# 30

# From Parents to Children – Elements of Genetics

Collect a few photographs of your family members and close relations. You will be surprised to find marked resemblances in physical features such as the shape of nose, eyes, hands, feet, forehead, colour and texture of hair and many other observable characterics.

Similarities between members of a family are due to 'heredity'. Heredity means passing down of characteristics from parents to children. Differences between members of the same family are due to different combinations of parental characteristics. These differences are termed 'variations'. Heredity and variation are due to genes and gene combinations and study of heredity is termed 'Genetics'. In this lesson, you will learn about some basics of genetics.

**OBJECTIVES**

After completing this lesson, you will be able to:

- define heredity and variation;
- highlight Mendel's and Sutton's contributions to genetics;
- state the number of chromosomes in a normal human being;
- state the distinction between autosomes and sex chromosomes;
- explain the chromosomal basis of sex determination in humans;
- name at least three genetic disorders;
- state the symptoms and explain the cause of haemophilia, colour blindness and thallasemia.

## 30.1 MENDEL AND SUTTON'S CONTRIBUTION TO GENETICS

**Gregor Johann Mendel** (1822-1884) an Austrian monk, was the first to observe the manner in which characteristic features pass down from parents to offsprings. He performed his experiments on the garden pea plant, which has striking contrasting characteristics such as, purple or white flowers, tall or dwarf plants, green or yellow seeds, which may also be either round or wrinkled. He selected

seven pairs of contrasting features in the pea plant and carried out selective breeding. For example, he chose a tall plant whose seeds always produced tall plants, and a dwarf plant from whose seeds only dwarf plants could be raised. He used the pollen of a tall plant to pollinate and fertilise the pea flower of a dwarf plant. Such a type of pollination that is done manually is known as artificial pollination. Similarly, he artificially pollinated plants with other contrasting features. After experimenting for several generations he was able to formulate certain 'laws of inheritance'. That was the beginning of genetics.

One of Mendel's laws states that for every feature or character (e.g. colour of flowers, height of plants, etc.) there is a pair of 'factors'. One factor each from each pair goes into the gamete (sperm or egg). Upon fertilisation, these factors express themselves according to a set pattern.

*Fig. 30.1 Gregor Johann Mendel (1822 - 1884)*

Gregor Johann Mendel was born in 1822. He grew up on a small farm in Northern Moravia, then in Austria. In 1847, he became a priest. In 1856, in the monastery of St. Thomas, Mendel began his historic research work with the garden pea and published his work entitled "Experiments on plant hybrids" in 1866. Unfortunately his work was rediscovered sixteen years after his death in 1900. The first significant contribution in the field of genetics was given by Mendel and so he is known as "the father of genetics".

Later in 1920, Sutton while observing grasshopper chromosomes confirmed that 'Mendelian factors' are present on chromosomes. The factors later got to be known as **genes**. It was accepted that genes are responsible for heredity. In other words, genes are the units of heredity. These are present at fixed loci (locations) on the chromosomes.

**ACTIVITY 30.1**

Check your ear lobes and those of your friends and family members. The lower end of the ear lobe may be attached or free. 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 who all in your family can roll their tongues and notice who all in your family can curve the tip of the thumb and who all cannot, for this ability is also hereditary. Also, note any two other features such as colour of eyes or shape of the nose or any other feature among your friends. Differences that you note are variations.

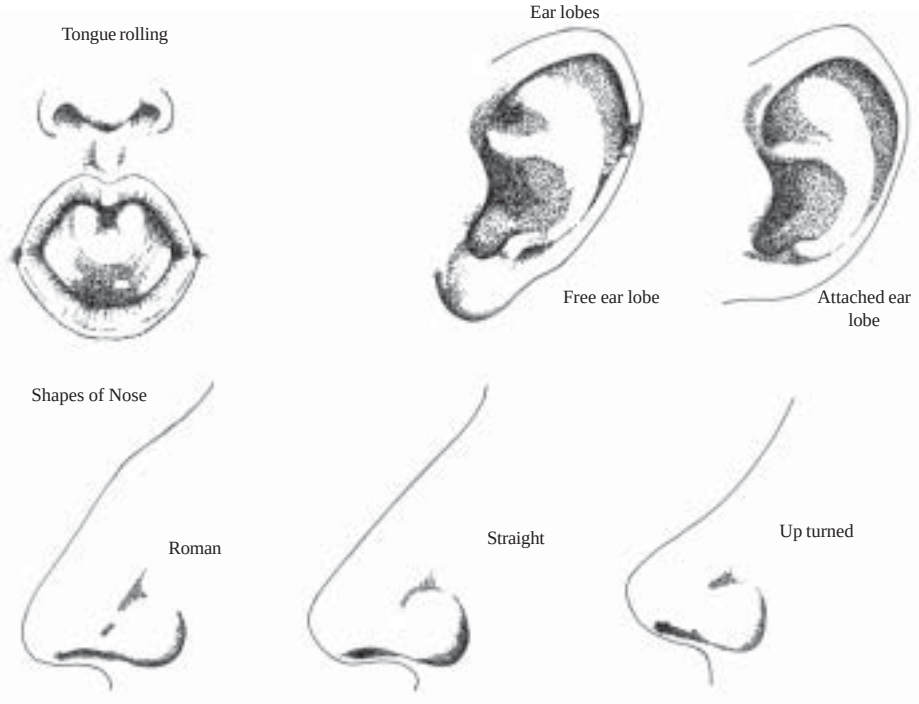



*Fig. 30.2 Some variations found in people*

## 30.2 HUMAN CHROMOSOMES

Every cell in the human body contains 46 (23 pairs) chromosomes. You have already learnt that chromosomes are present in the nucleus of a cell. Chromosomes are present in pairs. One chromosome of each pair comes from the father and the other from the mother. The chromosome number is thus a "diploid" (i.e. paired) number and is represented as 2n. The number of chromosomes remains constant in all normal human beings.

Of the 23 pairs of human chromosomes (2n = 46), one pair represented as X and Y-chromosomes have genes that determine the sex of an individual. X and Y chromosomes are, therefore, called sex chromosomes while, the rest 22 pairs are termed autosomes. You can see the chromosomes arranged according to size and also based on certain other considerations in the Fig. 30.3. Chromosomes can be seen only during cell division. Pairs of similar chromosomes (called homologous chromosomes) are selected and arranged in the mitotic metaphase of a dividing cell. You have

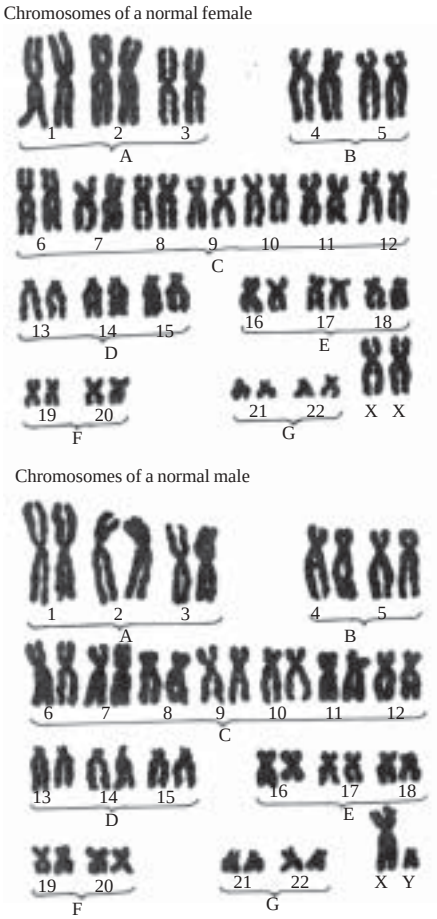



*Fig. 30.3 Human chromosomes*

already learnt about cell division earlier and know that at metaphase chromosomes are clearly seen lying at the equator.

**CHECK YOUR PROGRESS 30.1**

1. Who proposed that hereditary units are located on chromosomes?
2. What are 'Mendelian factors' called today?
3. How many autosomes do humans have?
4. Why are X and Y-chromosomes called sex chromosomes?
5. What is the diploid number of chromosomes in human body cells?

## 30.3 CHEMICAL NATURE OF GENES

By now you know that genes are bearers of hereditary characters and they are present on chromosomes. From the work of many scientists, today we know that genes are segments of chemical molecules **called DNA or deoxyribonucleic acid.** One chromosome contains one molecule of DNA and genes are fragments of this DNA molecule. You might have heard that criminals can now be identified by DNA tests called "DNA fingerprinting". This is because DNA of an individual is 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 parents. Just like the fingerprint, DNA of every individual is unique and even if a hair or drop 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 compare with the suspect to ascertain the truth.

**Dr. Hargobind Khorana: 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.

*Fig. 30.4 Dr. Hargobind Khorana*

## 30.4 SEX DETERMINATION IN HUMANS

In the earlier section you have learnt about autosomes and sex chromosomes. The gametes, i.e., sperms or eggs have haploid or half the number of chromosomes as you have learnt earlier in lesson 29 on gamete production and fertilization. Gamete has 22 autosomes and 1 sex chromosome. When an egg of the mother and sperm of the father fuse to produce a zygote, the diploid number is restored. Zygote develops into an individual whose sex depends on whether there are two X chromosomes or one X and one Y-chromosome. Zygotes having two X-chromosomes develop into females and zygotes with one X and one Y-chromosome develop into males.

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 below). When X bearing sperm fuses with egg, a female child results with 44 autosomes and two X chromosomes. If Y bearing sperm fuses with egg then a male child results with chromosomal constitution 44 autosomes (Fig. 30.5) and X and Y chromosomes.

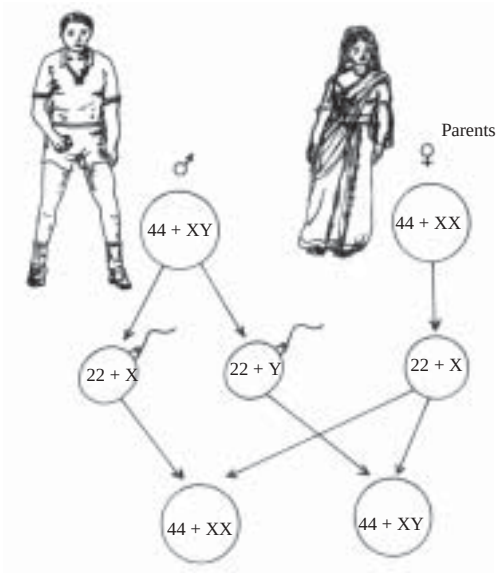



*Fig. 30.5 Chromosomes - the basis of sex determination in humans*

**Note :** Thus, the chromosomes in a male human are 44 autosomes + XY and that 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.

## CHECK YOUR PROGRESS 30.2

1. What is a gene made of?
2. Why is DNA fingerprinting a foolproof test?
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 (i) a boy, and (ii) a girl.
5. How many molecules of DNA are present in one chromosome?

## 30.5 INHERITANCE OF BLOOD GROUPS

You have already learnt in lesson 26 that every human being belongs to one of the four blood groups i.e., A, B, AB or O. The blood group of a person is inherited from parents and depends on the combination of genes for blood group inherited from either parent. One gene for blood group is inherited from the father and the other from the mother. These genes are designated as $I^A$, $I^B$ and i. The following table shows the combination of genes and the resulting blood group.

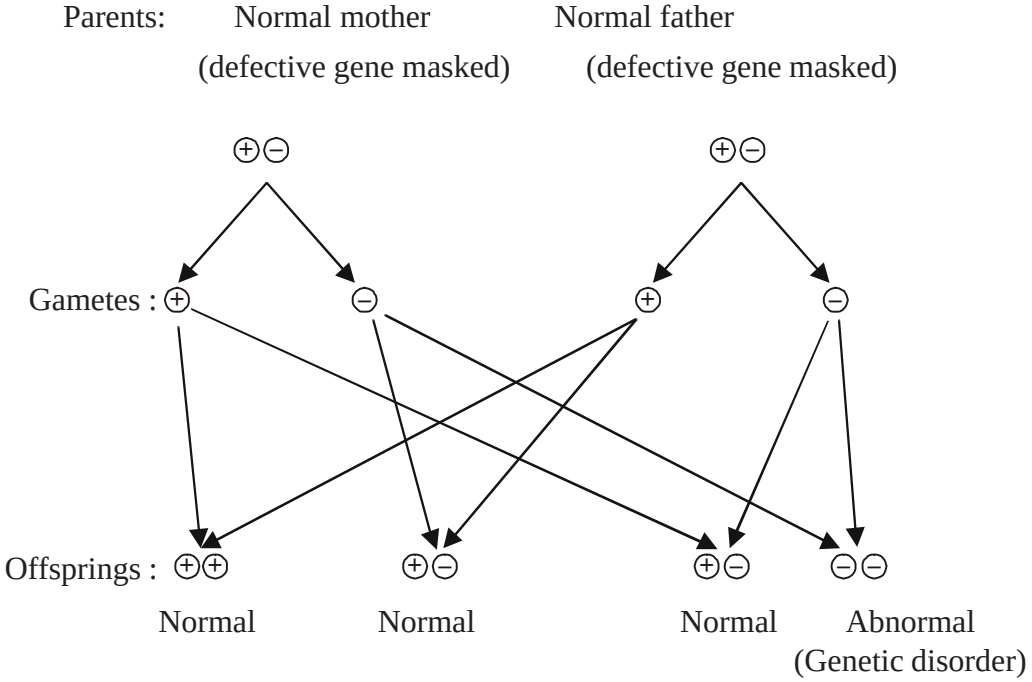
**Table 30.1: The combination of genes and the resulting blood group**

| Gene combination | Blood group |
|---|---|
| $I^A I^A$ or $I^A i$ | A |
| $I^B I^B$ or $I^B i$ | B |
| $I^A I^B$ | AB |
| I i | O |

Similarly the $Rh^+$ (Rhesus positive) blood group is inherited when one or two genes for $Rh^+$ antigen are present in a person. A person with $Rh^-$ (Rhesus negative) blood group lacks $Rh^+$ gene. Thus, $Rh^+$ $Rh^+$ or $Rh^+$ $Rh^-$ combinations result in $Rh^+$ blood group and $Rh^-$ $Rh^-$ into $Rh^-$ (Rh negative) blood group.

## 30.6 HEREDITARY (GENETIC) DISORDERS

Sometimes a defective gene present in a parent may be passed down to the offspring. The defective gene may not express itself in the parent. This is because the expression of the defective gene may be masked by its pair, which is normal. But the child may inherit one defective gene from each parent and hence have both genes defective. Such a child suffers from the genetic disorder as shown below.




There are several kinds of hereditary disorders, some of which may be caused due to the presence of only one defective gene or sometimes as shown above by the presence of two defective genes. 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.**

Three common hereditary disorders are Thallasemia, Haemophilia and colour blindness.

**a) Thallasemia**

Patients suffering from this disorder are unable to manufacture haemoglobin, the pigment in red blood corpuscles which carries oxygen to tissues. This is because the pair of genes controlling hemoglobin production are defective. Thallasemics (persons suffering from thallasemia) require frequent blood transfusion in order to survive.

**b) Haemophilia**

Those persons suffering from haemophilia have either a defective gene or lack genes, which control production of substances responsible for blood clotting. In the absence of such substances blood does not coagulate. So, once bleeding starts, it does not stop.

**c) Colour blindness**

Different kinds of colour blindness are met with but commonly those suffering from this genetic disorder are unable to distinguish blue colour from green. Again, this is due to the presence of a defective gene or absence of a gene.

Both haemophilia and colour blindness genes are located on X-chromosomes, and hence, the disorder is passed down form mother to the son. In the mother with two X-chromosomes, the defect may not show up. Also, in the daughter, the effect of defective gene on X-chromosome from mother may be masked by a normal gene on the X chromosome derived from the father. But males have only one X-chromosome, which is inherited from the nother and if it bears the defective gene, the person suffers from the genetic disorder.

| | | | | |
|---|---|---|---|---|
| (1) | $X^C$-chromosome (colour blindness gene in mother's X–chromosome) | + | X-chromosome (normal gene in father's X-chromosome) | $X^CX$ (daughter carrying colour blindness gene but not suffering from it) |
| (2) | $X^C$ (From mother) | + | Y (From father) | $X^CY$ Son born colour blind does not have any gene for colour vision so son with defective gene suffers. |

Thallasemia is an autosomal genetic disorder, while, haemophilia and colour blindness are sex-chromosomal or X-chromosomal disorders.

## CHECK YOUR PROGRESS 30.3

1. What will be the blood group of an individual with genetic combination $I^AI^B$?
2. How can a person be normal for a trait even when carrying one defective gene for that trait.
3. On which chromosome are genes for haemophilia and colour blindness located?
4. On which kind of chromosome–autosome or sex chromosome is defective gene causing Thallasemia located?
5. Name the therapy in which defective gene is substituted by normal gene.

## LET US REVISE

- Passing down of characters from parents to children is called heredity.
- Children of same parents differ because they posses different combinations of parental genes. This is called variation.

- Heredity and variation are due to genes and their varied combinations.
- Study of heredity is called genetics.
- Mendel was the first to postulate laws of inheritance (heredity) and said heredity was due to "factors".
- Sutton explained that "Mandelian factors" were the genes and that genes are present on chromosomes.
- 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. One Chromosome has one molecule of DNA. Genes are fragments of DNA.
- Sex determination in humans is based on combination of sex chromosomes. Females have two X-chromosomes, while males have one X and one Y chromosome.
- Defective genes or absence of genes may cause genetic disorders e.g. thallasemia, haemophilia and colour blindness.
- Thallasemics lack genes responsible for production of haemoglobin so they need frequent blood transfusion for survival.
- Haemophiliacs are bleeders. Their blood cannot coagulate as they lack genes responsible for production of substances required for blood coagulation.
- Colourblind people cannot distinguish blue colour from green due to defective genes for colour vision located on X-chromosomes.
- Thallasemia is an autosomal genetic disorder while, haemophilia and colour blindness are sex chromosomal disorders.

## TERMINAL EXERCISES

### A. Multiple choice type questions.

**Select the correct answer in the following.**

1. Which statement is true for 'genes'?
   (a) Genes are imaginary factors. (b) Genes are fragments of DNA.
   (c) Genes are present in the ribosomes of the cell. (d) Genes are not inherited.
2. In which genetic disorder is the patient unable to manufacture haemoglobin?
   (a) Haemophilia (b) Thallasemia
   (c) Tuberculosis (d) Jaundice
3. The number of chromosomes in a human sperm is,
   (a) 46 (b) 44
   (c) 23 (d) 22
4. The sex chromosomes in females are
   (a) XX (b) one X and no Y
   (c) XY (d) one Y and no X

5. Who can be called the founder or father of genetics?
   (a) Sutton (b) Mendel
   (c) Darwin (d) Bateson

**B. Descriptive type questions.**

1. Name the scientist who gave the laws of inheritance.
2. What are "factors" named by Mendel called today?
3. What is the chemical nature of a gene?
4. Where are genes located?
5. Why is haemophilia called bleeder's disease?
6. State two differences between autosomes and sex chromosomes.
7. Define: heredity; variation; genetic disorder; sex chromosomes.
8. What has been the contribution of Mendel and Sutton to science of genetics? State in one sentence for each of the two scientists.
9. State any two facts about human chromosomes.
10. Why is haemophilia found mostly in boys?
11. With the help of a line diagram explain the chromosomal basis of making of a male child.
12. What is the basis of sex determination in humans?
13. Why is DNA fingerprinting a sure test for identification of a person?
14. Write notes on any one genetic disorder.
15. What is meant by "gene replacement therapy"?
16. Difficult but try
    Rahul's maternal grandfather (mother's father) was colourblind. What are the chances of Rahul being colour blind if his father has normal colour vision?

## ANSWERS TO CHECK YOUR PROGRESS

**30.1**

1. Sutton
2. Genes
3. 22 pairs or 44
4. They determine sex of a person.
5. 46

**30.2**

1. DNA
2. DNA of a person is unique
3. Male
4. (i) one (ii) two
5. One

**30.2**

1. AB
2. The normal gene masks the effect of defective gene.
3. X chromosome
4. autosome
5. Gene replacement therapy

## GLOSSARY

**Autosomes:** Chromosomes containing genes for characters other than those for sex determination.

**Colour blindness:** Genetic disorder in which a person cannot distinguish between blue and green colours due to defective genes.

**DNA:** Deoxyribonucleic acid, chemical substance of which genes are made.

**DNA fingerprinting:** A technique by which a person's identity can be established by the study of his DNA.

**Diploid:** Full (double) set of chromosomes in pairs in a cell.

**Genetics:** Science of heredity and variation.

**Heredity:** Passing down of characteristics from parents to offsprings.

**Haemophilia:** Genetic disorder in which blood does not clot because of the presence of a defective gene.

**Sex chromosomes:** Chromosomes containing genes for sex determination (Designated X and Y).

**Thallasemia:** Genetic disorder in which haemoglobin cannot form in RBCs due to the presence of defective gene.

**Variation:** Genetic differences between individuals.