

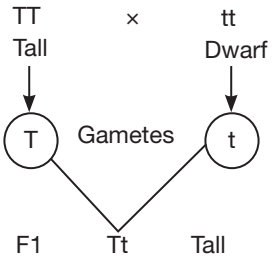
1. (c)
2. (b)
3. (a)
4. (a)
5. The number of chromosomes in a particular species is constant.
6. The proportion of plants heterozygous with respect to both the traits are in F2 generation of Mendel's dihybrid cross is 4/16.
7. The probability of birth of male child in humans is 50 per cent.
8. Human Y chromosome is shorter than X chromosome.
9. Allosomes or sex chromosomes are responsible for the transmission of characters from parents to the offspring.
10. 9:3:3:1
11. Dwarf
12. Rr
13. Round seed
14. 1:2:1
15. XY
16. 9
17. TtPp or TTPp or TtPP
18. Nucleus is present in cytoplasm whereas others are present in nucleus.
19. Ry represents heterozygous condition whereas others represent homozygous condition.
20. Yy represents a pair of alleles for contrasting traits whereas others represent a single allele for a particular trait.
21. Rryy represents a phenotypically different combination whereas others are all phenotypically similar.
22. Haemophilia is sex-linked genetic disorder whereas others are autosomal genetic disorders.
23. Crow show female heterogamety or ZZ–ZW type of sex determination whereas others show male heterogamity or XX–XY type of sex determination.
24. A second cross of the same genotypes in which the sex of the parental generation is reversed is called reciprocal cross.

	Female	Male	Offspring
Original cross	Tall (TT)	Dwarf (tt)	Tt
Reciprocal cross	Dwarf (tt)	Tall (TT)	Tt

25. Law of segregation or law of purity of gametes states that during gamete formation, the two alleles in an individual segregate or separate from each other so that each gamete gets only one kind of allele. It is also known as "law of purity of gametes" because each gamete is pure itself, that is, having either 'T' (gene for tallness) or 't' (gene for dwarfness)

    A cross between an individual of unknown genotype and homozygous recessive parent is called test cross. It is used to test whether an individual is homozygous or heterozygous. Test cross is of two types–monohybrid and dihybrid. A monohybrid test cross deals with single trait and give F1 phenotypic ratio 1:1. A dihybrid test cross deals with two traits at a time and gives phenotypic ratio of 1:1:1:1.
26. Law of dominance states that "When a pair of contrasting alleles or traits are brought together in F1 hybrid, then only one of them will express itself, and other will be suppressed. The trait which is expressed is called dominant and the one which is suppressed is called recessive trait." For example, in a cross between homozygous tall (TT) plant and

homozygous dwarf plant (tt), F1 hybrid plants were all tall.

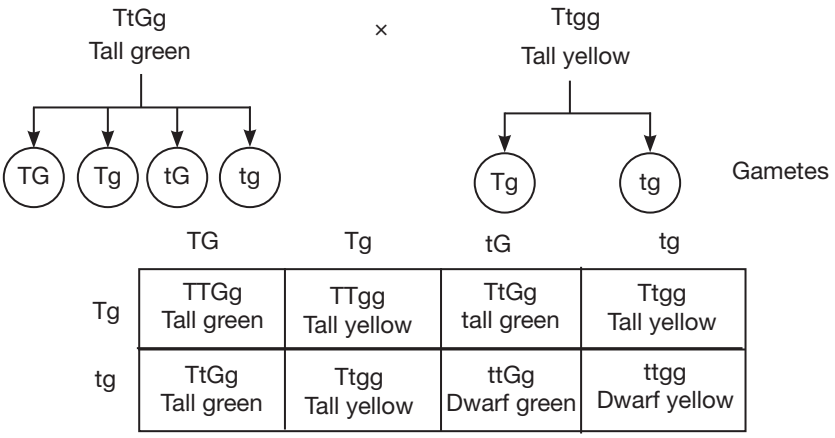


27. The cross of an F_1 hybrid with one of the two parents is called back cross. In plant breeding, back cross is performed a few times in order to increase the traits of that parent. There are two possibilities for back cross. In one possibility, there is a cross between F_1 hybrid (Tt) and dominant parent (TT). In such a cross, plants will be 100 per cent tall. In second possibility, there is a cross between F1 hybrid (Tt) and recessive parent (tt). In such cross, plants will be 50 per cent tall and 50 per cent dwarf.

28. One gene influences two or more seemingly unrelated phenotypic traits. This phenomenon is known as pleiotropism and the gene responsible for pleiotropism is known as pleiotropic gene.

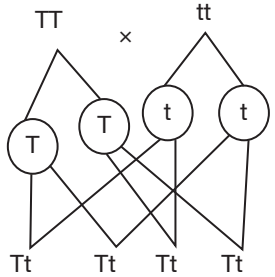
In pea plant, starch synthesis, size of starch grains, and seed shape are controlled by one gene. The gene can be represented as 'B', which has two allelic forms 'B' and 'b'. The homozygous pea plants with BB genotype are efficient in starch synthesis, and hence, the starch grains are large and seeds at maturity are round. The heterozygous Bb plants produce round seeds indicating B is the dominant allele, but the starch grains are intermediate in size, that is, for the starch grain size, alleles show incomplete dominance. Homozygous pea plants with 'bb' phenotype are less efficient in starch synthesis, and they have small starch grains and the seeds are wrinkled.

29.



	TG	Tg	tG	tg
Tg	TTGg Tall green	TTgg Tall yellow	TtGg tall green	Ttgg Tall yellow
tg	TtGg Tall green	Ttgg Tall yellow	ttGg Dwarf green	ttgg Dwarf yellow

30. (a) Homozygous tall and homozygous dwarf

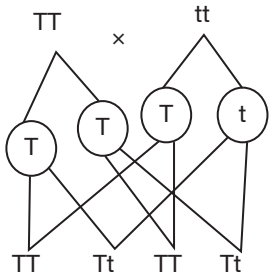


- A homozygous tall plant produces gametes with genes for tall trait. A homozygous dwarf plant produces gametes with genes for dwarf trait.
- The cross between the above plants results in the fusion of gametes produced by them in different combinations, which gives rise to all heterozygous plants with genes for both the

HINTS AND EXPLANATION

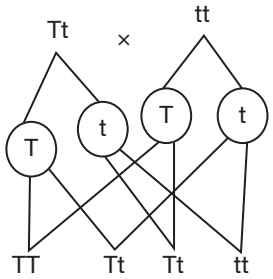
contrasting traits (Tt) in the F1 generation. However, all the progenies are phenotypically tall as the recessive dwarf trait is masked.

(b) Homozygous tall (dominant) and heterozygous tall

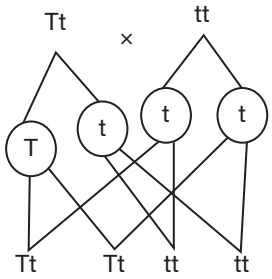


- A homozygous tall plant produces gametes with genes for tall trait. A heterozygous tall plant produces one gamete with gene for tall trait and another gamete with gene for dwarf trait.
- The cross between the above plants results in the fusion of gametes produced by them in different combinations, which gives rise to two homozygous tall plants with both the genes for tall trait and two heterozygous plants with genes for both the contrasting traits (Tt) in the F1 generation. However, all the progenies are phenotypically tall as the recessive dwarf trait is masked in the heterozygous.

(c) Heterozygous tall and heterozygous tall



- A heterozygous tall plant produces one gamete with gene for tall trait and another gamete with gene for dwarf trait.
- The cross between the above plants results in the fusion of gametes produced by them in different combinations, which gives rise to one homozygous tall plant with both the genes for tall trait, two heterozygous plants with genes for both the contrasting traits (Tt), and one homozygous dwarf plant with both genes for dwarf traits in the F1 generation. However, three progenies are phenotypically tall as the recessive dwarf trait is masked in the heterozygous plant. Only homozygous dwarf plant is phenotypically dwarf.



- A heterozygous tall plant produces one gamete with gene for tall trait and another gamete with gene for dwarf trait. A homozygous dwarf plant produces gametes with genes for dwarf trait.
- The cross between the above plants results in the fusion of gametes produced by them in different combinations, which gives rise to two heterozygous plants with genes for both the contrasting traits (Tt) and two dwarf plants with both the genes for dwarf traits in the F1 generation. However, the two heterozygous progenies are phenotypically tall as the recessive dwarf trait is masked.

31. • Allosomes are sex chromosomes that are responsible for the determination of sex. These are responsible for the transmission of hereditary characters in organisms.

- Autosomes are the chromosomes other than sex chromosomes.

32. Sudden variations may take place in a particular individual due to the change in the structure of a gene (DNA sequence) or chromosome or in the number of chromosomes. Such sudden changes are called mutations. These mutations may bring about a change in the genetic message of a cell thereby resulting in the consequent change in the phenotype of the respective organisms.

- Mutation is a random phenomenon that takes place spontaneously at a low frequency. Some external or environmental factors may induce mutations artificially in certain organisms. Such agents that cause mutations are called mutagens. Radiations, chemicals, and temperature are the important mutagens.

33. (a) Heterozygous

(b) Gene

(c) Recessive allele

(d) Homologous chromosomes

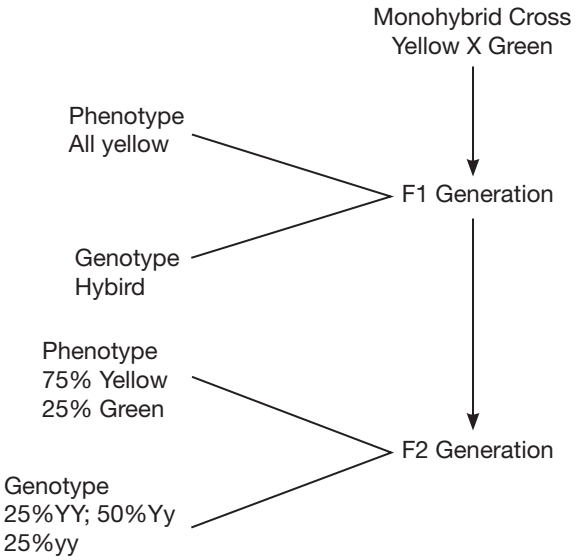
(e) Pleiotropy

(f) Autosomal traits

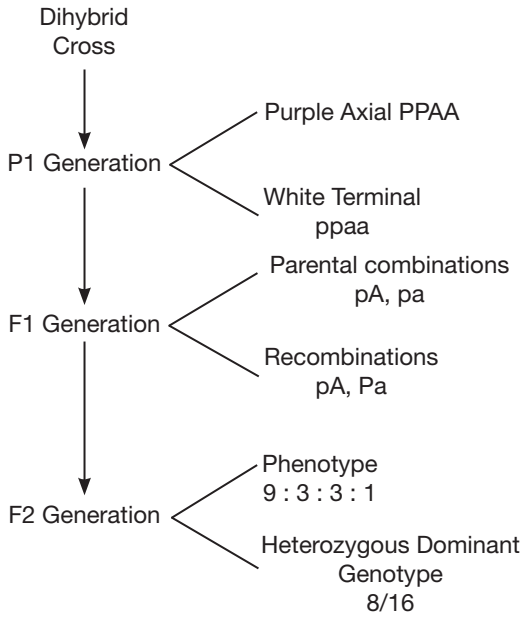
(g) Codominance

(h) Clones

34. Fill in the blanks in the following flow chart with appropriate terms.



35. Fill in the blanks in the following flow chart with the appropriate terms: (yellow: dominant; green: recessive)



HINTS AND EXPLANATION