



212en25

25

HEREDITY

Why does a human baby look like a human being and also resemble closely or distantly the parents, a grandparent or even cousins or uncle/aunt? Why is a kitten like a miniature cat to look at? Why does a seedling acquire the same kinds of leaves, stem or flowers as the parent plants? Why do, for that matter, all organisms resemble, in structure, their parents? The passing down of similar characters generation after generation is termed '**heredity**'. Heredity is controlled by genes. Differences in gene combinations lead to '**variations**' or differences even among members of the same family. The science of heredity and variation is termed **Genetics**.

In this lesson you shall learn some fundamental aspects of genetics such as Mendel's laws, chromosomes, genes, how DNA duplicates, what makes a fertilized egg male or female and what kind of advice helps to prevent hereditary disorders.



OBJECTIVES

After completing this lesson, you will be able to:

- *define the term heredity and variation;*
- *state pattern of Mendelian inheritance;*
- *describe the location, structure and function of chromosomes and genes and briefly explain DNA fingerprinting and its significance;*
- *outline the process of DNA replication;*
- *give an account of the four blood groups in humans and the manner of their inheritance;*
- *explain the chromosomal basis of sex determination in humans;*
- *list certain hereditary disorders and mention hazards of consanguineous marriage;*

- emphasize the relevance of genetic counseling;
- briefly describe the human genome;
- outline the salient points of genetic engineering.



Notes

25.1 HEREDITY AND VARIATION

Heredity

The passing down of characters from parents to offspring is termed **heredity**. Heredity is controlled by genes.

Variation

Look around and you shall find so many differences even between individuals of the same kind. For example, in the rose garden, the colour of rose flowers on different plants are different, puppies, of the same mother dog are different in their coat colour (Colour of their hair or fur). All such individual differences are termed **variations**. Variations are due to genes or environment. Now perform the following activity to find variation in the ear lobes of human beings



ACTIVITY 25.1

Check your ear lobes and those of your friends and family members. The lower end of the ear lobe may be attached or free as shown in the figure 25.1. This feature of the ear lobe is hereditary. Observe the ear lobes of your parents and your siblings (brothers and sisters) and note from which of your parents you have inherited this feature. You may similarly try and observe the rolling of your tongue and notice



Fig. 25.1 Ear lobe, whether free or fixed, is hereditary



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who all in your family can roll their tongues. Similarly, you may note who all in your family can curve the tip of the thumb backwards and who all cannot, for this ability is also hereditary. Note any two other features such as colour of the eyes or the shape of the nose or any other feature among your friends. Differences that you note are termed **variations**.

You may perform other activities to find out about the occurrence of variation for yourselves. For example the variation with respect to the ability to rolling the tongue or having the hitch hikers thumb (figure 25.2 a and b)



(a)

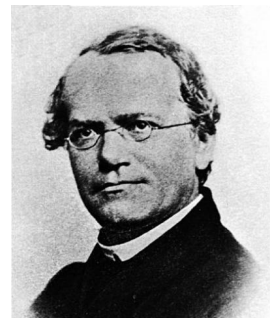


(b)

Fig. 25.2 (a) Rolling Tongue (b) Hitch hikers thumb

25.2 CONTRIBUTION OF GREGOR JOHANN MENDEL, THE FOUNDER OF GENETICS

The question about heredity intrigued many scientists of yesteryears. Gregor Johann Mendel (1822 -1884), an Austrian monk undertook the laborious task of finding the answers. He selected some pea plants, grew them year after year, compiled a lot of data, analysed and postulated certain laws of inheritance for the first time. His remarkable work, however, got recognized years after his death when Correns, Tschermak and Hugo de Vries came to the same conclusions as Mendel did, after independently carrying out experiments in their own countries.



Gregor Johann Mendel (1822-1884)

25.2.1 Mendel's Laws of Inheritance

Mendel's laws state that:

1. Every feature or character (for example colour of flowers, height of plant, colour and texture of seed, colour and texture of pods and location of flower on the plant) is controlled by a pair of **factors**. During the formation of gametes, one factor goes to one gamete and its pair to another gamete. **Thus the two factors of a pair segregate or separate during gamete formation.** Upon fertilization, the combination of factors expresses the feature. **(1st law).**

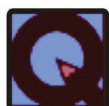
2. Out of the two factors controlling a certain feature, the **dominant** one may express inspite of the presence of the other. The other factor expresses only in the absence of the dominant factor and is termed **recessive (2nd law)**.

For example: factor for tallness in the pea plant always expresses in the offspring but dwarfness expresses only if factor for tallness is not present.

Mendel also postulated two other laws called 'law of parental equivalence' and, 'law of independent assortment'. They are not elaborated here.

The first law defined here is universal. Scientists later observed deviations from the other Mendelian laws.

Sutton in 1902, working with grasshopper chromosomes confirmed that **Mendelian factors were present in chromosomes**. Still later the **term 'gene' replaced the term 'factor.'** In other words genes are present on chromosomes.



INTEXT QUESTIONS 25.1

1. What is meant by the terms (1) Heredity and (2) Variation.

2. Why is Mendel considered as the founder or father of genetics?

3. Formulate a sentence to demonstrate your understanding of the terms '**dominant**' and '**recessive**.'

4. Name the scientist who discovered that Mendelian factors are present on chromosomes.

5. Give the synonym for Mendelian factor.

25.3 CHROMOSOMES AND GENES

Genes are responsible for heredity. They are present on chromosomes at fix points.

25.3.1. Chromosome

The nucleus of every cell (except RBC of mammals) contains a fixed number of chromosomes. In all the cells of eukaryotes, chromosomes show the following typical characteristics -



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1. They are present in **pairs**, one from the father and the other from the mother.
2. They can be **seen only during cell division**. In a non-dividing cell, they appear in the nucleus as a jumbled up network termed **chromatin**.
3. The paired chromosomes are present **in a fixed number**. A fixed set of chromosomes is termed the '**diploid**' (paired) number and designated as 2n.
4. Each chromosome is made of one molecule of the chemical called DNA or deoxyribonucleic acid and some proteins.
5. Before cell division, the DNA molecule of a chromosome replicates (duplicates) to give two molecules of DNA which are called '**chromatids**.' The two chromatids of a chromosome remain attached at a point called **centromere** and separate to form two chromosomes during cell division.

In the bacteria, only **one chromosome** (that is only one molecule of DNA) is present and since there is no well formed nucleus, the single chromosome lies in the cytoplasm in the region termed **nucleoid**. (Fig. 25.3a and 25.3b)

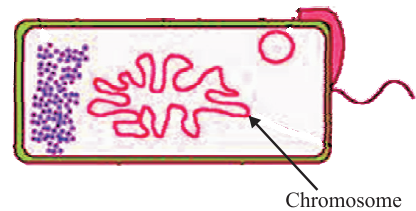


Fig. 25.3a Bacterial chromosome

25.3.2 Human chromosomes

Every cell of a human being contains 46 chromosomes. In other words, the diploid number in humans is 46. This can be expressed as $2n=46$. Since gametes contain only half the number of chromosomes or the **haploid** number, a sperm and an ovum or an egg has only 23 chromosomes.

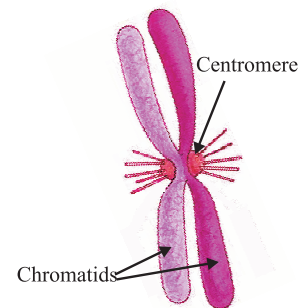


Fig. 25.3b Eukaryote chromosome

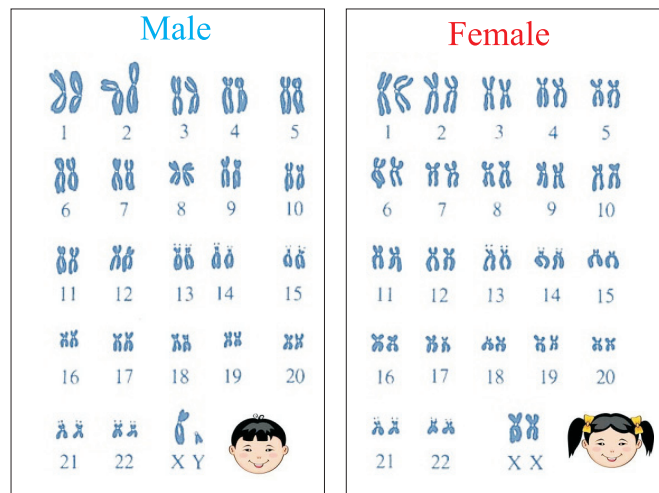


Fig 25.4 Human Chromosomes

Every species has a fixed number of chromosomes.

Each chromosome seen in the figure (25.4) possesses two identical chromatids joined by the centromere. The chromatids become independent chromosomes when they acquire a centromere at anaphase of cell division and ultimately move to different cells.

The chromosomes may be photographed from dividing cells at metaphase stage of mitosis and then displayed in pairs as in figure 25.5.

A pair of similar chromosomes (one received from the father and one from the mother) containing the same genes are termed **homologous chromosomes**.

Out of the 23 pairs of chromosomes, 22 pairs are termed **autosomes**. The 23rd pair (X and X in females and X and Y in males) are called **sex chromosomes**.

X chromosome has several genes, some of which are necessary for survival. Y chromosome bears genes for maleness only. One such gene is the 'testes determining factor'.

25.3.3 Genes

Genes are present on chromosomes. The Genes are the 'Mendelian factors,' present in pairs (one received from the father, other from the mother), on the chromosomes. Thus, one member of a pair of genes present on the chromosomes has its pair on the homologous chromosome at the same location.

Genes are the bearers of hereditary characters or the units of heredity. It has already been mentioned that a chromosome contains one molecule of the chemical called DNA. Genes present on chromosome are **segments of DNA**. (figure 25.5)

Since every individual begins life as a single cell, the DNA contained in all the cells of an individual is identical.

25.3.4 DNA fingerprinting

You might have heard that criminals can now be identified by DNA tests called "DNA fingerprinting". This is because DNA of an individual is the same in each and every cell of the body and also resembles the DNA of parents. Needless to say this is because children inherit DNA from their parents. Just like the fingerprint, DNA of every individual is unique and even if a hair or drop

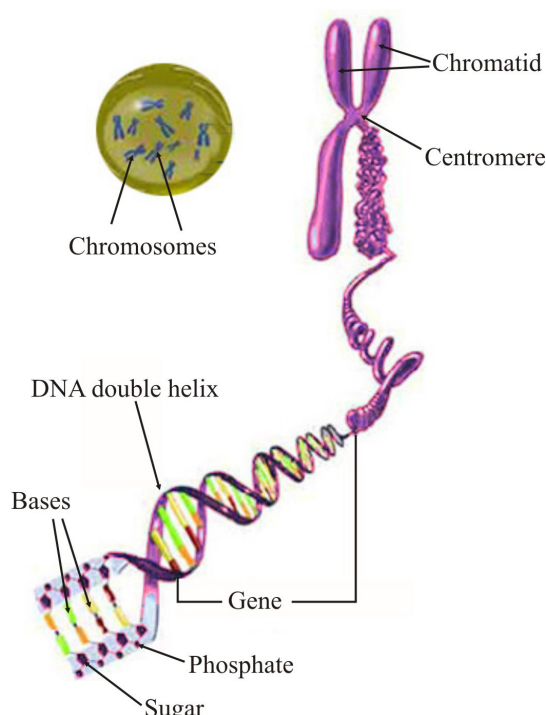


Fig 25.5 Location and Appearance of Chromosome and Genes

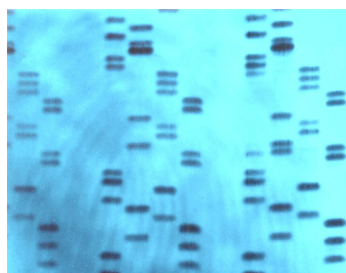


Fig. 25.6 DNA Fingerprint



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of blood or semen of the criminal is left at the site of the crime, it can be used to detect the DNA of the criminal and compared with that of the suspect to ascertain the truth. (Figure.25.6)

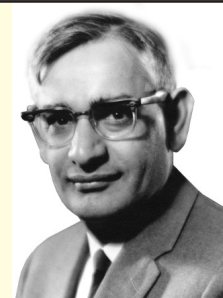


Did you know

Dr. Hargobind Khorana was the creator of man-made gene

It is a matter of pride that Nobel laureate Dr. Hargobind Khorana who was born in our country synthesized an artificial gene in the laboratory for the first time.

Dr. Khorana got the Nobel Prize in 1970 for this contribution to molecular biology.



Dr. Hargobind Khorana

25.4 THE DNA MOLECULE

A DNA molecule is a **polynucleotide**. (poly = many) It is made of units called **nucleotides**, each of which contains

- A nitrogenous base
- A deoxy ribose sugar
- A phosphate

There are four nitrogenous bases **Adenine, Guanine, Thymine** and **Cytosine** and hence four kinds of nucleotides in a DNA molecule.

The various combinations of these nucleotides in a segment of DNA form the different genes.

In physical structure, a DNA molecule is a double helix containing two polynucleotide strands.

25.4.1 DNA replication

Cell division takes place in a manner so that one cell divides into **two identical cells** with the **same number of identical chromosomes**. Therefore, prior to cell division, every chromosome should contain two **chromatids** made of two identical DNA molecules. This is achieved through the process of **DNA duplication** or **DNA replication**. (Figure 25.7) The major steps of DNA duplication are simplified below.

- i. The double stranded DNA molecule unwinds with the help of certain enzymes to expose two strands of DNA.



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- ii. A DNA polymerase enzyme catalyzes the formation of a new daughter strand which can form a double helix with one strand of parental DNA molecule. So two DNA molecules, each with a parental strand and a new strand get generated.
- iii. The two identical DNA molecules then become two chromatids which remain attached by a centromere.

Thus, upon DNA replication, each chromosome contains two identical molecules of DNA housed in its two chromatids. During cell division, the two chromatids, separate out as two chromosomes, one each, passing into the two daughter cells.

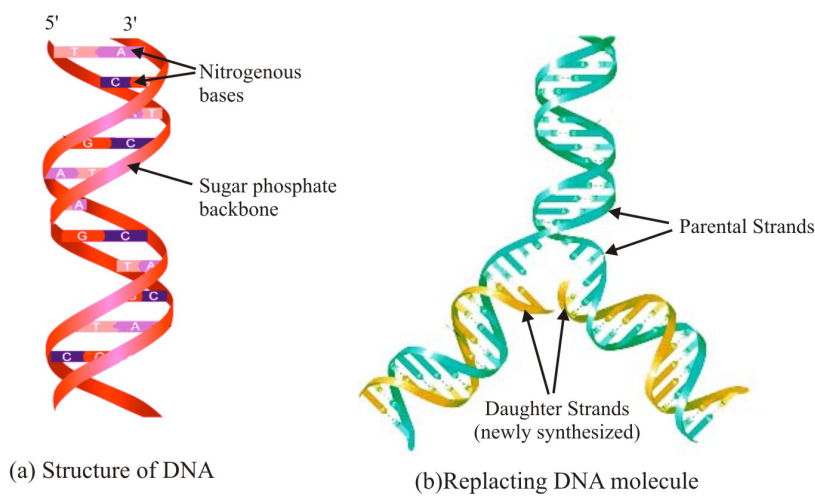
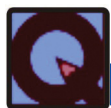


Fig 25.7 DNA Structure and Replication



INTEXT QUESTIONS 25.2

1. Name the sex chromosomes.

2. How many autosomes do humans have?

3. Why does an organism (except bacteria) have diploid number of chromosomes?

4. State any two typical features of chromosomes.

5. Define a gene with respect to its chemical nature.



Notes

Rh+ Rh-

2. _____

Rh- Rh-

3. _____

Why should you know your blood group?

In case of any emergency such as an accident, or a diseased condition, blood transfusion may be required. Only a matching blood group of the blood donor can be transfused. A person with blood group A can donate blood to patient with blood group A and AB. AB can receive blood from any of the four blood groups. O can receive blood only from O but donate blood to all four blood groups. Sometimes there may not be time or facility available for prior ascertaining of the blood group. Immediate blood transfusion is possible if the blood group is known. If unknown, the safest blood group for transfusion is O negative (O group and Rh-). B can donate to B and AB. O is the universal donor and AB is the universal recipient.

The entire human race can be divided into four groups on the basis of blood groups. Do you think further distinctions made by human beings on the basis of caste, creed and gender are justified?

25.6 SEX DETERMINATION IN HUMANS

The combination of sex chromosomes with autosomes determines whether the foetus will be a boy or a girl. (figure 25.8). The foetus develops from the zygote which is formed by the fusion of the two gametes, the male gamete or sperm and the female gamete or egg. Gametes are haploid [(have only 'n' number of chromosomes) while the zygote is diploid (2n)].

Ova or eggs are of one kind only. These contain 22 autosomes and a single X chromosome. Sperms are of two kinds (i) having 22 autosomes and one X chromosome, or (ii) having 22 autosomes and a Y chromosome (see figure 25.8). When X bearing sperm fuses with the egg, a female child results with 44 autosomes

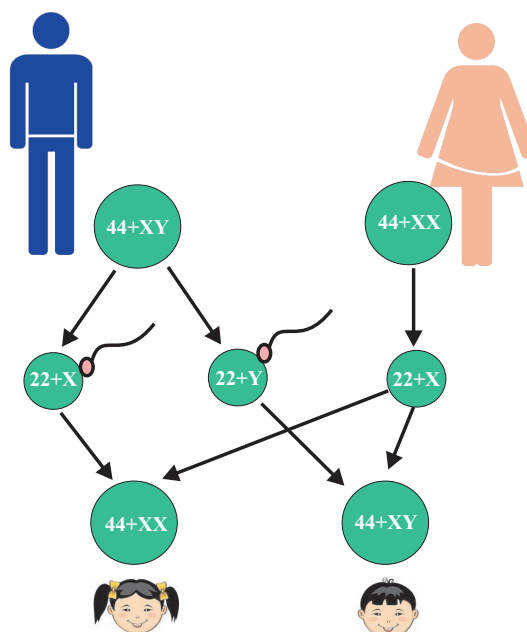


Fig. 25.8 Chromosomal basis of sex determination in Humans

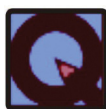


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and two X chromosomes. If Y bearing sperm fuses with the egg then a male child results with chromosomal constitution of 44 autosomes and one X and one Y chromosome.

You have already learnt about cell division earlier and know that at metaphase chromosomes are clearly seen lying at the equator and may be studied under the microscope or by taking a photograph. You can then easily identify and state that the chromosomes in a male human are 44 autosomes +XY and those in a female are 44 autosomes+ XX. It is, therefore, wrong to blame a woman if she does not bear a male child as is done in some ignorant families of our country. Sex of an individual is purely due to chance and neither the mother nor the father can be blamed.

The Pre-natal Diagnostic Techniques (Regulation and Prevention of Misuse) Act, 1994, was enacted and brought into operation from 1st January, 1996, in order to check female foeticide. The Act prohibits determination and disclosure of the sex of foetus. It also prohibits any advertisements relating to pre-natal determination of sex and prescribes punishment for its contravention. The person who contravenes the provisions of this Act is punishable with imprisonment and fine.



INTEXT QUESTIONS 25.3

1. What is a gene made of?

2. To which blood group would a person having genes IAi belong?

3. If a Y bearing sperm fuses with an egg, what will be the sex of the individual developing from the zygote?

4. How many X chromosomes can be found in the cells of the body of (i) a boy, and (ii) a girl.

5. How many molecules of DNA are present in one chromosome?

25.7 HEREDITARY DISORDERS

You already know that genes control all features of an organism. Some times a gene may change or **mutate** either in the gamete or zygote. Mutated gene

may not remain normal. Also, sometimes a defective gene present in the parent may not be expressed in the parent as the dominant normal member of its pair may mask the effect of the defective gene. But if the child inherits the defective gene from both the parents, the presence of the defective pair of genes has a harmful effect. **Such a disorder is termed hereditary or genetic disorder.** (figure 25.9)

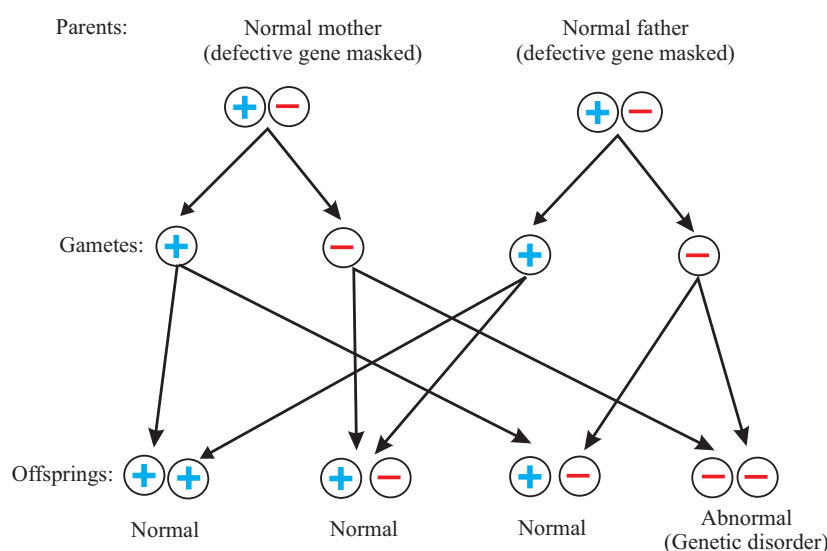


Fig 25.9 Hereditary or genetic disorder

There are several kinds of hereditary disorders, some of which may be caused due to presence of only one defective gene which is dominant or sometimes by the presence of two defective recessive genes. As shown above genetic disorders cannot be cured by medicines. Scientists are trying to discover methods by which a defective gene occurring in an individual may be removed or replaced by a normal gene. This is called **gene replacement therapy**.

25.7.1 Common genetic (hereditary) disorders

There are several genetic (hereditary) disorders. Three common hereditary disorders are Thalassemia, Haemophilia and Colour blindness.

(i) Thalassemia

Patients suffering from this disorder are unable to manufacture haemoglobin, the pigment present in red blood corpuscles which carries oxygen to tissues. This is because the pair of genes controlling haemoglobin production are defective. Thalassemics (persons suffering from thalassemia) require frequent blood transfusion in order to survive. The thalassemia gene is present on an autosome.

(ii) Haemophilia

Those persons suffering from haemophilia have either a defective gene or lack genes, which control production of substance responsible for blood clotting. In the



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absence of such substance blood does not coagulate. Once bleeding starts, it does not clot easily.

(iii) Colour-blindness

Different kinds of colour-blindness have been detected but in the most common form of the disorder, a person is unable to distinguish the blue colour from green. Again this is due to the presence of a defective gene or absence of the gene, responsible for colour vision.

The genes for both haemophilia and colour-blindness are located on X-chromosome, and hence, the disorder is passed down from mother to the son because a boy receives the X chromosome from the mother and Y chromosome from the father. In the mother, with two X-chromosomes, the defect does not show up. Also in the daughter, the effect of defective gene on X-chromosome inherited from mother may be masked by a normal gene on the X-chromosome, inherited from her father. Since X chromosome bears the defective gene, the son suffers from the genetic disorder, as male has only one X chromosome and one Y chromosome and so the defective gene does not get masked.

25.8 GENETIC COUNSELLING

Thalassemia is an autosomal genetic disorder, while, haemophilia and colour blindness are sex-chromosomal or X-chromosomal disorders. You would now appreciate why marriages between close relatives termed consanguineous marriages are discouraged. In marriages between relatives, chances of inheriting defective genes by the offspring is enhanced as both parents, being related may possess the same defective genes. If recessive, the two defective genes express when passed on to offspring by both parents. Therefore, it is essential to know the probability of a genetic defect in the offspring and seek the advice of genetic counsellors who are professionals. Genetic counselling helps one to know the chances of inheritance of a genetic disorder so that people can make informed decisions.

25.9 THE HUMAN GENOME

You can well imagine that human beings are complex in structure, behaviour and body functions. Thus, many genes control the features that make a human being. In 2003, it became possible to identify many of these genes of humans – their location on the chromosomes and the combinations of nucleotides that constitute them. All the various genes together constitute the **genome**. Knowing the human genome can help in finding and devising therapies for many genetic defects. This promises hope for people suffering from genetic disorders, as the location of every gene of the human genome is now known.

25.10 GENETIC ENGINEERING

Genetic Engineering is also called recombinant DNA technique. In this technique, gene from an organism of a species can be transferred to become part of the or genome of an organism belonging to another species which is then termed GMO **genetically modified organism**. The transfer is possible through “plasmids” present in bacteria. Plasmid is a circular DNA molecule found in bacteria. It is not part of the bacterial chromosome. Transfer is also possible through viruses that attack bacteria and are called bacteriophage. These carry genes from a cell culture (bacterial cells grown in a culture dish) and transfer into bacteria. A bacterial “clone” containing the required gene may then yield the “foreign” gene which can be used for replacing a defective gene during gene therapy. Genetic engineering also has many other benefits. Try and find out at least two such benefits. You may take the help of books or internet. (figure. 25.10)

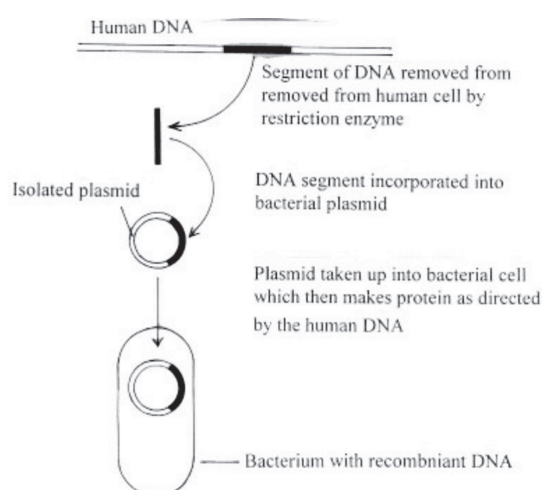
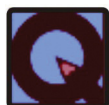


Fig. 25.10 Major steps in genetic engineering.



INTEXT QUESTIONS 25.4

1. What will be the blood group of an individual with genetic combination IA IB?

2. How can a person be normal for a trait even when carrying one defective gene for that trait?

3. Which is the safest blood group for donation if an accident victim of an unknown blood group has to be given immediate blood transfusion?

4. On which kind of chromosome, the autosomes or the sex chromosomes, are defective genes causing, Thallasemia, colour blindness and Haemophilia located?

5. Name the therapy in which a defective gene is substituted by a normal gene.



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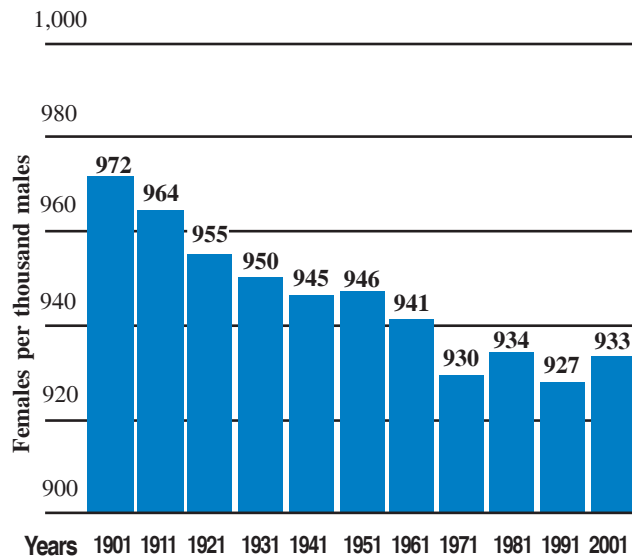


Notes

6. The given box diagram represents the ratio of females to males or the sex ratio in our country for 10 decades (1901 to 2001). Answer the following questions in the light of your knowledge of sex determination and the data presented in the box diagram.

- What does the bar diagram show? _____
- As per scientific knowledge regarding sex determination, what should be the sex ratio or the male to female ratio at a given point of time. _____
- Assign one reason to the trend showing deviation from the expected sex ratio.

- In what ways is such a trend unfavourable? _____
- Suggest a way by which such a trend can be stopped. _____



Graph related to sex ratio is included here

- Do you notice any reversal in the trend? What would you attribute it to?



WHAT YOU HAVE LEARNT

- Passing down of characters from parents to children is called heredity.
- Children of same parents differ because they possess different combinations of parental genes. These difference are termed variations. We are all human beings but can be distinguished easily from each other due to variation.
- Heredity and variation are due to genes and their varied combinations.



Notes

- Study of heredity is called Genetics.
- Mendel was the first to postulate laws of inheritance (heredity) and he said that heredity was due to “factors” and that every feature was controlled by a pair of factors which separate into different gametes during gamete formation.
- Another Mendelian law of inheritance stated that in a pair of genes one may be dominant and the other recessive. The dominant gene of the pair masks the effect of the recessive member of the pair.
- Sutton found out that “Mendelian factors” were the genes and that genes are present on chromosomes.
- Chromosomes are present in pairs in the nucleus and each is made of one molecule of DNA and proteins.
- The diploid number of chromosomes in humans is 46, of which 22 pairs are autosomes and 2 chromosomes X and Y are sex chromosomes.
- Genes are made of DNA. They are segments of the DNA molecule of the chromosome.
- A DNA molecule is a polynucleotide. Each of its nucleotides is made of a nitrogenous base, a sugar, and a phosphate.
- A DNA molecule is made of two strands of DNA helically coiled around each other.
- Before cell division, DNA in every chromosome replicates forming two identical DNA molecules which are present as the two chromatids forming the chromosome. In DNA replication, the two strands of a DNA molecule unwind and each acquires a new strand so that two molecules of DNA are formed.
- Sex determination in humans is based on combination of sex chromosomes. Females have two X-chromosomes, while males possess one X and one Y chromosome.
- Defective genes or absence of genes may cause genetic disorders e.g. thalassemia, haemophilia and colour blindness.
- Thalassemics lack genes responsible for production of haemoglobin so they need frequent blood transfusion for survival.
- Haemophiliacs bleed profusely. Their blood cannot coagulate as they lack genes for factors necessary for blood clotting. Hence their bleeding does not stop easily.



Notes

- Colour-blind people cannot distinguish the colour blue from the green due to defective genes for colour vision located on X-chromosomes.
- Thalassemia is an autosomal genetic disorder while, haemophilia and colour-blindness are sex chromosomal disorders.
- The collection of all the genes of a species constitute its genome. Human genome has been unravelled that is, location of all human genes on the chromosomes is now known.
- Genetic engineering involves transfer of a gene from one species into member of another species with the help of plasmids. Organisms carrying foreign genes, that is, genes of another species are called genetically modified organisms or GMO.
- DNA fingerprinting is a technique with the help of which the identity of a person can be known from the genetic make up.



TERMINAL EXERCISES

1. Which statement is true for 'genes'? Select the correct answer
 - (a) Genes are imaginary factors.
 - (b) Genes are fragments of DNA.
 - (c) Genes are present in the cytoplasm.
 - (d) Genes are not inherited.
2. What are "factors" named by Mendel called today?
3. What is the chemical nature of a gene? Name the three components of this chemical.
4. Where are genes located?
5. State two differences between autosomes and sex chromosomes.
6. Define heredity, variation, genetic disorder and sex chromosomes.
7. Why does DNA have to be duplicated before cell division?
8. Mention the main steps in DNA replication.
9. What will be the blood group of the following which contain the genes $I^A i$.
10. Why is haemophilia found mostly in boys?
11. With the help of a line diagram explain the chromosomal basis of a zygote developing into a male child.

12. What is the basis of sex determination in humans?
13. Write notes on any one genetic disorder.
14. What is meant by “gene replacement therapy”?
15. Rahul’s maternal grandfather (mother’s father) was colour-blind. What are the chances of Rahul being colour-blind if his father has normal colour vision?



Notes



ANSWERS TO INTEXT QUESTIONS

25.1

1. (i) Passing down of similar characters generation after generation.
(ii) Differences in gene combinations.
2. He had initiated work on heredity/genetics.
3. (i) The gene which may express in spite of the presence of the other (Dominant).
(ii) Expression only in the absence of the dominant gene. (Recessive).
4. Sutton
5. Gene.

25.2

1. X and Y
2. 22 pairs or 44 chromosomes
3. Because one chromosome of a pair is received from father and one from the mother.
4. (i) present in pairs, (ii) seen only during cell division, (iii) present in fixed number etc. (any other).
5. Genes are segments of a DNA molecule. So it is made of Deoxyribonucleic acid or DNA.
6.
 - Unwinding of double helix.
 - Formation of new molecules of DNA complimentary to each DNA strand
 - Winding of one new and one parental DNA strand
7. Because DNA of every individual is unique.

**Notes****25.3**

1. DNA
2. Blood group A
3. Male
4. For boy – 44 autosomes and one X and one Y chromosomes
For girl – 44 autosomes and 2 X chromosomes
5. One molecule of DNA

25.4

1. AB
2. The other member of the pair is dominant and masks the effect of the recessive gene.
3. 'O' positive.
4. Thalassemia – autosome, colour blindness and haemophilia on X chromosome.
5. Gene replacement therapy.
6.
 - Bar diagram shows the proportion of females in the population over a decade
 - 1:1
 - Female foeticide;
 - Male to female ratio becomes lopsided
 - Banning sex tests of unborn baby; increasing awareness
 - Yes, awareness and education/ No- with reasons.

MODULE - 6
NATURAL RESOURCES

- Lesson 26 Air and Water
Lesson 27 Metals and Non-metals
Lesson 28 Carbon and Its Compounds