Chapter 16

Heredity and Variation

Chapter Outline

16.1 Inheritance

16.2 Heredity: Characteristics and their appearance

16.3 Mendel's laws of inheritance

16.4 Diseases due to chromosomal aberrations

Important Points

1. Inheritance

(1) **Inheritance:** The tendency to transmit the variations from one generation to the next is called inheritance.

(2) **Offspring** produced through sexual reproduction show more variations. Those produced through asexual reproduction have comparatively much lesser variations.

(3) **Genetics:** Genetics is the branch of biology which studies the transfer of characteristics of organisms in the form of genes from one generation to the next.

Observe. (Textbook page 179)

(1) Carefully observe your classmate's ear lobes.

Ans. Some of the students have ear lobes completely attached, some have partially attached ear lobes while some have totally free ear lobes.

(2) Irrespective of all of us being humans, what difference do you notice in our skin colour?

Ans. Every person has different skin colour. Some are fair, some are dark while some are intermediate. No two persons are exactly alike with respect to skin colour.

(3) All of you are in Std. IX. Why then are some students tall and some short?

Ans. Though all of us are in the same standard, each one has different type of inherited characters from their respective parents. Therefore, there are variations in the skin colour.

(4) Heredity

Heredity: Transfer of characteristics from parents to offspring is called heredity. Therefore, every living organism has similar characteristics as its parents.

2. Inherited Traits and Expression of Traits

Can you tell? (Textbook page 180)

Q. How do specific traits or characteristics appear in ofganisms?

Ans. The specific characteristics seen in any living organisms are due to the genes transmitted from its parents to the offspring.

The characteristics and variations seen in the organisms are due to inherited characteristics.

(1) The inherited characters are based on chromosomes present in the cell, the DNA molecule present in the chromosome, and the sequence with which the nucleotides are arranged in the DNA molecule. Based on the sequence of the nucleotides present in the DNA molecule, a particular protein is synthesized. This segment of DNA is called a gene.

(2) The growth in the height of a plant depends upon growth hormone. Due to quantity of the growth hormone, the height of the plant is determined. The quantity of growth hormones produced by a plant depends upon the efficiency of the concerned enzyme. Efficient enzymes produce a greater quantity of the hormone resulting in taller plant.

Page 2 — Chromosomes, DNA, genes and DNA fingerprinting

Key Takeaway

The height of an individual depends on the sequence of nucleotides present in DNA.



3 Chromosomes

Chromosomes are thread-like structures made up of nucleic acids and proteins. They carry hereditary characters in the form of DNA. The number of chromosomes is characteristic of each species.

Structure of Chromosomes

i. The proteins on chromosomes are **histones**. DNA is present along with histones in chromosomes.

ii. Chromosomes are visible only during cell division. They appear dumbbell-shaped.

iii. Each chromosome has a **'primary constriction'** or **centromere**. The centromere divides the chromosome into two arms. The short arm is called **'p' arm** and the long arm is called **'q' arm**.

iv. According to the position of the centromere, chromosomes are of four types: **metacentric, sub-metacentric, acrocentric** and **telocentric.**

v. According to their function, chromosomes are of two types: **somatic** (body chromosomes) and **sex chromosomes** (those that determine sex). Somatic chromosomes can be homologous or heterologous.

Structure of DNA

i. Watson and Crick proposed the model for DNA structure in 1953.

ii. According to Watson and Crick, the DNA molecule is a double helix.

iii. Each strand of this helix is made up of nucleotides.

iv. Each nucleotide is made up of a phosphoric acid, a deoxyribose sugar and a nitrogenous base.

v. Nitrogenous bases are of two types: purines and pyrimidines. **Purines**: adenine and guanine. Pyrimidines: cytosine and thymine.

vi. The two strands of the helix are held together by hydrogen bonds on opposite sides. Adenine pairs with thymine via double hydrogen bonds, while cytosine pairs with guanine via triple hydrogen bonds.

vii. The deoxyribose sugar and the phosphoric acid alternate with each other and are arranged on the outer side of each helix.

4 D.N.A. (Deoxyribonucleic acid)

(1) **Discovery:** Friedrich Miescher discovered DNA molecule in 1869 from the nuclei of white blood cells. It is found in the nucleus and is acidic in nature; hence it is called nucleic acid. DNA is present in all organisms, e.g. viruses, bacteria, plants, animals and humans.

(2) **Functions of DNA:** DNA controls the functioning, growth and division (reproduction) of the cell. It also carries hereditary characters and is responsible for genetic transmission. It is called the **'Master Molecule'**.

5 Genes

- Genes are the functional units of heredity.
- The nucleotide sequence in DNA forms a gene.
- Genes are found on the DNA molecule.
- Genes control the structure and function of the body and help to transmit hereditary characters from parental generation to the offspring.

6 D.N.A. finger printing

- The sequence of genes in the DNA of a person is called a **genome.**
- DNA fingerprinting is a technique used to identify a person's genome.
- It is also used to identify the lineage and to identify criminals because it is unique to every person.

At a Glance: Key Terms and Meanings

Term	Meaning
Chromosome	Thread-like structure of nucleic acids and proteins that carries hereditary characters.
DNA	Nucleic acid that controls cell functions and carries hereditary information; the 'Master Molecule'.
Gene	Functional unit of heredity; a specific nucleotide sequence on DNA.
Genome	Entire sequence of genes of a person.
DNA fingerprinting	Technique used to identify a person's genome; also used to trace lineage and identify criminals.

Question Bank

Q1. What are chromosomes? Mention their composition.

Q2. State the different types of chromosomes according to the position of centromere.

Q3. What are somatic and sex chromosomes? How do they differ?

Q4. Write a short note on the discovery and functions of DNA.

Q5. Who proposed the model for DNA structure? What is its structure?

Q6. Name the nitrogenous bases present in DNA and state their types.

Q7. What is a gene? Where is it located and what is its function?

Q8. What is DNA fingerprinting? Write two of its uses.

1 Human Genome Project (HGP)

- Human Genome Project is a major work in human genetics, conducted by a few nations together.
- As per HGP findings, humans have about 20,000 to 30,000 genes in the body.
- Detailed results of these findings are available on the web site.

2 RNA (Ribonucleic Acid)

1. RNA is another important nucleic acid, made of ribonucleotide molecules.
2. RNA is composed of ribose sugar, phosphate molecules and four nitrogenous bases—adenine, guanine, cytosine and uracil. Uracil is present in RNA instead of thymine (which is found in DNA).
3. A single-stranded polynucleotide forms one RNA molecule.
4. **Types of RNA according to their function :**

rRNA	mRNA	tRNA
Ribosomal RNA	**Messenger RNA**	**Transfer RNA**
Component of the ribosome organelle. Helps in protein synthesis.	Carries the information of protein synthesis from genes on DNA chain in the nucleus to the ribosomes in the cytoplasm.	According to the message of the mRNA, tRNA carries the amino acid up to the ribosomes.

Know something more !

▶ How does protein synthesis take place?

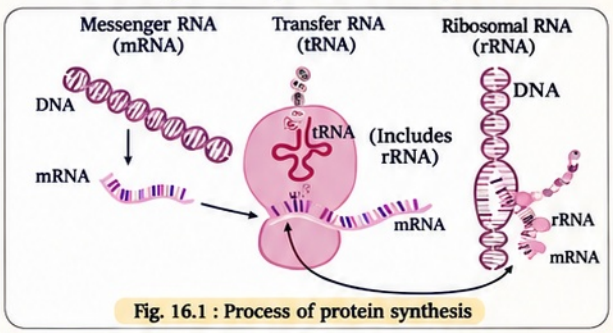


Fig. 16.1 : Process of protein synthesis

Genetic information and mRNA

Genetic information is copied from DNA on mRNA. mRNA comes out of the nucleus and enters the cytoplasm. It links with ribosomes present in the cytoplasm. tRNA molecules are also present there. tRNA transfers the required amino acids after reading the genetic information brought by mRNA. The amino acids are then linked to form a protein.

3 Mendel's Principles of Heredity

- Principles of heredity state that genetic material is transferred from parents to offspring in equal quantity.
- More than a century ago, Gregor Johann Mendel performed experiments on the pea plant and proposed the laws to explain the principles of heredity.
- Mendel's experiments were based on seven visible characteristics of the pea plant, *Pisum sativum*.

Characters of Pea Plant Studied by Mendel

Characters	Dominant	Recessive
Shape of the seed	Round (R)	Wrinkled (r)
Colour of seed	Yellow (Y)	Green (y)
Colour of flower	Purple (C)	White (c)
Shape of the pod	Inflated (I)	Constricted (i)
Colour of a pod	Green (G)	Yellow (g)
Position of a flower	Axillary (A)	Terminal (a)
Height of a plant	Tall (T)	Dwarf (t)

Rules from Mendel's Findings

- When the characteristics are passed to the next generation, they follow certain rules.
- Mendel's laws are based on these findings.
- Mendel's laws are said to be the foundations of modern genetics.

4 Monohybrid Cross

- Cross between two pea plants with only one pair of contrasting characters is called monohybrid cross.
- Parental generation (P_1) consisted of tall and dwarf plants. These plants were crossed with each other.

Question Bank

1. What is the Human Genome Project? Write its main findings.

Ans. A large-scale project in human genetics; it revealed that humans have about 20,000 to 30,000 genes, and detailed results are available on the website.

2. What is RNA? State its composition.

Ans. RNA is an important nucleic acid made of ribonucleotide molecules. It consists of ribose sugar, phosphate and four nitrogenous bases—adenine, guanine, cytosine and uracil (uracil replaces thymine of DNA).

3. List the types of RNA and their functions.

Ans. (i) rRNA – component of ribosome; helps in protein synthesis. (ii) mRNA – carries the information of protein synthesis from genes on DNA in the nucleus to ribosomes in the cytoplasm. (iii) tRNA – carries the amino acid to ribosomes according to the message of mRNA.

4. What is a monohybrid cross?

Ans. A cross between two pea plants with only one pair of contrasting characters is called a monohybrid cross.

Page 4 Mendel monohybrid cross, Punnett squares, genotype and phenotype

Mendel's conclusions from monohybrid cross

- After crossing the plants of the P_1 (parental) generation, F_1 generation is produced. F is the symbol for filial. All the F_1 offspring were tall and none was dwarf.
- Mendel called tallness a dominant character, while dwarfness a recessive character because dwarfness did not appear in F_1.
- Mendel concluded that the factors (determinants) for inheritance of characters occur in pairs. These factors are called genes.
- Dominant characters are written with capital letters, whereas recessive characters are written with small letters.
- The experiment and its results are shown in the Punnett square.

E.g. TT – Pair of genes of one parent
tt – Pair of genes of another parent
TT and tt – Separate at the time of gamete formation.

Gametes of P_1 generation

- Gametes of P_1 generation : TT = T and tt = t.

First filial (F_1) generation

Female tt ↓ / TT Male →	T	T
t	Tt	Tt
t	Tt	Tt

- The offspring of the F_1 generation were all tall.
- Even when one parent was dwarf, none of the offspring were dwarf. This observation led Mendel to the principle of dominance and recessiveness.
- Tallness is the dominant character, whereas dwarfness is the recessive character.
- The character of tallness hides the expression of dwarfness. If the factor for tallness is absent, only dwarfness is expressed.
- Tallness or dwarfness is the outward (external) appearance. This is called a phenotype, whereas the genetic constitution is called a genotype.

Key terms

- **Phenotype :** External appearance.
- **Genotype :** The factors or genes present in the offspring.

The F_2 progeny

Genotype	Phenotype	Characters	Type
TT	Tall	Pure dominant	Homozygous
tt	Dwarf	Pure recessive	Homozygous
Tt	Tall	Hybrid plants	Heterozygous

F_2 filial generation formed by selfing

- F_2 generation is obtained by self-cross between F_1 plants.
- The F_2 generations show the following gametes : Tt = T and t; Tt = T and t.

Male ↓ / Female →	T	t
T	TT	Tt
t	Tt	tt

Genotypic ratio in F_2

- In F_2 the cross shows the following genotypic ratio.

1TT : 2Tt : 1tt

1 Homozygous dominant : 2 heterozygous : 1 homozygous recessive

Phenotypic ratio in F_2

- 3 tall plants : 1 dwarf plant are visible in F_2 progeny.

Phenotypic ratio = 3 : 1

Mendel's figures

- Total plants = 929, Tall plants = 705, Dwarf plants = 224.

Mendel's dihybrid cross

- When two pairs of contrasting characteristics are under consideration, then it is called a dihybrid cross.
- Round and yellow seed-bearing plant was crossed with green and wrinkled seed-bearing plant to have a dihybrid cross.
- Round and yellow are dominant characters. Green and wrinkled are recessive characters.

Parental generation P_1

- First parent : RR (Round seeds) + YY (Yellow seeds) and Second parent : rr (Wrinkled seeds) + yy (Green coloured seeds).

Question Bank

1 What is a phenotype?
A External appearance.

2 What is a genotype?
A The factors or genes present in the offspring.

3 What is the gamete of TT?
A T.

4 What is the gamete of tt?
A t.

5 What is the genotypic ratio in F_2?
A 1TT : 2Tt : 1tt.

6 What is the phenotypic ratio in F_2?
A 3 : 1.

Page 5 Dihybrid Cross, F_1/F_2 Punnett Square and Ratios

- Gametes from RRYY parent = RY
- Gametes from rryy parent = ry

F_1 Generation

rryy ↓ \ RRYY →	R	Y
ry	RrYy	RrYy
ry	RrYy	RrYy

- Genotype of these plants was RrYy, their phenotype was producing rounded-yellow seeds.
- **F_2 generation :** Plants of the F_1 generation of dihybrid cross produce four types of gametes **RY**, Ry, rY, ry. Of these gametes, RY and ry are similar to those of the P_1 generation.

F_1 First filial generation

- **Phenotype :** All the plants formed show round and yellow seeds.
- **Genotype :** All are heterozygous RrYy.

- The offspring obtained through such a cross is called **dihybrid.**
- The plants with RrYy genes produced four different kinds of combinations of gametes. These are **RY, Ry, rY and ry.**

F_2 Second filial generation

Female gametes ↓ \ Male gametes →	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

Phenotypes

- **9** Round and yellow seeds
- **3** Round and green seeds
- **3** Wrinkled and yellow seeds
- **1** Wrinkled and green seeds

Total phenotypic ratio : 9 : 3 : 3 : 1

F_2 Dihybrid offspring

Dihybrid	Genotype	Phenotype	Total number
	RRYY	Round and yellow	1
	RRyy	Round and green	1
	RRYy	Round and yellow	2
	RrYY	Round and yellow	2
	RrYy	Round and yellow	4
	Rryy	Round and green	2
	rrYy	Wrinkled and yellow	2
	rrYY	Wrinkled and yellow	1
	rryy	Wrinkled and green	1
Ratio	1 : 1 : 2 : 2 : 9 : 3 : 3 : 1 4 : 2 : 2 : 1 : 1		16

10. Some human characters

- **Some dominant and recessive characteristics of human beings**

Dominant	Recessive
Rolling tongue	Non-rolling tongue
Presence of hair on arms	Absence of hair on arms
Black and curly hair	Brown and straight hair
Free ear lobe	Attached ear lobe

- In each human cell, there are 46 autosomes consisting of 22 pairs of chromosomes that decide the body characters and 1 pair of sex chromosomes or allosomes that decide the sex of the individual.
- Every pair is given a number based on its length and characteristics.
- There are 44 + XX chromosomes in every cell of human females and 44 + XY chromosomes in every cell of human males.

11. Genetic disorders

- Genetic disorders are caused due to abnormalities in chromosomes and mutations in genes.

(1) Causes of genetic disorders

- **Changes in the number of chromosomes :** Increase or decrease in number.
- Deletion of any part of the chromosome.
- Translocation of chromosomes.

Chapter Heredity and Variation

Page 6 | Chromosomal, Monogenic and Mitochondrial Disorders

2 Disorders due to chromosomal abnormalities

- **Cause: Numerical changes in chromosomes.**

Disorder	Down Syndrome	Turner Syndrome	Klinefelter Syndrome
Cause of disorder	Trisomy 21	Abnormality in sex chromosome number	Abnormality in sex chromosome number
Total chromosomes	47, extra chromosome with the 21st pair	45 (44 + XO)	47 (44 + XXY)
Symptoms	• Mental Retardation Short lifespan • Life expectancy • Short height • Short wide neck • Flat nose • Short fingers • Scanty hair • Single horizontal crease on palm	• One X chromosome less • Sterile females • Improper growth of reproductive organs • Mental retardation	• Extra X chromosome in the cells • Sexually sterile males • Under-developed reproductive organs

3 Monogenic disorders

- A sudden change (mutation) in a normal gene can convert it into a defective gene. The defective gene causes a monogenic disorder.
- Around 4000 different monogenic disorders are known.
- Each gene has a specific sequence to make a definite product (an enzyme), but the defective gene cannot do so. Either the essential enzyme is not produced or it is made in a deficient amount.
- Due to this, some metabolic reactions do not occur in the body and unwanted toxic substances may accumulate.
- Hence, these disorders are called inborn errors of metabolism.
- Such abnormal metabolism may lead to death at a tender age.
- **Examples:** Hutchinson's disease, Tay-Sachs disease, galactosaemia, phenylketonuria, sickle cell anaemia, cystic fibrosis, albinism, haemophilia, night blindness, etc.

(i) Albinism

- In albinism, the body cannot produce melanin.
- As a result, the skin becomes pale and eyes look reddish pink due to absence of melanin pigment in the retina and sclera.
- The person has white hair and lacks normal skin colour.



Albinism
(lack of melanin)

(ii) Sickle cell anaemia

- Sickle cell anaemia is a monogenic disorder. The mutation in the foetus causes this disease.
- Due to mutation, the glutamic acid is changed to valine. Normal haemoglobin has glutamic acid as the 6th amino acid in its molecular structure.
- When it is replaced by valine, the shape/structure of the haemoglobin molecule changes.
- This change causes oval erythrocytes (RBCs) to become sickle-shaped.
- Therefore, the oxygen-carrying capacity of haemoglobin is reduced and clumping and destruction of erythrocytes occurs.



Normal RBCs
(oval / disc-shaped)

Sickle-shaped RBCs
(in sickle cell anaemia)

Sickle-cell anaemia – Symptoms

(i) Swelling of hands and legs,
(ii) Pain in joints, severe general body aches,
(iii) Frequent colds and cough,
(iv) Constant low grade fever,
(v) Exhaustion,
(vi) Pale face,
(vii) Low haemoglobin content.

Diagnosis of sickle-cell anaemia

'Solubility Test' and 'Electrophoresis' are performed to detect sickle cell anaemia.

Remedies for sickle cell anaemia

(i) Marriages between the carriers or sufferers should not take place.
(ii) A tablet of folic acid daily should be taken by a patient of sickle cell anaemia.

(4) Mitochondrial disorders



- The mitochondrial DNA is inherited from mother only.
- If the mitochondrial DNA is mutated, then the disorders produced are called mitochondrial disorders.

Question Bank

Q1. What is the total number of chromosomes in Klinefelter syndrome?
(A) 45 (44 + XO) (B) 46 (44 + XY)
(C) 47 (44 + XXY) (D) 47 (44 + XXX)
Ans. (C) 47 (44 + XXY)

Q2. Which amino acid replaces glutamic acid in sickle cell anaemia?
(A) Glycine (B) Valine
(C) Alanine (D) Leucine
Ans. (B) Valine

Q3. The mitochondrial DNA is inherited from —
(A) Father only (B) Mother only
(C) Both parents (D) Neither
Ans. (B) Mother only

Polygenic Disorders

- These disorders are transmitted only from the mother. For example, Leber's hereditary optic neuropathy.

(5) Polygenic disorders

(Disorders due to mutations in multiple genes)

- Mutations in more than one gene at the same time can cause a polygenic disorder.
- Severe foetal disorders may result from the effects of environment on the foetus. Examples: cleft lip, cleft palate, constricted stomach, spina bifida (a defect of the spinal cord), etc., are polygenic disorders.
- Diabetes, blood pressure, heart disorders, asthma, obesity are also polygenic disorders.
- Polygenic disorders do not strictly follow Mendel's principles of heredity. They arise from a complex interaction between environment, life style and defects in several genes.

12. Inter-relationship between tobacco addiction and cancer :

(1) Tobacco is either smoked or chewed. Any kind of tobacco product is carcinogenic.

- Nicotine in the tobacco is the main carcinogenic agent.

(2) Harmful effects of smoking :

(i) adverse effects on digestion
(ii) instability and trembling of fingers
(iii) dry cough causing sleeplessness
(iv) shortening of life span
(v) chronic bronchitis
(vi) cancer of the lungs, mouth, larynx (voice box), pharynx, pancreas, urinary bladder, etc.
(vii) central and peripheral nervous system affected
(viii) arteriosclerosis and hypertension
(ix) visual disorders or tremors

(3) Harmful chemicals in the tobacco smoke :

(i) pyridine
(ii) ammonia
(iii) aldehyde furfural
(iv) carbon monoxide
(v) nicotine
(vi) sulphur dioxide
(vii) minute carbon particles



QUESTIONS & ANSWERS

*** Q. 1 Complete the following sentences by choosing the appropriate words from the brackets :**

(Inheritance, sexual reproduction, asexual reproduction, chromosome, DNA, gene)

(1) Hereditary characters are transferred from parents to offspring by ..... , hence they are said to be structural and functional units of heredity.

(2) Organisms produced by ..... show minor variations.

(3) The component which is in the nuclei of cells and carries the hereditary characteristics is called ..... .

(4) Chromosomes are mainly made up of ..... .

(5) Organisms produced through ..... show major variations.

Ans. (1) gene (2) asexual reproduction (3) chromosome (4) DNA (5) sexual reproduction

Q. 2 Choose the correct alternative and write it along with its allotted alphabet :

(1) Information necessary for ..... synthesis in the cell is stored in DNA.

(a) protein *(b) glucose*
(c) fat *(d) minerals*

(2) The structure in the nucleus of cells that carries the hereditary characteristics is called the ..... .

(a) genome *(b) chromosome*
(c) mitochondria *(d) nucleolus*

Multiple Choice Questions (MCQs)

3 Crab's chromosome number is ..... whereas in humans it is ..... .

(a) *200, 46* (b) 20, 200
(c) *20, 46* (d) 46, 200

4 ..... discovered DNA while studying the white blood cells.

(a) Watson, Crick (b) Frederick Miescher
(c) Alexander Fleming (d) Gregor Johann Mendel

5 The RNA molecule which, according to the message of the mRNA, carries the amino acid up to the ribosomes is called ..... .

(a) *messenger RNA* (b) *ribosomal RNA*
(c) *transfer RNA* (d) *All the above*

6 The experiments performed on pea plants are presented by the '.....' method.

(a) *Mendel square* (b) *Punnet Square*
(c) *Mendel columns* (d) *Punnet columns*

7 Down syndrome is a disorder arising due to ..... abnormality.

(a) *chromosomal* (b) *mental*
(c) *physical* (d) *physiological*

8 ..... retardation is the most prominent characteristic of all the genetic syndromes.

(a) *Mental* (b) *Physical*
(c) *Emotional* (d) *Financial*

9 Normal haemoglobin has ..... as the 6th amino acid in its molecular structure.

(a) *valine* (b) *aspartic acid*
(c) *glutamic acid* (d) *proline*

10 ..... disorders are inherited from the mother only.

(a) *Chromosomal* (b) *Genetic*
(c) *Mitochondrial* (d) *Physical*

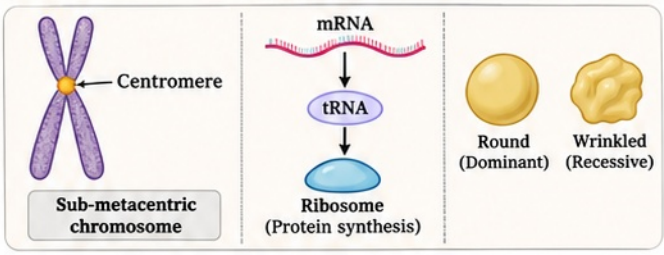
Answers (MCQs)

(3) (a) 200, 46 **(4)** (b) Frederick Miescher
(5) (c) transfer RNA **(6)** (b) Punnet Square
(7) (a) chromosomal **(8)** (a) Mental
(9) (c) glutamic acid **(10)** (c) Mitochondrial

Q. 3 State whether the following statements are True or False :

Note : Students should write whether the given statement is True or False. There is no need to correct a false statement and rewrite it.

(1) Wrinkled seed is the dominant character.
(2) Some of the Mendelian principles now form the basis of modern genetics.
(3) In a Monohybrid ratio, the phenotypic ratio is 9 : 3 : 3 : 1.
(4) The centromere is exactly at the mid-point in a sub-metacentric chromosome.
(5) Segments of the RNA molecule are called genes.
(6) The tRNA molecule carries the information of protein synthesis from genes.
(7) In Albinism, the body cannot produce glutamic acid.



Ans. (1) False (2) True (3) False (4) False
(5) False (6) False (7) False

Q. 4 Note the relationship between the first two words and suggest suitable words in the fourth place :

*(1) 44 + X : Turner's syndrome : : 44 + XXY : .....
*(2) 3 : 1 : Monohybrid : : 9 : 3 : 3 : 1 : .....
*(3) Women : Turner's syndrome : Men : .....
(4) Male : 44 + XY : : Female : .....
(5) Homozygous : rr : : Heterozygous : .....
(6) Centromere exactly at the mid-point : Metacentric : : Centromere at the end of chromosome : .....
(7) J-shaped chromosome : Acrocentric : : V-shaped chromosome : .....
(8) Down's syndrome : 47 chromosomes : Turner's syndrome : .....

Ans. (1) Klinefelter's syndrome (2) Dihybrid
(3) Klinefelter's syndrome (4) 44 + XX
(5) Rr (6) Telocentric
(7) Sub-metacentric (8) 45 chromosomes

Ans. (1) Klinefelter's syndrome (2) Dihybrid (3) Klinefelter's syndrome (4) 44 + XX (5) Rr (6) Telocentric (7) Metacentric (8) 45 chromosomes.

Q. 5 Find the odd one out and give reason :

(1) Adenine, thymine, Uracil, Guanine.

Ans. Uracil (Others are nitrogenous bases present in DNA.)

(2) Absence of hair on arms, Brown and straight hair, Attached ear lobes, Rolling tongue.

Ans. Rolling tongue (Others are recessive human genes.)

(3) Green pod, Tall plant, White flower, Axillary flower.

Ans. White flower (Others are dominant characters of the pea plant.)

(4) Cystic fibrosis, Klinefelter's syndrome, Hutchinson's disease, Albinism.

Ans. Klinefelter's syndrome (Others are monogenic disorders.)

Q. 6 Match the columns / How are the items in groups A, B and C inter-related ?

A	B	C
Leber hereditary optic neuropathy	44 + XXY	Pale skin, white hair
Diabetes	44 + X	Men are sterile
Albinism	Mitochondrial disorder	Women are sterile
Turner syndrome	Polygenic disorder	This disorder arises during development of zygote.
Klinefelter syndrome	Monogenic disorder	Effect on blood-glucose level.

Ans.

A	B	C
Leber hereditary optic neuropathy	Mitochondrial disorder	This disorder arises during development of zygote.
Diabetes	Polygenic disorder	Effect on blood-glucose level.
Albinism	Monogenic disorder	Pale skin, white hair
Turner syndrome	44 + X	Women are sterile
Klinefelter syndrome	44 + XXY	Men are sterile

Q. 7 Complete the table :

(1) Chromosome Number :

Sr. No.	Organism	No. of chromosomes
(1)	.....	46
(2)	.....	48
(3)	Crab	.....
(4)	.....	04
(5)	.....	48
(6)	Maize	.....

Ans.

Sr. No.	Organism	No. of chromosomes
(1)	Man	46
(2)	Potato	48
(3)	Crab	200
(4)	Roundworm	04
(5)	Frog	48
(6)	Maize	20

Heredity and Variation

Notes and Question Bank

Q. (2) Complete the following table :

Sr. No.	Disorder	Type	Cause
(1)	Albinism	.....	.....
(2)	.....	.....	Amino acid valine instead of glutamic acid
(3)	.....	Mitochondrial disorder	.....
(4)	Cleft lip and palate	.....	Mutations in multiples genes

Ans.

Sr. No.	Disorder	Type	Cause
(1)	Albinism	Monogenic disorder	No synthesis of melanin
(2)	Sickle cell anaemia	Monogenic disorder	Amino acid valine instead of glutamic acid
(3)	Leber hereditary optic neuropathy	Mitochondrial disorder	Mutations in multiple genes
(4)	Cleft lip and palate	Polygenic disorder	Mutations in multiples genes

Q. (8) Give definitions :

(1) Gene : The sequence of nucleotides present in the DNA molecule which contains the information to synthesise protein is called a gene.

(2) Genetics : Genetics is the branch of biology which studies the transfer of characteristics and genes of organisms from one generation to the next.

(3) Heredity : Transfer of characteristics from parents to offspring is called heredity.

(4) Phenotype : External appearance or visible characteristics of organisms is called a phenotype.

Important Definitions (Contd.)

(5) Genotype : The pairs of genes (factors) responsible for the visible characteristics is called a genotype.

(6) Monohybrid : The cross between two living organisms with only one pair of contrasting characters is called monohybrid.

(7) Dihybrid : When in a cross between the two organisms has two pairs of contrasting characteristics under consideration, then such a cross is called dihybrid cross.

(8) Dominant : Dominant is the term used for a genetic trait which expresses itself even in the presence of a recessive trait and which shows the presence even in the heterozygous condition is called a dominant.

(9) Recessive : Recessive is the term used for a genetic trait which cannot express itself in the presence of a dominant trait but remains masked in F_1, generation and which can show presence only in the absence of a dominant trait are called recessive.

(10) Homozygous : When both the genes are same in a genotype, then it is called homozygous. Homozygous can be either homozygous dominant or homozygous recessive based on the type of combination the alleles.

(11) Heterozygous : When both the genes are different in a genotype then it is known as heterozygous. In heterozygous genotype, only dominant allele expresses itself.

(12) DNA Fingerprinting : DNA fingerprinting is the technique by which the sequence of genes in the DNA of a person is identified.

Q. (9) Distinguish between the following :

(1) DNA and RNA.

Ans.

DNA	RNA
1. DNA means Deoxyribonucleic Acid.	1. RNA means Ribonucleic Acid.

Page 11 : Key Questions and Answers

Q. (Source Questions)	Ans.
1. In a DNA molecule two nucleotide strands are intertwined around one another, making the DNA double helix.	2. RNA has a single nucleotide strand. It folds upon itself.
2. The nitrogen bases are adenine, thymine, cytosine and guanine in the DNA molecule.	3. The nitrogen bases are uracil, cytosine adenine and guanine in the RNA molecule.
3. The sugar present in the DNA molecule is deoxyribose sugar.	4. The sugar present in the RNA molecule is ribose sugar.
4. DNA is responsible for transmission of genetic characters.	5. RNA is mainly responsible for protein synthesis.

(2) Phenotype and Genotype

Ans.

Phenotype	Genotype
1. Phenotype refers to the outward appearance of an individual such as shape, colour, sex, etc.	1. Genotype refers to the genetic composition of an individual.
2. Phenotype can be observed directly in an individual.	2. Genotype can be determined from the ancestry of an individual.
3. Individuals resembling each other may or may not have the same genotype.	3. Individuals possessing the same genotype usually have the same phenotypic expression.
4. The phenotypic ratio obtained in the F_2 generation of a monohybrid cross is **3 : 1.**	4. The genotypic ratio obtained in the F_2 generation of a monohybrid cross is **1 : 2 : 1.**

Key Terms

- **Phenotype :** The observable external traits of an individual (e.g. shape, colour, sex, etc.).
- **Genotype :** The genetic makeup (allelic composition) of an individual.

(3) Monohybrid and Dihybrid cross

Ans.

Monohybrid cross	Dihybrid cross
1. Crosses involving a single pair of alleles are called monohybrid crosses.	1. Crosses involving two pairs of alleles are called dihybrid crosses.
2. Monohybrid crosses yield a phenotypic ratio of 3 : 1 in the F_2 generation.	2. Dihybrid crosses yield a phenotypic ratio of 9 : 3 : 3 : 1 in the F_2 generation.
3. Genes are not assorted to form new combinations of characters.	3. Genes are assorted to form new combinations of characters.

(4) Dominant characters and Recessive characters

Ans.

Dominant characters	Recessive characters
1. The characters that are expressed in the F_1 generation are called dominant characters.	1. The characters that are not expressed in the F_1 generation (i.e. they are prevented from expressing themselves) are called recessive characters.
2. Tall habit and red colour of the flower in pea plants are dominant characters.	2. Dwarf habit and white colour of the flower in pea plants are recessive characters.
3. Dominant character is expressed in the presence of one or both the dominant genes.	3. Recessive character is expressed only when both the recessive alleles of a gene are present.
4. Dominant characters cannot be masked by recessive characters.	4. Recessive characters can be masked by dominant characters.

Remember

- **F_1 generation :** First filial generation (offspring of P generation).
- **F_2 generation :** Second filial generation (offspring of F_1).

(5) Turner's syndrome and Klinefelter's syndrome.

Ans.

Turner's syndrome	Klinefelter's syndrome
1. Turner's syndrome has single X chromosome along with autosomes in every cell.	1. Klinefelter's syndrome has two X chromosomes and a single Y chromosome with autosomes in every cell.
2. Turner syndrome has 45 chromosomes in each cell of her body with (44 + XO) combination.	2. Klinefelter's syndrome has 47 chromosomes in each cell of his body with (44 + XXY) combination.
3. Their phenotype is like a female but they are sterile and underdeveloped.	3. Their phenotype is like a male but they are sterile.

Q. 10 Name the following :

(1) Dominant characters in human beings.

Ans. Rolling tongue, presence of hairs on arms, black and curly hair, free ear lobes are some of the dominant characters in human beings.

(2) Recessive characters in human beings.

Ans. Non-rolling tongue, absence of hairs on arms, brown and straight hair, attached ear lobes are some of the recessive characters in human beings.

(3) Disorders caused due to abnormalities in chromosome number.

Ans. Down's syndrome, Turner's syndrome and Klinefelter's syndrome are disorders caused due to abnormalities in chromosome number.

(4) Symbols to denote the sickle cell disorder.

Ans. Symbols for sickle cell anaemia : Normal person = AA, Carrier of the disease = AS, Sufferer of sickle cell = SS.

(5) Tests of the sickle cell anaemia.

Ans. (1) 'Solubility Test', (2) Confirmatory diagnostic test called 'Electrophoresis'.

(6) Harmful chemicals are present in the tobacco smoke.

Ans. Harmful chemicals like pyridine, ammonia, aldehyde furfural, carbon monoxide, nicotine, sulphur dioxide, etc.

Q. 11 Complete the paragraph by choosing the words given in the bracket :

(consumption, smoking, tremors, cancer, uncontrolled, harmful)

Tobacco smoke contains many ..... chemicals. They cause ..... cell division. Tobacco smoke is full of minute carbon particles which causes normal tissue of the lung to transform into thickened black tissue. This causes ..... . While chewing tobacco or tobacco products much of the extract is absorbed into the body. Excessive tobacco ..... may cause cancer of lips or tongue, visual disorders or..... . To protect one's body from cancer one must avoid ..... and consumption of tobacco and tobacco products in any form.

Ans. Tobacco smoke contains many **harmful** chemicals. They cause **uncontrolled** cell division. Tobacco smoke is full of minute carbon particles which causes normal tissue of the lung to transform into thickened black tissue. This causes **cancer**. While chewing tobacco or tobacco products much of the extract is absorbed into the body. Excessive tobacco **consumption** may cause cancer of lips or tongue, visual disorders or **tremors**. To protect one's body from cancer one must avoid **smoking** and consumption of tobacco and tobacco products in any form.

Q. 12 Answer the following questions :

(1) What is a gene? What are its functions?

Ans. (1) Chromosomes are made up of DNA molecule.

Genes and DNA : Key Points

1. The DNA molecule is made up of nucleotides, each consisting of a nitrogenous base, deoxyribose sugar and phosphoric acid. The sequence of nucleotides in DNA is called a gene. A segment of DNA is thus considered a gene.
2. Since the nucleotide sequence of DNA varies from one organism to another, there are many genes in different organisms.
3. Due to the structure and function of genes, every cell is determined. The functions of cells are also regulated by genes.
4. The information about protein synthesis is also stored inside genes.
5. Genes are functional units of heredity. They transmit the hereditary characters of the parents to their offspring. A father's genes, in the form of his DNA, pass to the offspring through his sperms, while a mother's DNA passes through her ovum.
6. Therefore, there are many resemblances between parents and their children.

(2) Express your opinion about the use of DNA fingerprinting. (Open-ended question)

Ans. (1) DNA fingerprinting is the ultramodern technique. The sequence of genes in the DNA of any person can be understood using this technique.
(2) This technique is used to identify the criminals in forensic cases.
(3) Identification of the unclaimed dead bodies can also be done using DNA fingerprinting.
(4) The DNA sequence of mother, father and child can be matched with each other. In case of disputed parentage, therefore DNA fingerprinting technique is used.
(5) If unknown remains are obtained during excavation, then they can be identified using the same technique.



DNA Fingerprinting

(3) Which were the seven pairs of contrasting traits in pea plant that were studied by Mendel?

Ans. The seven pairs of contrasting traits in pea plant studied by Mendel are as follows :

No.	Trait	Contrasting traits (pairs)
(1)	Stem height	Tall plants / Dwarf plants
(2)	Flower colour	Red flowers / White flowers
(3)	Flower position	Axial position / Terminal position
(4)	Pod colour	Green pods / Yellow pods
(5)	Pod shape	Full pods / Constricted pods
(6)	Seed colour	Grey seeds / White seeds
(7)	Seed shape	Round seeds / Wrinkled seeds

(4) What are F_1 and F_2 generations? Why are genotypic and phenotypic ratios different in these generations?

Ans. (1) F_1 is the first filial generation consisting of the offspring born to their parents i.e. P_1 generation.
(2) F_2 is the second filial generation consisting of the offspring born to F_1 generation.
(3) F_1 generation has all heterozygous individuals born to homozygous dominant and homozygous recessive parents. Due to dominant genes, the recessive gene is masked and therefore the phenotype of all the individuals of the F_1 generation look alike. Genotypically they all are heterozygous.
(4) F_2 generation is formed because two heterozygous parents are crossed. In such a case, if the recessive genes are combined in a double dose then only the recessive character appears among the offspring.
(5) F_2 generation therefore has 3 : 1 ratio in a monohybrid cross. Three individuals of the same phenotype are seen due to one homozygous dominant and two heterozygous individuals.
(6) Genotypically, the ratio is 1 : 2 : 1 in the proportion of 1 homozygous dominant, 2 heterozygous and 1 homozygous recessive.

Thus, the genotype and phenotype ratios are different in F_1 and F_2 generations.

(5) What are the reasons for hereditary disorders?

Ans. (1) Due to some reasons, the chromosome number changes. Any chromosome can be added or deleted during cell division causing the numerical disorder.

Key Terms at a Glance

Gene	A segment of DNA that carries information for a trait.
DNA fingerprinting	A modern technique to identify an individual from the sequence of genes in DNA.
P_1 generation	The parental generation (parents).
F_1 generation	First filial generation (offspring of P_1).
F_2 generation	Second filial generation (offspring of F_1).
Genotype	The genetic make-up of an individual (e.g. TT, Tt, tt).
Phenotype	The observable characters of an individual (e.g. tall or dwarf).
Heterozygous	An individual having two different alleles (e.g. Tt).
Homozygous	An individual having two identical alleles (e.g. TT or tt).
Monohybrid cross	A cross between two individuals differing in only one pair of contrasting traits.
Dominant gene	A gene that expresses its character even in the heterozygous condition.
Recessive gene	A gene whose character is expressed only in the homozygous condition.

(2) Sometimes, there is **structural aberrations** in the chromosome. Part of the chromosome may be **lost or added**. Sometimes there is **translocation** in which the sequence of genes is changed.

(3) Sometimes there is **sudden mutation** causing deleterious effects.

(4) The **number of chromosomes** present, their **sex-related types**, the **types of genes** on the chromosomes, whether they are **dominant** and **recessive**, etc. decide the type of genetic disorders and their **inheritance pattern**.

6 What are the functions carried out by National Health Mission?

Ans. (1) The National Health Mission has two units, viz. the National Rural Health Mission which started since April 2005 and the National Urban Health Mission that is functional since 2013.

(2) National Urban Health Mission looks after the different plans of health of urban people.

(3) Under the National Health Mission scheme, disease like sickle cell anaemia is kept under control by diagnosis.

(4) The main objective of both these missions is to strengthen the rural and urban health facilities, controlling various diseases and illnesses, increasing public awareness about health and offering financial assistance to patients through various schemes.

7 What harm is caused to body by tobacco consumption?

Ans. (1) Tobacco consumption in any form can cause cancer. Hence it is proved to be a carcinogenic.

(2) Smoking of cigarettes and bidi has adverse effects on the process of digestion. The constant smoke creates irritation and burning sensation in the throat and results in the cough. Excessive smoking leads to instability and trembling of fingers. A dry cough causes sleeplessness.

(3) Addiction to any tobacco product causes shortening of the life span due to chronic bronchitis, arteriosclerosis, high blood pressure, pericarditis, cancer of the lungs, mouth, larynx (voice box), pharynx, pancreas, urinary bladder, etc.

Key Harms of Tobacco

- Carcinogenic (can cause cancer).
- Affects digestion; irritates and burns the throat.
- Causes cough, instability and trembling of fingers.
- Dry cough leads to sleeplessness.
- Shortens life span; may cause chronic bronchitis, arteriosclerosis, high blood pressure, pericarditis and cancers of lungs, mouth, larynx, pharynx, pancreas, urinary bladder, etc.

(4) Excessive tobacco consumption may cause cancer of lips or tongue, visual disorders or tremors caused due to ill effects on central and peripheral nervous system.

* (8) Why is it necessary for people to have their blood examined before marriage?

(Open-ended question)

Ans. Many people are carriers of the genetic diseases. They are not aware of it too. If mother and father both are carriers, there are greater chances of their young one to become a victim of genetic condition. E.g. Sickle-cell anaemia, thalassaemia, different types of syndrome conditions, etc. may be present in such children. By performing blood tests on prospective husband and wife, the chances of having abnormal children can be understood. The genetic disorders cannot be cured any time. It comes with the birth and ends with the death.

Moroever, if one wants to be protected from the dreadful disease like AIDS, then the blood test of our prospective spouse should be done in time.

If the Rh blood group is incompatible with each other, the disorder of erythroblastosis foetalis can occur resulting in death or retardation of the child. If mother is Rh negative and father is Rh positive the children born to such couple may have many problems. Taking into consideration all these facts, it becomes evident that people should have their blood examined before marriage.

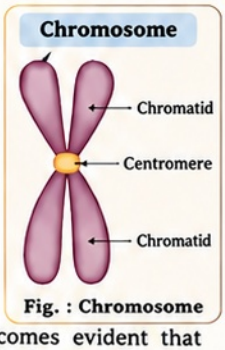


Fig. : Chromosome

* (9) Is it right to avoid living with a person suffering from a genetic disorder?

(Open-ended question)

Ans. Genetic disorder is not an infective disease. It is transmitted only from parents to offspring. Similarly, it is never cured. The genetic disorder is formed in the embryonic stage due to abnormal genome. This spreads to all the cells of the patient. Later in life it is incurable. Such people need help and support. Few borderline syndromes are given remedial education and made self-sufficient. The parents of syndrome also need lots of empathy and support. It is the moral duty of the society to include such people and make them a part of the mainstream.

Inclusion and Support

- A genetic disorder is not communicable (not an infective disease).
- It arises due to an abnormal genome in the embryonic stage and is incurable.
- Such individuals need care, support and empathy.
- Some borderline syndromes can be managed by remedial education to make them self-sufficient.
- Their parents also need understanding and support.
- They should be included in the society and mainstreamed.

We must help them as much as we can. It is therefore wrong to avoid living with a person suffering from a genetic disorder.

Q. 13 With neat and well labelled diagrams, answer the following questions :

(1) What is meant by 'chromosome'? Explain its types.

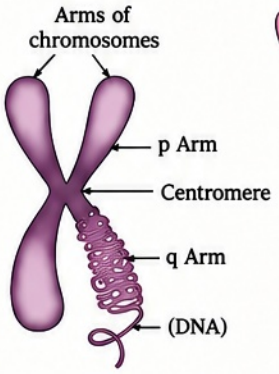


Fig. 16.2 : Organisation of Chromosome

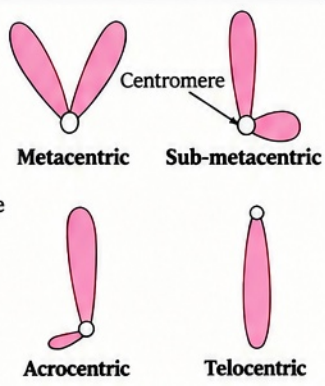


Fig. 16.3 : Types of Chromosome Arm

Ans. (1) Chromosomes are definite number of thread-like structures present in the nucleus of every cell.

(2) They are visible only at the time of cell division. Each chromosome is made up of DNA, RNA and proteins.

(3) The nucleotides present in the DNA molecule form sequences. These nucleotides are arranged in a specific order in the DNA molecules, which is called a gene. The segments of DNA thus form genes. Genes are carried to the next generation through chromosomes. This is an important function of chromosomes.

(4) Each chromosome has a 'primary constriction' or 'centromere'. The centromere divides the chromosome into two parts. Each part is called an 'arm' of the chromosome. The centromere has a specific position in each chromosome. Depending on the position of the centromere and the length of the arms of the chromosome, there are four types of chromosomes.

Key Facts – Chromosomes

- Chromosomes are definite in number in every cell.
- Composed of DNA, RNA and proteins.
- Gene = a specific sequence of nucleotides in DNA.
- Genes are carried to the next generation via chromosomes.
- Centromere (primary constriction) divides a chromosome into two arms—p arm and q arm.
- Based on centromere position and arm length, chromosomes are of four types: metacentric, sub-metacentric, acrocentric and telocentric.

(5) Types of chromosomes :

(A) Types of chromosomes according to the position of centromere and the shape.

Name	Structure	Chromatid arm	Visible pattern
(1) Metacentric	Centromere in the centre of chromosome	The arms of this chromosome are equal in length	V shaped
(2) Sub-metacentric	The centromere is somewhere near the mid-point in chromosome	One arm is slightly shorter than the other.	L shaped
(3) Acrocentric	The centromere is near one end of chromosome	One arm is much smaller than other	J shaped
(4) Telocentric	The centromere is right at the end of chromosome	Consists of only one arm	I shaped

(B) Types of chromosomes according to their structure and function.

1. **Homologous chromosomes :** If the pair consists of similar chromosomes by shape and organization, they are called homologous chromosomes.
2. **Heterologous chromosomes :** If the chromosomal pairs are not similar they are called heterologous chromosomes.
3. **Sex chromosomes or allosomes :** The chromosomal pair that decides the sex of the sexually reproducing organisms.
4. **Somatic chromosomes or autosomes :** Chromosomes that decide the body characters other than sex.

(2) What are the functions of DNA molecule?

Ans. (1) DNA molecule is present in all living organisms right from bacteria to human beings.

(2) DNA is called the master molecule as it controls the functioning, growth and reproduction of the cell.

(3) The two main functions of the DNA are protein synthesis and the transmission of hereditary characters.

(4) Genes are actually the sequence of nucleotides in the DNA molecule. They are transmitted from parental generation to the offspring through chromosomes. In this way DNA is responsible for the genetic characters of the organisms.

***(3) Describe the structure of the DNA molecule.**

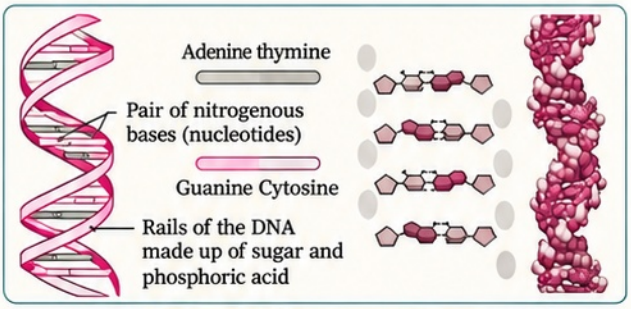


Fig. 16.4 : Structure of DNA

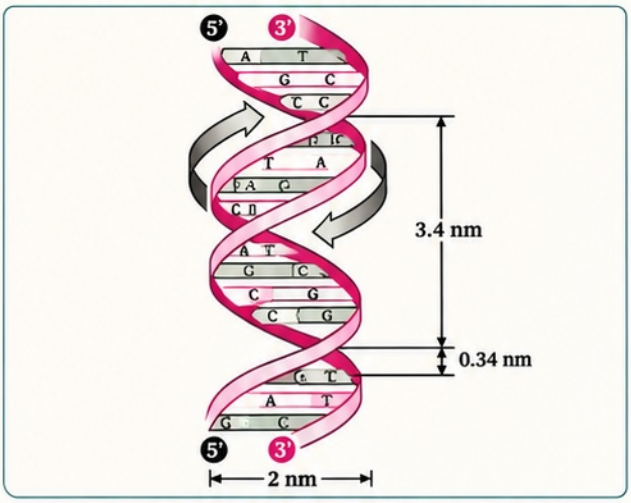


Fig. 16.5 : DNA (Watson and Crick's Model)

Ans.

1. DNA structure was proposed by Watson and Crick in 1953.
2. According to their model, DNA has two parallel threads of nucleotides which are coiled around each other. Thus it is called a double helix.
3. Each strand is made up of nucleotides. Each nucleotide consists of a Deoxyribose sugar, phosphoric acid and a nitrogen base.
4. The sugar and phosphate joins alternately with each other on the DNA strand.
5. There are two main types of nitrogenous bases, known as purines and pyrimidines. Adenine and guanine are two purines while cytosine and thymine are two pyrimidines.
6. One purine is bonded with other pyrimidine in the opposite strand of the DNA helix.
7. As a rule, purine adenine always makes pair with pyrimidine thymine by double hydrogen bond.
8. Purine guanine always pairs with pyrimidine cytosine by triple hydrogen bond.
9. These hydrogen bonds appear like steps of the ladder.

***(4) Explain the structure, function and types of RNA.**

Ans. (1) The RNA is a nucleic acid that is found both in nucleus as well as in cytoplasm. It is made up of single strand that is twisted upon itself.

(2) The components of RNA are nucleotides which are made up of ribose sugar, phosphoric acid and the nitrogenous bases.

(3) The four types of nitrogenous substances in the RNA are adenine, guanine, cytosine and uracil. Thymine is not present in the RNA molecules. Of these adenine and guanine are purines while cytosine and uracil are the pyrimidines of the RNA. RNA is found both in nucleus as well as in cytoplasm.

(4) The RNA is of three types according to its functions.

(A) Messenger RNA or mRNA : During the process of protein synthesis the mRNA molecule carries the information of coding a particular protein from genes i.e. DNA chain. DNA is present in the cell nucleus but the information is passed to the ribosomes in the cytoplasm with the help of messenger RNA. Then later the proteins are synthesized in the ribosomes.



(B) Ribosomal RNA (rRNA) : Ribosomal RNA is present in the ribosomes which are protein factories of the cell. This rRNA molecule performs the function of protein synthesis.

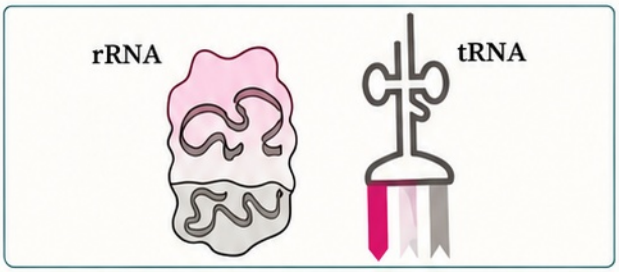


Fig. 16.6 : Types of RNA

(C) Transfer RNA (tRNA)

tRNA is the RNA that carries amino acids to the ribosomes according to the message of mRNA. It is also called transfer RNA. It brings the appropriate amino acid to the ribosome during protein synthesis.

Q. 14 Give scientific reasons :

1 The albino person has pale skin and white hair.

Ans. Albinism is a hereditary (monogenic) disorder in which the body cannot produce melanin, the pigment responsible for colour. As a result, the hair, eyes and skin remain uncoloured, leading to pale skin and white hair.

2 The person suffering from sickle-cell anaemia has reduced oxygen carrying capacity of the haemoglobin.

Ans. Sickle-cell anaemia is a monogenic disorder caused by a change in the structure of haemoglobin. In the normal haemoglobin molecule, glutamic acid at the 6th amino acid position is replaced by valine due to a genetic mutation. This alters the shape of haemoglobin, making the erythrocytes sickle-shaped instead of normally biconcave. These abnormal red blood cells carry less haemoglobin and hence have a reduced oxygen carrying capacity. In addition, they can clump together and also get destroyed, which further lowers the oxygen carrying capacity.

3 Mitochondrial disorders are transmitted only from mother to her progeny.

Ans. Mitochondria are cell organelles present in the cytoplasm only. The cytoplasm of the egg is inherited by the embryo, whereas the sperm contributes only its nucleus and not cytoplasm. Therefore, any abnormality of mitochondria is passed on to the offspring only through the mother.

4 Polygenic disorders do not strictly follow Mendel's principles of heredity.

Ans. Polygenic disorders are caused by mutations in more than one gene. Their severity is influenced by environmental factors on the foetus. Thus, they show a complex interaction between environment, lifestyle and genetic defects involving several genes, and therefore do not strictly obey Mendel's principles.

5 There is hypertension due to smoking.

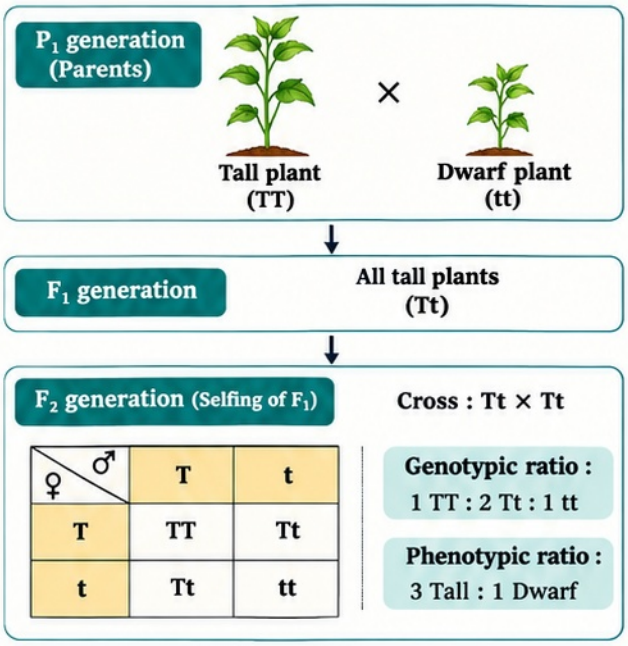
Ans. Tobacco in cigarette or *bidi* contains nicotine, a harmful chemical. Nicotine affects the central and peripheral nervous systems. It causes arteries to become hard (arteriosclerosis). When the lumen of arteries becomes narrow due to arteriosclerosis, blood flow is reduced, leading to a rise in blood pressure. This results in hypertension.

Q. 15 Explain the following :

★ **(1) Explain Mendel's monohybrid progeny with the help of any one cross.**

Ans.

1. Monohybrid cross is a cross involving only one pair of contrasting characters or traits.
2. In P_1 generation, tall and dwarf plants are selected for the cross. The progeny obtained from such a cross are all tall. Hence, Mendel called tallness as the dominant character.
3. In the F_1 generation, all the plants are tall as tallness is dominant. The dwarf character is not expressed and thus remains recessive.
4. Though all the plants in F_1 are tall, the character of dwarfness is hidden in them, Hence, the phenotype of all F_1 individuals is tall, but their genotypes are heterozygous.
5. When two such F_1 plants are crossed with each other, two types of phenotypes are seen in the F_2 generation—tall and dwarf.
6. In the F_2 generation, three genotypes are obtained, viz. homozygous dominant, homozygous recessive and heterozygous.



Monohybrid Cross : Conclusion

- (7) According to the data collected after the experiments of Mendel, out of 929 pea plants, 705 were tall and 224 were short.
- (8) This shows that in F_2 generation there is phenotypic ratio of 3 tall : 1 dwarf whereas genotypic ratio was 1TT : 2Tt : 1tt.

Mendel's experiment of the monohybrid cross

Parental Generation (P_1)

Phenotype : Tall Dwarf
Genotype : TT tt
Gametes : T t

First Filial Generation (F_1) : Tt
Phenotype : Tall

Parental Generation (P_2) **Selfing in F_1**

Phenotype : Tall Tall
Genotype : Tt Tt
Gametes : T and t T and t

First Filial Generation (F_2)

Male gamete / Female gamete	T	t
T	TT (Tall)	Tt (Tall)
t	Tt (Tall)	tt (Dwarf)

(2) Explain Mendel's dihybrid ratio with the help of any one cross.

OR

What is a dihybrid cross? Explain with a suitable example.

Ans. (1) The cross involving two pairs of contrasting characters is known as a dihybrid cross.

(2) Two pairs of contrasting characters, namely seed shape and seed colour were considered by Mendel.

(3) The characters of round and yellow seeds are dominant characters while wrinkled and green seeds are recessive characters.

(4) In parental (P_1) generation, a plant with round and yellow seeds was crossed with a plant bearing wrinkled and green seeds. RRYY (round and yellow) was one parent while rryy (wrinkled and green) was another parent.

- (5) Each pair of genes segregates independently from the other.
- (6) RRYY parents produced RY type of gametes, and rryy parents produced ry type of gametes.
- (7) In F_1 generation all the offsprings produced had genotype RrYy and their phenotype was yellow and round.
- (8) The F_1 generation produced four different types of gametes, viz. RY, Ry, rY and ry. Of these, RY and ry were parental combinations whereas Ry and rY were recombinations.
- (9) From four male gametes and from four female gametes, 16 different combinations are produced as follows : (Refer to the table)
- (10) The phenotypic ratio produced was 9 : 3 : 3 : 1 which is called a dihybrid ratio.

Mendel's experiment of dihybrid cross

Parental Generation (P_1)

Phenotype : Rounded-yellow seeds Wrinkled-green seeds
Genotype : RRYY rryy
Gametes : RY ry

First Filial Generation (F_1) : RrYy
Phenotype : (Rounded-yellow seeds)

Parental Generation (P_2) : Selfing in F_1

Phenotype : Rounded-yellow seeds Rounded-yellow seeds
Genotype : RrYy RrYy
Gametes : RY, Ry, rY, ry RY, Ry, rY, ry

Second Filial Generation (F_2)

Gametes	RY	Ry	rY	ry
RY	RRYY (Round yellow)	RRYy (Round yellow)	RrYY (Round yellow)	RrYy (Round yellow)
Ry	RRYy (Round yellow)	RRyy (Round green)	RrYy (Round yellow)	Rryy (Round green)
rY	RrYY (Round yellow)	RrYy (Round yellow)	rrYY (Wrinkled yellow)	rrYy (Wrinkled yellow)
ry	RrYy (Round yellow)	Rryy (Round green)	rrYy (Wrinkled yellow)	rryy (Wrinkled green)

Dihybrid Cross : Key Results

- In a dihybrid cross (e.g. seed shape and seed colour in pea), the F_2 generation shows the following results :

Phenotypic ratio :	Genotypic ratio :
• Round-Yellow : 9 • Wrinkled-Yellow : 3 • Round-Green : 3 • Wrinkled-Green : 1	= 1 : 1 : 2 : 2 : 4 : 2 : 2 : 1 : 1 RRYY – 1, RRYy – 2, RRyy – 1, RrYY – 2, RrYy – 4, Rryy – 2, rrYY – 1, rrYy – 2, rryy – 1

*(3) Distinguish between monohybrid and dihybrid cross.

For the answer, refer to answer to Q. 9 (3).

(4) Explain the inheritance of sickle-cell anaemia disease.

Ans.

1. For showing inheritance of sickle cell anaemia, normal person is denoted as AA, carrier as AS and patient as SS.
2. There is normal progeny if both parents are normal. In such case, the mother and father both have normal gene (AA) and the child born to them through cross (AA X AA) also becomes AA.
3. If one of the parent is carrier (AS) then the chance of child becoming carrier of sickle-cell is 50% and the chance of remaining normal is also 50% (AS X AA).
4. If one of the parent is normal (AA) and another parent is patient of sickle-cell anaemia (SS), then all the children will become carriers (AA X SS).
5. If both the parents are carriers then the proportion of children will be 25% normal, 25% affected with sickle-cell anaemia and 50% will be carrier (AS X AS).
6. If one of the parent is carrier and other is a patient then the proportion among children will be 50% carrier and 50% affected with disease (AS X SS).
7. If both the parents are affected then all the children will be affected too (SS X SS).

Q. 16 Write a brief note on each :

(1) Human Genome Project (HGP) :

(1) Scientists and biotechnologists from different countries came together to carry out Human Genome Project in early 1990.

2. In June 2000, scientists of HGP and Celera Genomics Corporation which is a private industry in USA collectively announced the discovery of the complete DNA sequence of the human genome.
3. According to the finding of HGP, the number of genes in the human genome is about 20,000 to 30,000. Later, genomic sequence of many micro-organisms were also discovered.
4. Due to research in genomics, genetic diseases can be diagnosed and properly treated. The exact causative gene for a particular disease can be located. Thus, the remedy can be found for such incurable genetic diseases in near future.
5. On the Website : www.genome.gov the entire information of HGP is available.

*(2) Down syndrome :

Ans.

1. Down syndrome is a disorder arising due to autosomal abnormality.
2. Down syndrome was the first discovered and described chromosomal disorder in human beings.
3. This disorder is also called trisomy 21 as the 21st chromosome occurs thrice in such patients. Due to extra 21st chromosome, in each cell of Down syndrome there are 47 chromosomes.
4. These children are mentally retarded due to improper brain growth.

   Their main symptoms include short height, short wide neck, flat nose, short fingers, scanty hair, single horizontal crease on the palm.
5. They have short life span ranging to about 16 to 20 years.

Note : Their face resembles a Mongolian face, therefore in olden days this disorder was called Mongolism. But now due to ethical reasons such phrase is not used.

(3) Turner syndrome :

Ans.

1. The sex chromosomal abnormality causes Turner's syndrome.
2. In Turner syndrome there are 22 pairs of autosomes and a single X chromosome.
3. The chromosomal complement of Turner syndrome is thus (44 + XO) i.e. there are 45 chromosomes in each cell.

» Turner's Syndrome (Contd.)

(4) The phenotype of this person is that of the female. But she is underdeveloped and sterile.

(5) They are unable to have children due to improper growth of the reproductive organs.

Monogenic Disorders

*** (4) Monogenic disorders :**

Ans. (1) When there is mutation in any single gene into a defective gene, it causes disorders which are called monogenic disorders.

(2) There are about 4000 different disorders of this type.

(3) Due to abnormal genes, the protein synthesis does not take place properly. Therefore, their products are either produced in deficient amounts or are not produced at all. It causes abnormal metabolism leading to death at a tender age.

(4) E.g. Hutchinson's disease, Tay-Sachs disease, galactosaemia, phenylketonuria, sickle cell anaemia, cystic fibrosis, albinism, haemophilia, night blindness, etc. are all monogenic disorders.

(5) Sickle Cell Anaemia : Symptoms and Treatment

I. Symptoms of sickle-cell anaemia :

1. Swelling of hands and legs,
2. Pain in joints,
3. Severe general body aches,
4. Frequent colds and cough,
5. Constant low grade fever,
6. Exhaustion,
7. Pale face,
8. Low haemoglobin content.

II. Treatment / Remedies :

1. Premarital blood check up should be done as this disease spreads only through reproduction.
2. A carrier or sufferer should never marry with another carrier or sufferer. Or if they marry they should not have children.
3. A person suffering from sickle cell anaemia should regularly and daily take a tablet of folic acid.
4. Since it is a genetic disorder, there is no treatment to completely cure this disease.

Note : A carrier or sufferer should never marry with another carrier or sufferer.

*** Q. 17 Complete the tree diagram below based on types of hereditary disorders :**

