21

PRINCIPLES OF GENETICS

It is common observation that seeds of mango trees grow into mango plants, dogs give birth to puppies and not into the young ones of any other animal. Humans give birth to human beings. The tendency of offspring to inherit parental characteristics is termed '**heredity**' and the science of heredity in '**Genetics**'. Genetics also seeks to answer questions like why two offspring of same parents look different, why are some people dark and others light skinned etc. In other words, why is there **variation** among individuals of the same kind. This lesson deals with heredity and variation. It also includes a section on hereditary disorders and gives an idea of the human genome too.

OBJECTIVES

After completing this lesson, you will be able to :

- *explain the terms heredity and variation;*
- *describe Mendel's experiments on garden pea and the principles derived;*
- *define the term hybridization, alleles, trait, dominance, recessive, homozygous, heterozygous, genotype, phenotype;*
- *explain the term incomplete dominance with the example of flower colour in 4 o'clock plant (Mirabilis jalapa);*
- *define the terms lethal genes and pleiotropic genes;*
- *explain quantitative (polygenic) inheritance supported by the examples of kernel colour of wheat and skin colour of humans;*
- *differentiate between monogenic and polygenic inheritance;*
- *explain the chromosome theory of heredity;*
- *define and give examples of linkage, crossing over and criss-cross inheritance;*
- *justify mitochondrial inheritance as a case of maternal inheritance;*
- *describe the human karyotype;*
- *differentiate between sex chromosomes and autosomes;*

- *list and describe the causes and symptoms of some common genetic disorders. Colour blindness, haemophilia, Down syndrome, Turner syndrome, Klinefelter syndrome;*
- *describe the inheritance of Rh factor and explain its significance during pregnancy;*
- *explain ABO blood group;*
- *explain the diagnostic technique of aminocentesis and give its significance;*
- *give a brief idea of human genome.*

21.1 HEREDITY AND VARIATION

Whenever an infant is born in a family, the relatives begin to wonder about the resemblance of the infant's eyes, facial features, complexion, colour of hair with those of the parents, siblings and grandparents. The source of such resemblances and differences are in the "genes" that are passed down form parents to children and so on generation after generation. This inheritance of genes is termed '**heredity**' the study of heredity is '**Genetics**'. New individual develop according to the genes inherited from their parents.

> The transmission of characters from one generation to the next, that is from parents to offspring is known as heredity.

It is further observed that siblings from same parents are unique and differ from each other except the identical twins. Such differences are termed **variations**.

> Variation means differences between parents and offspring or between offspring of same parents or between members of the same population.

Variation in a population is very important. It has survival value for the population. This is because if the environment changes, some individuals (variants) may be able to adapt to new situations and save the population from dying out. Variation arises due to **mutation** or sudden change in the genes. Variation also arises because genes get shifted and exchanged during meiosis at formation of gametes, giving rise to new gene combinations (Recall from lesson 8 on cell and cell division about chiasma formation and lesson no. 20 on reproduction in animals for gamete formation and fertilization). At fertilization, there is random mixing of parental chromosomes with different gene combinations. Such a source of variation which is most common is called **recombination**.

> Variation arises because of mutation and recombination.

21.2 MENDEL'S EXPERIMENTS ON THE GARDEN PEA AND PRINCIPLES OF INHERITANCE

Sir Gregor Johann Mendel (1822 to 1884) was Austrian monk who used garden pea (*Pisum sativum*) for his experiments and published his results in 1865. His work, however, was rediscovered in 1900, long after Mendel's death, by Tschermak,

Correns and DeVries. But since Mendel was the first to suggest principles underlying inheritance he is regarded as the founder or **father of genetics**.

21.2.1 Mendel's Experiments

Mendel designed his experiments such that a pure tall variety of pea plants could be crossed to a pure dwarf variety. The anthers from flowers of tall plants were removed and their stigmas dusted with pollen from flowers of dwarf plants. The reverse experiment was also carried out.

In the following spring, seeds from the new plants were collected and sown. He found that all the plants of this generation called **first filial generation** or F_1 grew to be tall plants. He allowed them to self pollinate. Again he collected the seeds. The following year, after the seeds had been sown, he found that three quarters of these plants were tall and the rest dwarf. He repeated the experiment several times and found that the ratio of tall to dwarf plants was 3 : 1 (Fig. 21.1).

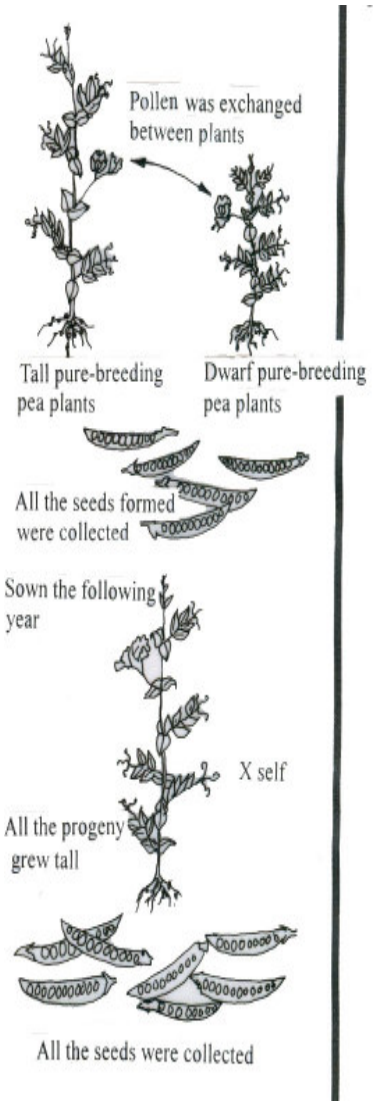


Fig. 21.1 Mendel's experiment with fall and dward peas.

In this way he tried to cross pea plants with seven such contrasting characters. These were 1. red flowered and white flowered plants; 2. axillary flowered (flower arising in the axial of the leaf) and terminal flowered (flower arising at tip of stalk); 3. yellow seeded versus green seeded; 4. round seeded verses wrinkled seeded; 5. green pod versus yellow pod 6. plants with inflated pods versus those with constricted pod and 7. pure tall plants versus pure dwarf plants. Plants with such contrasting characters exist in varieties that are 'self pollinating' so that generation after generation they express only one type of feature (Fig. 21.2).

Crosses considering the inheritance of **one feature only** are called **monohybrid crosses**. Mendel also tried crosses involving two contrasting features, such as tall and red flowered with dwarf and white flowered plant such a cross is termed **dihybrid cross**.

21.2.2 Mendel's Principles (laws) of inheritance

Basing on the results of his experiments, Mendel postulated the following laws of heredity.

1. **Law of segregation or purity of gametes.** At formation of gametes, the two chromosomes of each pair separate (segregate) into two different cell which form the gametes. This is a universal law and always during gamete formation in all sexually reproducing organisms, the two factors of a pair pass into different gametes. Each gamete receives one member of a pair of factors and the gametes are pure.

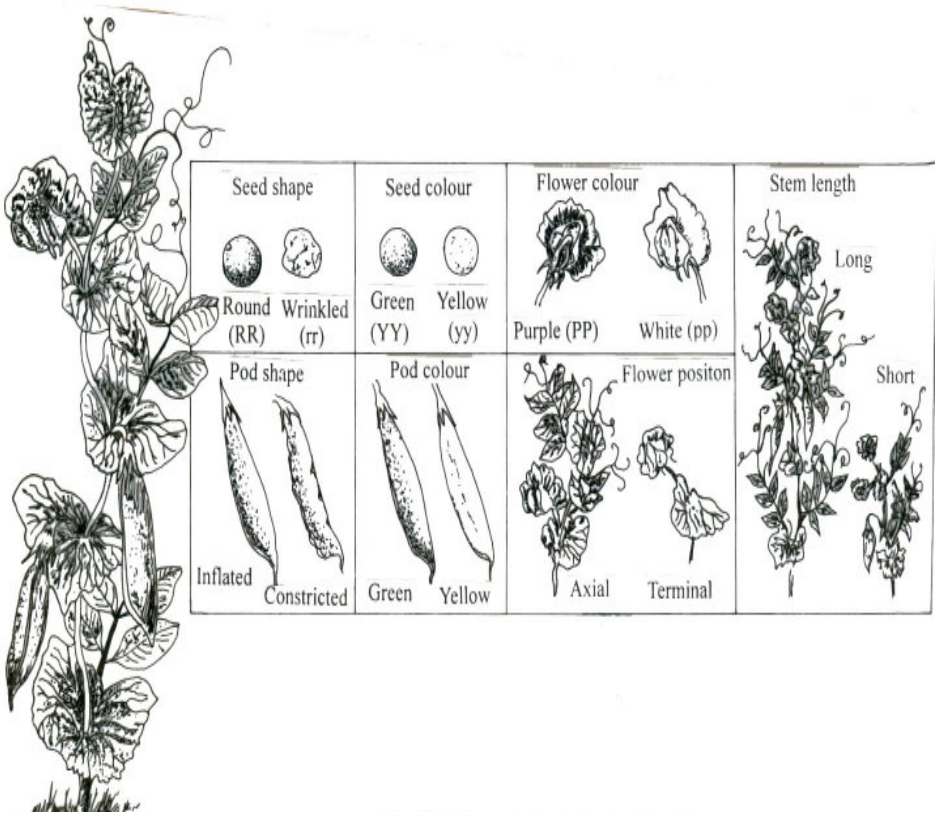


Fig. 21.2 Seven traits studies by Mendel

2. **Law of dominance.** Inheritance of many features (e.g. eye colour, flower colour, seed shape etc.) is controlled by **one pair of genes.** When the two genes are of the same kind (e.g. brown colour of eyes, red colour of flower) the condition is termed **homozygous**. When a pair of chromosomes has the gene controlling the same feature (flower colour) in two different forms (red flower gene on one chromosome and white flower gene on its pair (termed its **homologue,**) the condition is termed **heterozygous.**

   The second law of inheritance maintains that of the two contrasting genes of a pair, **the expression of one is dominant over that of the other**. Thus if both genes are for tallness (represented on TT) that is **homozygous** or for tallness and one for dwarfness (Tt), the pea plants will be tall. The opposite of dominant gene is termed **recessive gene**. The recessive feature (e.g. dwarfness of the plant) shows up only when its gene is in the homozygous condition (tt).

The law of dominance was found to be true in both monohybrid and dihybrid crosses in cases of the characteristics studied by Mendel in the garden pea.

3. **Law of independent assortment** meaning whereby that in the inheritance of two features (each feature controlled by a pair of genes), genes for the two different features are passed down into the offspring **independently** (Fig. 21.3) i.e. the segregation of one pair of factors is independent of the segregation of the other.

	Red Tall		White Dwarf	
Parents	TTRR	×	ttrr	
gametes	TR		tr	
F_1	Tt		Rr	Tall, red (self)

F_2 progeny worked out below.

Genes in male and female gametes	TR	Tr	tR	tr
TR	TTRR Tall red	TTRr Tall red	TtRR Tall red	TtRr Tall red
Tr	TTRr Tall red	TTrr Tall white	TtRr Tall red	Ttrr Tall white
tR	TtRr Tall red	TtRr Tall red	ttRR White red	ttRr White red
tr	TtRr Tall red	TtRr Tall red	ttRr Dwarf red	ttrr Dwarf white

Fig. 21.3 Dihybrid phenotypic ratio

9 Tall Red : 3 Tall Dwarf : 3 Dwarf red : 1 Dwarf white

Fig. 21.3 Results showing independent assortment in two pairs of genes. R stands for red flower colour, r for white flower colour, T for tall gene and t for gene for dwarfness.

You would have noticed that the composition of genes or **genotype** controls the outside expression which we can be see, that is the phenotype. The ratio of progeny in the crosses is therefore, the **phenotypic ratio**.

However, as more and more scientists began to devise genetic experiments, it became clear that Mendel's laws do not hold true in all cases. We shall learn about the deviations from Mendel's laws such as incomplete dominance, codominance etc.

21.2.3 Reasons for Mendel's success

1. **His choice of material** was good. He selected garden pea which has a short life cycle, has self pollinated bisexual flowers with closed corolla so pollination

can be easily controlled. Also it is easy to cultivate pea plants and a large number of pure line plants with several pairs of contrasting characters were available.

2. **His selection of traits :** The seven pairs of contrasting characters of pea plant considered by Mendel in his experiments were responsible for the desired results that helped Mendel postulate the laws of inheritance.

3. **Mendel's Technique :** His technique of experimentation was excellent and was as follows :

   (i) Homozygous pure line plants with contrasting characters were crossed.

   (ii) Self pollination was prevented by removing stamens.

   (iii) Female plants were dusted with pollen grains from another plant with the opposite feature and were tied in a bag to prevent any further pollination.

   (iv) Seeds were collected and shown in time.

   (v) He considered the inheritance of one character at a time.

   (vi) The results were analysed statistically.

   (vii) He performed reciprocal crosses and test crosses to confirm the results. (see section 21.3 for definition of these crosses)

21.3 IMPORTANT TERMS IN GENETICS

- **Factor :** The unit responsible for the inheritance and expression of a particular character. Now replaced by the term gene.

- **Gene :** It is a particular segment of a DNA molecule which determines the inheritance and expression of a particular character.

- **Alleles or Allelomorphs :** Two or more alternative firms of a factor or a gene are called alleles. For example in pea plant, the gene for producing seed shape may occur in two alternative forms: round (R) and wrinkled (r). Genes for round and wrinkled seeds are alleles of each other. Similarly, there are three alleles for gene controlling blood group in man I^A, I^B, i (I = immunoglobulin gene). Alleles occupy same locus on homologous chromosomes.

- **Trait :** is the expressed character, e.g. colour of flower, shape of seed etc.

- **Dominant trait :** Out of the two alternating forms (allelomorphs) of a trait, the one which expresses itself in a heterogygous organism in the F_1 hybrid is called the dominant trait (dominant allele) and the phenomenon is called dominance. For example in an organism with Tt, T (tallness) expresses itself and t (dwarfness) cannot, so T is the dominant allele.

- **Recessive trait :** Out of the two alternating alleles for a trait, the one which is suppressed (does not express) in the F_1 hybrid is called the recessive trait

(recessive allele). Recessive allele expresses only in the homozygous state (e.g. tt).

- **Genotype :** The genetic constitution of an individual (which he/she inherits from the parents) is called the genotype, e.g., the genotype of pure round seeded parent pea plant is RR.
- **Phenotype :** The outward (morphological) appearance of an individual for any trait or traits is called the phenotype, e.g. for seeds, round shape or wrinkled shape is the phenotype.
- **Homozygous :** an individual possessing identical alleles for a trait is called homozygous or pure for that trait, e.g. plant with RR alleles is homozygous for the seed shape.
- **Heterozygous :** An individual receiving dissimilar alleles for a trait is called (heterozygous or impure for that trait, e.g. a plant with Rr alleles is heterozygous for the seed shape.
- **Parent generations :** The parents used for the first cross represent the parent (or P_1) generation.
- **F_1 generation :** The progeny produced from a cross between two parents (P_1) is called **First filial** or **F_1 generation**.
- **F_2 generation :** The progeny resulting from self hybridization or inbreeding of F_1 individuals is called **Second Filial** or F_2 generation.
- **Monohybrid cross :** The cross between two parents differing in a single pair of contrasting characters is called monohybrid cross and the F_1 offspring as the **hybrid**. The phenotypic ratio of 3 dominants : 1 recessive obtained in the F_2 generation from the monohybrid cross is called **monohybrid phenotype ratio** (eg. 3 : 1 in Mendelian Inheritance).
- **Dihybrid cross :** The cross between two parents in which two pairs of contrasting characters are studied simultaneously for the inheritance pattern is called dihybrid cross. The phenotypic ratio obtained in the F_2 generation from a dihybrid cross is called dihybrid ratio phenotypic (e.g. 9 : 3 : 3 : 1 in Mendelian crosses.
- **Hybridisation :** Crossing organisms belonging to different species for getting favourable qualities in the offspring.
- **Test cross :** Crossing of the F_1 progeny with the homozygous recessive parent. If F_1 progeny is heterozygous, then test cross always yields the ratio of 1 : 1.
- **Reciprocal cross :** Is the cross in which the sex of the parents is reversed. That is if in the first cross father was dwarf and mother tall, then in the reciprocal cross, dwarf parent will be female and tall parent male.

INTEXT QUESTIONS 21.1

1. Who is called the founder of genetics and why?

   .....

2. State one difference between

   (i) homozygous and heterozygous

   .....

   (ii) dominant and recessive

   .....

   (iii) genotype and phenotype

   .....

   (iv) monohybrid and dihybrid crosses.

   .....

3. Define heredity and variation.

   .....

4. Give the monohybrid and dihybrid phenotypic ratios.

   .....

5. Mention two sources of variation.

   .....

21.4 DEVIATION FROM MENDEL'S LAWS INCOMPLETE DOMINANCE

In the four O'clock plant *Mirabilis jalapa* and Snapdragon or *Antirrhinum* law of dominance is not followed. Thus when a homozygous red flowered plant (RR) is crossed to a homozygous white flowered plant, all flowers in the F_1 are pink when F_1 plants are self pollinated, the phenotypic ratio is found to be 1 : 2 : 1.

Parents	RR × rr
Gametes	R, R × r, r
F_1	Rr Pink
F_2	1 Red : 2 Pink : 1 White

You will find that the heterozygous (Rr) plants have an intermediate colour pink. You must have also noticed that the genotypic ratio 1 RR : 2 Rr : 1 rr and phenotypic ratio 1 Red : 2 Pink : 1 white are both (1 : 2 : 1) same.

Multiple alleles and codominance

Tall or dwarf, red or white, brown eyes or blue eyes, these are just two varieties or **alleles** of genes for plant height and flower colour and eye colour. Most genes, however, have more than two alleles or **multiple alleles**. An example of multiple alleles is the gene for blood group.

The four blood groups of humans are determined by a single gene. The alleles I^A for A group, I^B for B blood group are both dominant. Therefore person with alleles I^A and I^B has the blood group AB as both the genes I^A and I^B are **co-dominant**. The gene i when homozygous (ii) gives the blood group O. Genotype and phenotype of blood groups in humans are given in Table 21.1.

Table 21.1 Genotypes and Phenotypes of human blood groups

Genotype	Blood group
I^AI^A and I^A i	A
I^B I^B and I^B i	B
I^A I^B	AB
ii	O

Lethal genes

Have you ever seen a yellow mouse? Probably not. The yellow coat colour in mice is due to the presence of the gene which is also responsible for killing the mouse born with that gene. Such genes are termed **lethal genes**. Some lethal genes kill only in the homozygous condition and are **recessive lethals. Dominant lethals** cause death even when present in the heterozygous condition.

Pleiotropy

While a gene may have multiple alleles and thus give multiple genotypes, one gene may control several genotypes. For example the recessive gene for white eye in *Drosophila* when present in the homozygous condition affects several other features such as wing shape and shape of abdomen. Thus a white eyed *Drosophila* is also born with vestigeal wings and curled abdomen.

Polygenic or quantitative inheritance

When a trait (feature or character) is controlled by a single gene it is termed **monogenic inheritance**. Many traits or features are controlled by a number of different genes. For example the skin colour of humans and the kernel colour of wheat results from the combined effect of several genes, none of which are singly dominant. Polygenes affecting a particular trait are found on many chromosomes. Each of these genes has equal contribution and cumulative the total effect. Three to four genes contribute towards formation of the pigment in the skin of humans. So there is a continuous variation in skin colour from very fair to very dark. Such inheritance controlled by many genes is termed **quantitative inheritance** or **polygenic** (poly meaning due many genes) **inheritance**.

$R_1R_1R_2R_2$ (Red) × $r_1r_1r_2r_2$ (White)

Pink

$R_1r_1R_2r_2$

	R_1R_2	R_1r_2	r_1R_2	r_1r_2
R_1R_2	$R_1R_1R_2R_2$ Red	$R_1R_1R_2r_2$ Dark Pink	$R_1r_1R_2R_2$ Dark Pink	$R_1r_1R_2r_2$ Pink
R_1r_2	$R_1R_1R_2r_2$ Dark Pink	$R_1R_1r_2r_2$ Pink	$R_1r_1R_2r_2$ Pink	$R_1r_1r_2r_2$ Light Pink
r_1R_2	$R_1r_1R_2R_2$ Dark Pink	$R_1r_1R_2R_2$ Pink	$r_1r_1R_2R_2$ Pink	$r_1r_1R_2r_2$ Light Pink
r_1r_2	$R_1r_1R_2r_2$ Pink	$R_1r_1r_2r_2$ Link Pink	$r_1r_1R_2r_2$ Light Pink	$r_1r_1r_2r_2$ White

1 Red : 4 Dark Pink : 6 Pink : 1 White

In polygenic inherlance:

(i) Each dominant gene controls equally the intensity of the character.

(ii) The effect of the dominant genes in cumulative.

(iii) The intensity of character or trait depend upon the number of dominant genes.

INTEXT QUESTIONS 21.2

1. Define :

   (i) An allele .....

   (ii) Codominance .....

   (iii) Polygenes .....

   (iv) Lethal gene .....

2. Which kind of inheritance is found for

   (i) blood groups of humans .....

   .....

   (ii) flower colour of snapdragons .....

   .....

(iii) wheat kernel colour .....

.....

(iv) human skin colour? .....

.....

3. State the phenotypic monohybrid ratio in case of incomplete dominance.

.....

21.4 CHROMOSOMAL THEORY OF INHERITANCE

It was observed by **Sutton and Boveri** in **1902** that

1. Maternal (from mother) and paternal (from father) characters come together in the progeny which is diploid or 2n and has chromosomes in pairs and later on segregate during the formation of gametes. The gametes have a single chromosome from each pair and is haploid or n.
2. Chromosomes from two parents come together in the same zygote as a result of the fusion of two gametes and again separate out during the formation of gametes. You have already learnt that charomosomes are filamentous bodies present in the nucleus and seen only during cell division.

The above two observations proved that there is a remarkable similarity between the behaviour of characters during inheritance and that of chromosomes during meiosis.

This led Sutton and Boveri to propose '**chromosomal theory of inheritance**' and its salient features are as follows.

1. The somatic (body) cells of an organism, which are derived by the repeated division of zygote have **two identical sets of chromosomes** i.e. they are **diploid.** Out of these, one set of chromosomes is received from the mother (maternal chromosomes) and one set from the father (paternal chromosomes). Two chromosomes of one type (carrying same genes) constitute a **homologous pair**. Humans have 23 pairs of chromosomes.
2. The chromosomes of homologous pair separate out during meiosis at the time of gamete formation.
3. The behaviour of chromosomes during meiosis indicates that Medelian factors or **genes are located linearly on the chromosomes.** With progress in molecular biology it is now known that a chromosome is made of a molecule of DNA and segments of DNA are the genes.

21.5 LINKAGE AND CROSSING OVER

Bateson and Punnett performed a dihybrid cross with true breeding varieties of sweet pea (*Lathyns sativus*) and instead of 9 : 3 : 3 : 1 ratio in F_2 they got the ratio 7 : 1 :1 : 7. So the parental forms were more than the new combinations of characters (recombinants). It means the two genes chosen for the experiment do not follow the principle of independent assortment. Instead they tend to be inherited together or are **linked** together. Thus genes present on same chromosome tend to be inherited together and are said to be **linked**. This phenomenon is called **linkage**.

A group of genes present on the same chromosome and tend to be inherited together forms a **linkage group**.

In the above experiment some recombinant types were also produced. How did that happen ? They are produced by another phenomenon called **crossing over**.

Crossing over is the physical exchange of parts of the non sister chromatids of chromosomes of homologous pair (Fig. 21.4).

Crossing over occurs during meiosis I prophase I at the time of gamete formation. The point where crossing over occurs is called **chiasma**. (plural : chiasmata) See Fig. 21.3. Linked genes get separated from each other by crossing over.

Because of linkage and crossing over a heterozygous individual can produce four types of gametes as shown in Fig. 21.4. The figure 21.4 shows linked genes of the parents and **recombinants** due to crossing over.

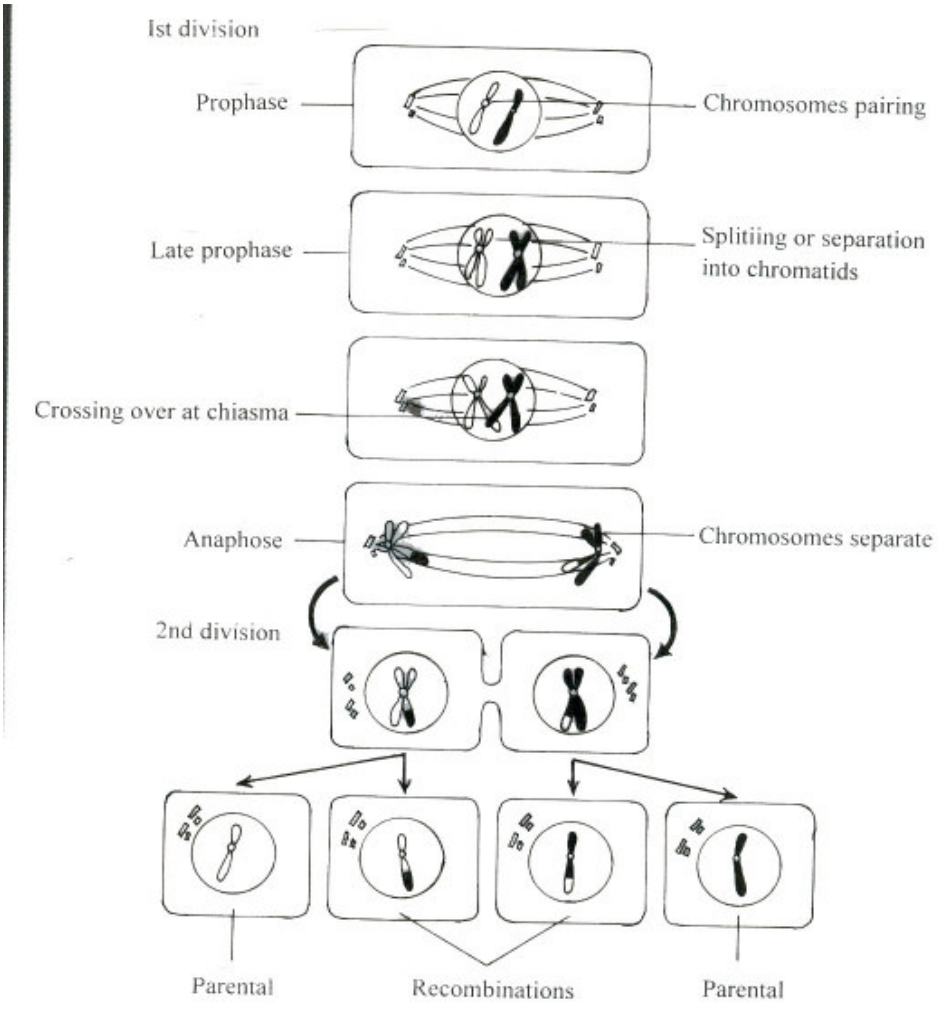


Fig. 21.4 Schematic diagram showing recombination by crossing over

Fig. 21.4 Schematic diagram showing recombination by crossing over

21.6 CHROMOSOMES AND SEX DETERMINATION

In some diploid organisms, specific chromosomes have a role in sex determination. Such chromosomes some are called **sex chromosomes** and the rest of the chromosomes of a set are called **autosomes**.

- If sex chromosomes are morphologically similar (i.e. XX) in an individual, the individual is termed **homogametic**. Such individuals, produce only one kind of gamete (containing X). For example : all eggs of the human female contain an X chromosome. So human female is termed homogametic.

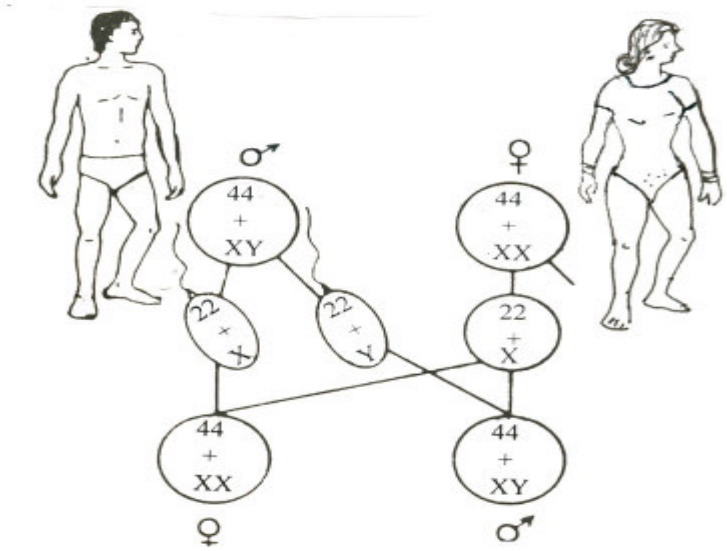


Fig. 21.5 Chromosomal basis of sex determination in humans.

- If sex chromosomes are morphologically dissimilar (i.e. XY) in an individual, is termed **heterogametic**. Such individuals produce two types of gametes. (one containing X and the other containing Y). For example : human male produces two kinds of sperms, X bearing and Y bearing sperms. When the human egg is fertilised by an X bearing sperm a boy is born (Fig. 21.6). Whether the unborn will be a male or female is purely a matter of chance and no parent can be blamed for the sex of the progeny.

21.7 CRIS-CROSS INHERITANCE (X-LINKED INHERITANCE

We already know that genes are located on chromosomes. The genes which are located on X chromosome (sex chromosome), are called sex linked genes. These genes show cris-cross inheritance as shown in Fig. 21.6.

When a male has a defective sex linked gene located on **X** chromosome during reproduction, he transmits the defective **X** chromosome to his **daughter only**. The female who has this gene transmits it to her sons and daughter both in equal probability. So the male passes on his recessive sex linked trait to 50% of his grandsons through his daughter. The sex linked trait being recessive is not expressed in female but is expressed in males. Therefore males suffer from genetic defect

while females are only **carriers** of these defective genes. Females express the trait if they are homozygous for the recessive gene.

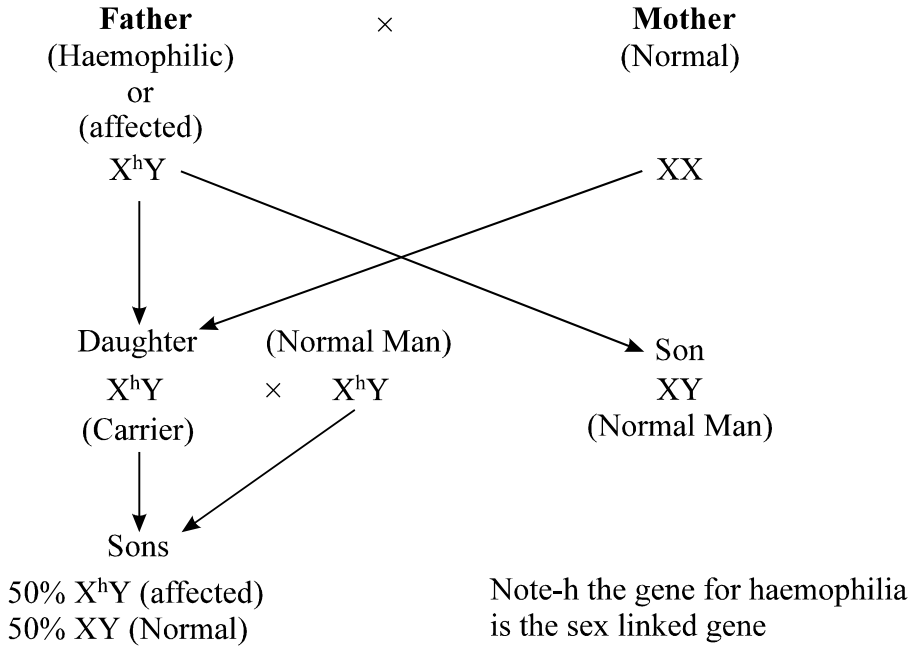


Fig. 21.6 Cris cross inheritance or X-linked inheritance

This type of inheritance of recessive sex linked character from father to daughter and then from the daughter to her sons is known as cris cross inheritance or sex linked or X-linked inheritance.

Cris Cross Inheritance in humans : Red green colour blindness and Haemophilia are examples of sex linked inheritance in humans. The defective gene is located on **X** chromosome. Thus a single defective gene causes disease in male while two defective genes (homozygous condition) only can cause the disease in female. Females in heterozygous condition are apparently normal but actually the carriers of the disease. Carrier females pass this defective gene to 50% of her sons. The disease is expressed only in males because male does not have the partners of the genes on **Y** Chromosome. See Fig. 21.7 (a), (b) and (c).

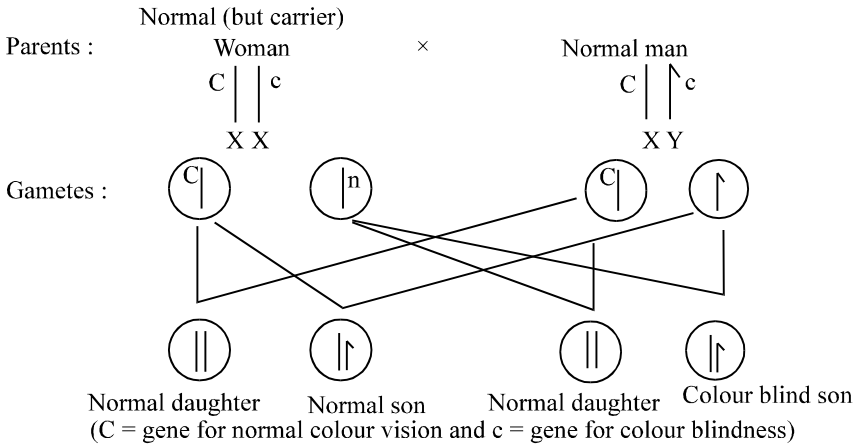


Fig. 21.7 (a) Progeny of carrier female for colour blindness and a man with normal colour vision.

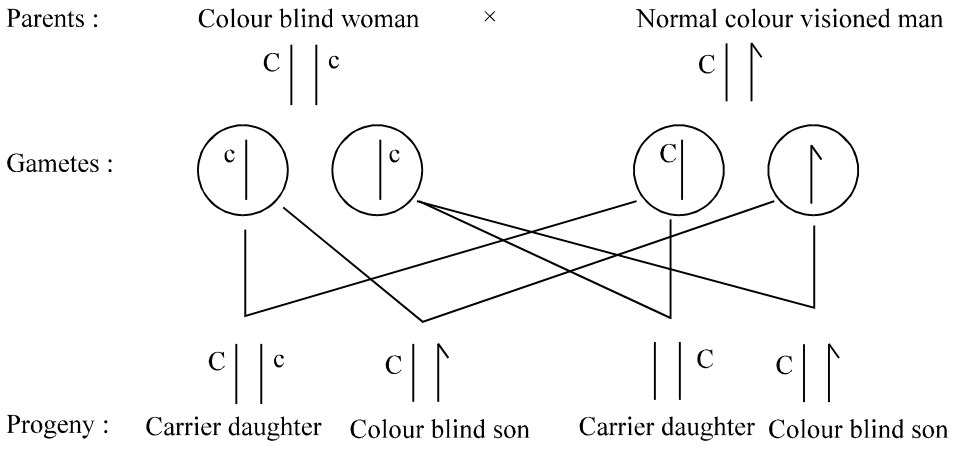


Fig. 21.7 (b) Progeny of colour blind female and a man with normal vision.

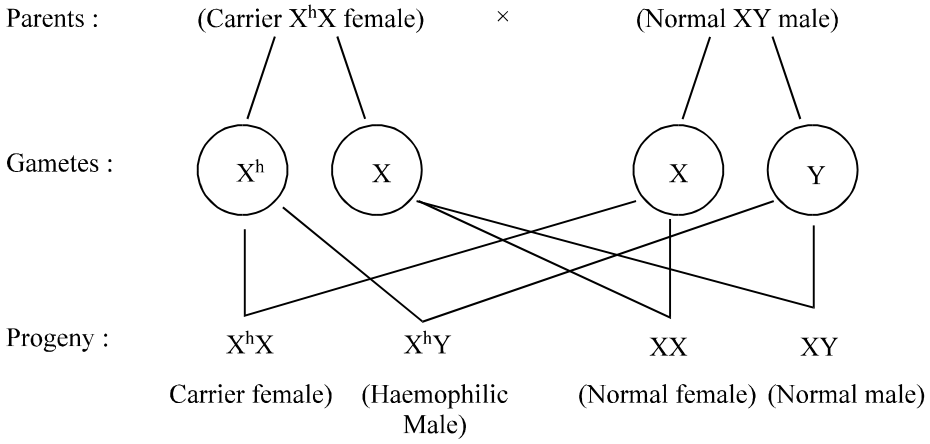


Fig. 21.7 (c) Progeny of an haemophilic carrier female and normal male

INTEXT QUESTIONS 21.3

1. What are genes and where are they located?

   .....

2. State the names of the scientists who proposed the chromosomal theory of inheritance.

   .....

3. Define (i) linkage and (ii) crossing over.

   .....

4. When does gene exchange through chiasma formation occur between homologous chromosomes?

   .....

5. Why is the human female called the homogametic sex?

   .....

6. A colour blind man married to a normal woman whose father and mother both had normal colour vision. Will any of their sons be colour blind? If not why not.

   .....

21.8 MITOCHONDRIAL INHERITANCE AS A CASE OF MATERNAL INHERITANCE

Apart from the nucleus, mitochondria and chloroplasts also possess DNA and you have just learnt that genes are segments of DNA. Till now you have studied about genes on the chromosomes present in the nucleus. Since mitochondria come into the zygote from the egg, inheritance of mitochondrial DNA or DNA is said to be a case of maternal inheritance.

In fact, certain diseases and therefore the genes responsible for them are due to defects in mitochondrial DNA and can be traced to the mother's family.

21.9 HUMAN KARYOTYPE

Human karyotype is the arrangement of chromosomes in seven groups according to the types of chromosomes and their size. It is prepared by arranging chromosomes seen at mitotic metaphase.

(i) Total no. of chromosomes or 2n = 46 (23 pairs).

(ii) Number of autosomes = 44 (22 pairs).

(iii) Sex chromosomes = X and Y = 2

(iv) Depending on size, location of centromere, and bands obtained by special staining methods, human chromosomes are grouped into 7 groups A to G as shown in Fig. 21.8.

Sex determination in humans, as you have already learnt is as follows :

Normal male 22 pairs of autosomes + one X chromosome and one Y chromosome

Normal female 22 pairs of autosomes + two X chromosome

Presence of Y is necessary for maleness.

Absence of Y chromosomes makes the individual a female

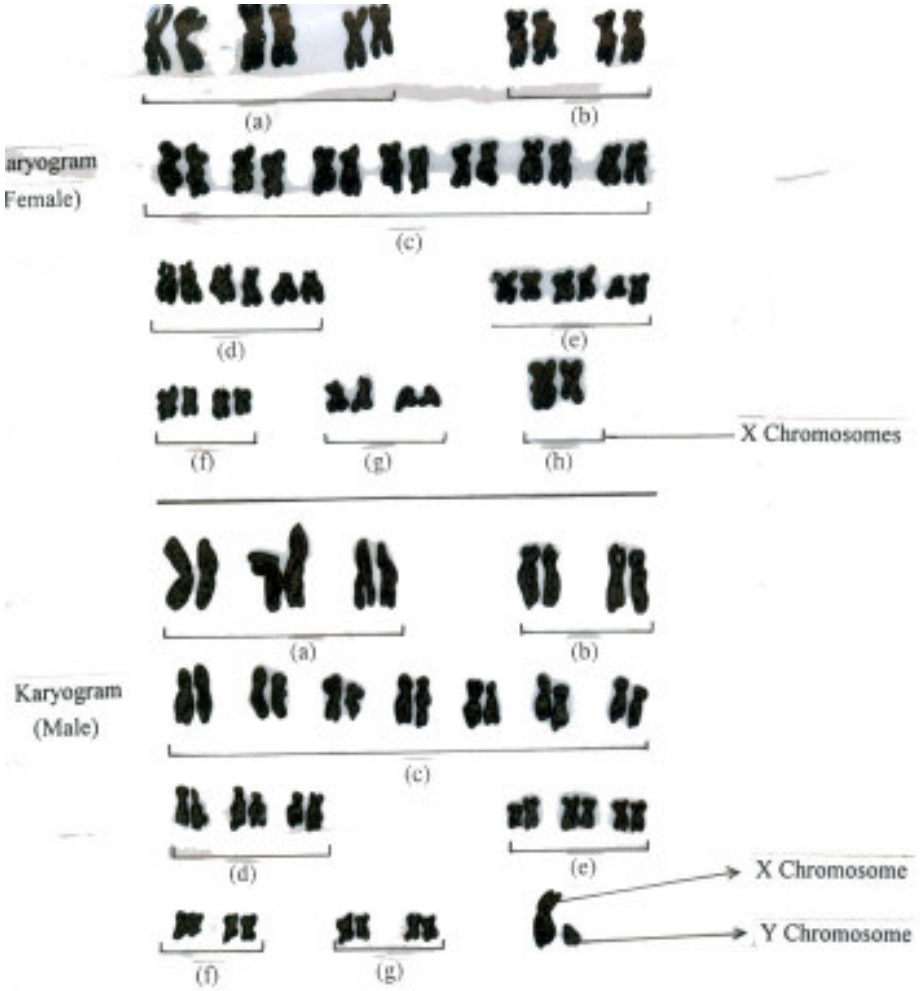


Fig. 21.8 Karyotype showing chromosomes of normal male. Female has the same autosomes but two X-chromosomes instead of XY

21.10 CHROMOSOMAL ABNORMALITIES AND GENETIC DISORDERS IN HUMANS

Any change from the normal number or structure of chromosomes causes abnormalities. Following are some of them.

1. Mongolism or Down's syndrome

The individual has 47 chromosome because of one extra chromosome in the 21st pair (Trisomy of chromosome 21). The outcome is that they are

- mentally retarded
- have a thick tongue
- and a drooping (false expression of pleasure) face. Fig. 21.9.

The possibility of giving birth to a mongol child is far greater in pregnant mothers above the age of forty.

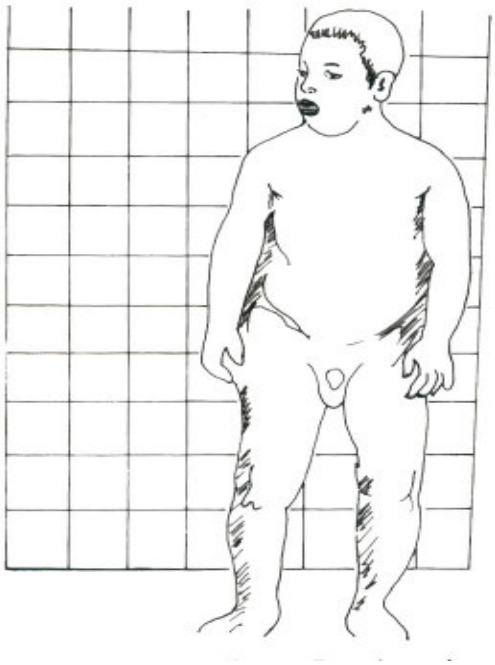


Fig. 21.9 Mongolism or Down's syndrome

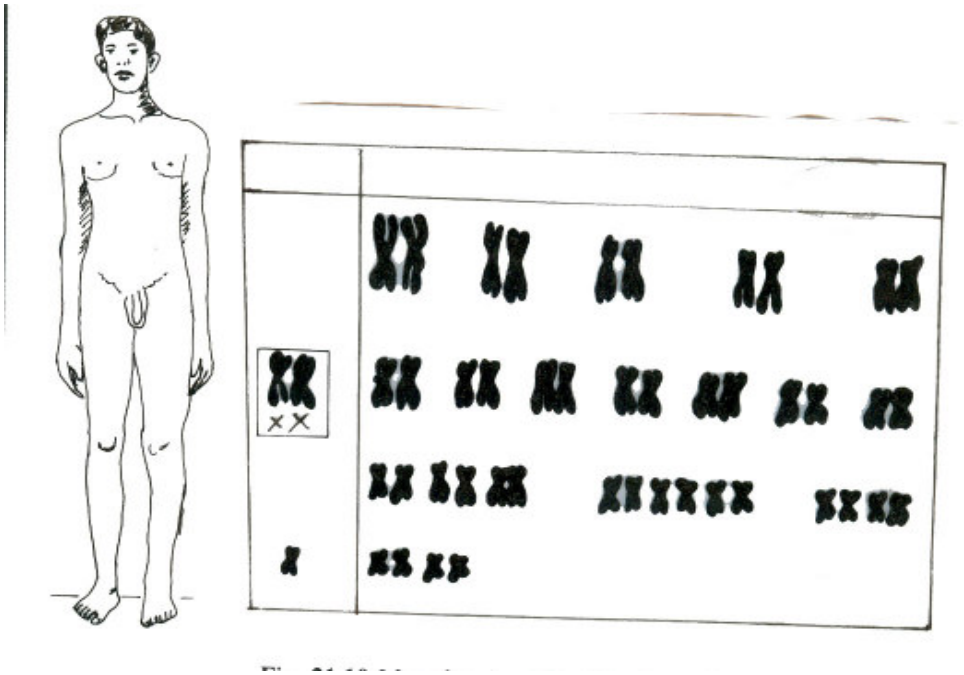


Fig. 21.10 Man showing Klinefelter's syndrome

2. Klinefelter's syndrome

Individual is a male with 47 chromosome with one extra X chromosome. (44 autosomes + XXY). Typical features of a Klinefelter are

- Tall, mentally retarded male;
- Sterile and shows breast development or **gynaecomastia** (gynae : female; massere : mammary glands). Fig. 21.10.

3. Turner's syndrome

Individual is a female with 45 chromosomes with only one X, chromosome (22 pairs of autosomes +XO). The characteristic features are

- Mentally retarded
- web like skin on neck.
- incompletely developed breasts. Fig. 21.11.

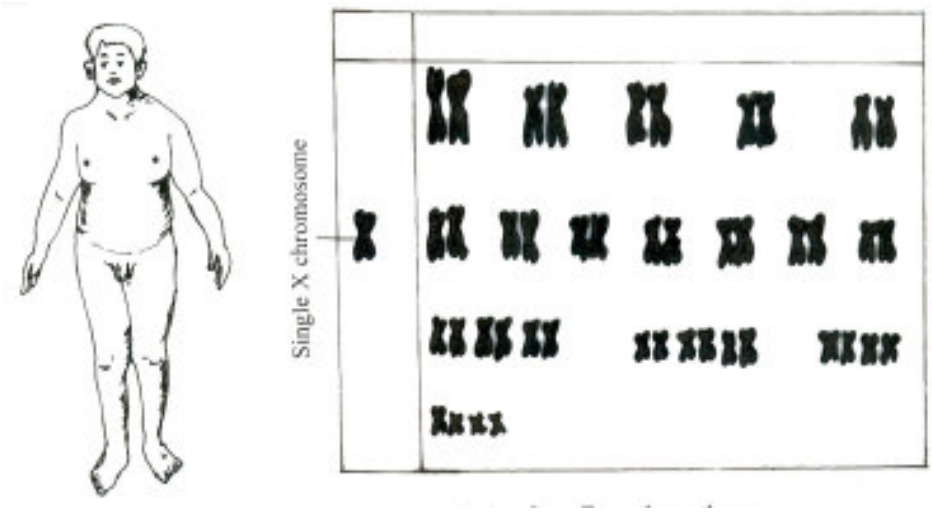
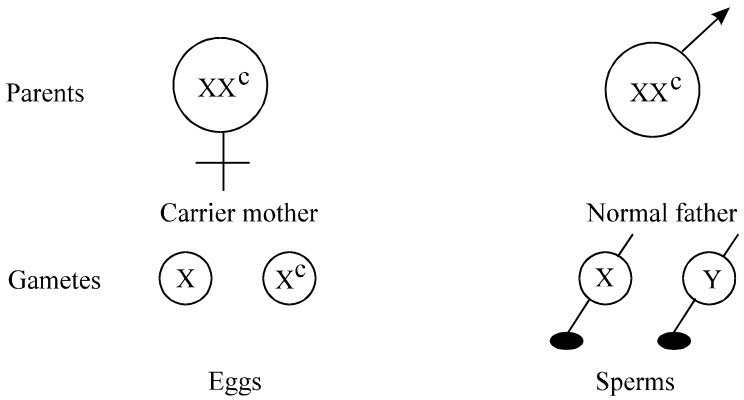


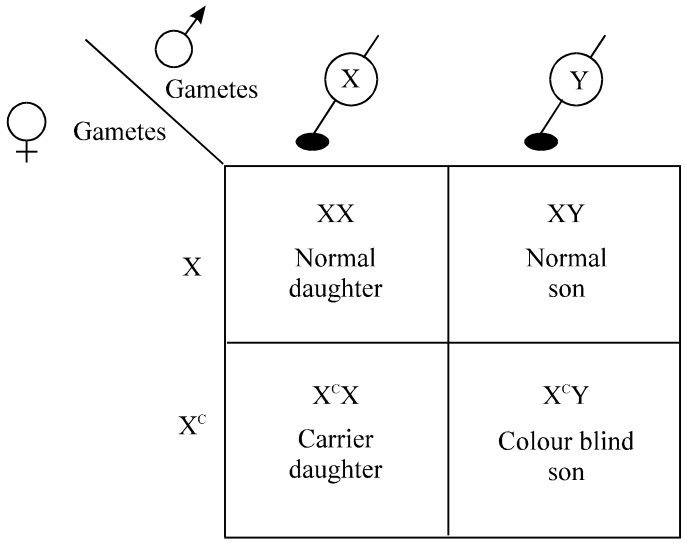
Fig. 21.11 A woman suffering from Turner's syndrome.

4. Colour blindness and Haemophilia (Bleeder's diseases)

Both these are sex linked disorders. (See figures 21.6 and 21.7)



The inheritance is as follows :



Note : X = *normal allele*; X^C = *recessive mutant*

Fig. 21.12 Inheritance of colour blindness

In male, the single X-chromosome is received from the mother.

Hence a defective, gene (for colour blindness or haemophilia) on X chromosome of the mother, is passed on to the son and expressed as a defect.

The daughter receives one X-chromosome from the mother and the other X from the father. The defective gene received from the mother may be masked by normal allele on the other X. The daughter is then called a carrier. (Fig. 21.12).

Colour blind males are unable to distinguish between red-green colours. In haemophilia afflicted male, blood does not clot easily and the patient may bleed to death. Its mode of inheritance is exactly like that of colour blindness.

5. Thalassemia

It is a disorder in which haemoglobin is not synthesised properly. So, frequent blood transfusions are required for survival.

The defective gene is recessive and therefore heterozygous parents may not show the disorder. The child who gets the defective genes 'from both the parents (homozygous recessive) suffers from Thalassemia.

6. Sickle Cell Anemia

This is another hereditary abnormality due to mutation of a single gene in which. red blood corpuscles lose their shape and become sickle shaped because of defective Haemoglobin. Individuals possessing two defective genes (homozygous recessive), cannot survive. In the heterozygous individuals, one gene is normal and so half the number of total red blood corpuscles are normal containing normal haemoglobin while the others are defective. A boon in disguise against malaria for children with one defective Haemoglobin gene can survive as they are less affected by malaria because the malaria parasite cannot thrive inside the defective RBC's.

7. Rh factor

Rh factor is an antigen (a protein) present on the surface of red blood corpuscles. About 15% of all women do not have the gene for Rh antigen. They are Rh-negative. Men can also be Rh-negative, but the problem which this trait creates is in Rh-negative women.

A pregnant Rh-negative women whose husband: is Rh + may bear a child who may have inherited the Rh + gene from the father. If the foetal blood enters mother's body stream, her immune system produces antibodies against Rh antigen of the embryo. Rh antigens cause no problems in first pregnancy. But in a subsequent early pregnancy, the mother's antibodies against Rh antigen may enter the foetal blood stream and destroy its red blood corpuscles causing severe anemia which may even be fatal. (erythroblastosis foetalis)

Now a days Rh-negative mother of a Rh-positive foetus is treated immediately after delivery, to destroy Rh antigens in her blood stream. (Fig. 21.13)

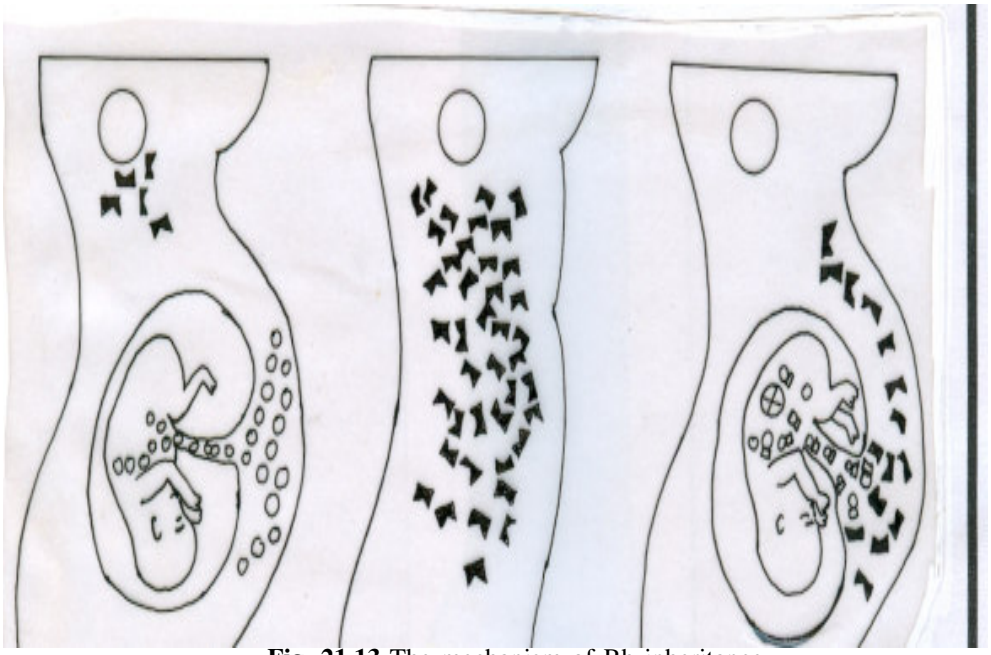


Fig. 21.13 The mechanism of Rh inheritance

(a) shows the first pregnancy where the mother is Rh (-) and foetus Rh (+). Protein (empty circles) of the foetus stimulates the production of anti factors (black blocks) in the mother.

(b) shows the retention of anti factors in the mother's body.

(c) shows the Rh (+) foetus in the womb of the same mother during the second pregnancy. The anti factors from the same mother during the second pregnancy. The anti factors from the mother's body will destroy the infant's red blood cells.

21.11 AMNIOCENTESIS

Amniocentesis in a technique by which hereditary disorders due to defects in genes can be detected. In this technique (Fig. 21.14)

(i) a small sample of amniotic fluid which surrounds the foetus is syringed out.

(ii) This fluid has cells which break off from the skin of the foetus.

(iii) Foetal cells are picked up and cultured.

(iv) Chromosomes in the dividing cells are analysed for genetic defects.

If incurable genetic defects are detected, pregnancy can be terminated.

It is illegal to use amniocentesis for detecting the sex of the unborn.

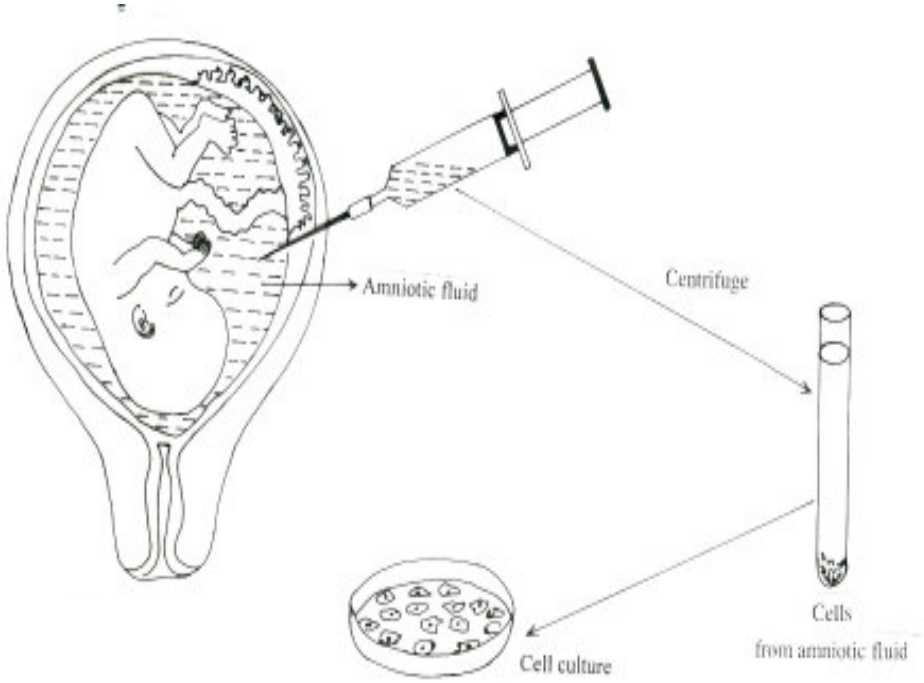


Fig. 21.14 Techniques of Aminocentesis

21.12 THE HUMAN GENOME

In the last over hundred years genetics and molecular biology have gene far ahead and the progress has been very rapid.

You have just read about genetic disorders and today there is hope for heir cure through gene therapy. This is because in 2003, most of the genes on human chromosomes in have been mapped or located on the 23(n) chromosomes. The genes responsible for inheritance of various structural features, genes that control various enzymes that catalyse the various biochemical reactions in the body, genes responsible for genetic disorders have been located. Genome means genes of a particular organism. Since genes are present in pairs (one inherited from mother and other from the father), all kinds of genes present in a particular type of organism are present on its haploid set (n). Thus human genome may be defined as all the genes present in the haploid set of chromosomes in humans. There are an estimated

20,000 to 25,000 genes and 3 billion base pairs in the total human DNA. Each human chromosome has apart from protein coding genes regulating base sequences, non coding DNA, promoter sequences (TATA boy) in between genes that code for proteins. There are genes that code for the production of ribosomal RNA and the many tRNAs.

It is estimated that only 1.5% of the human genome has protein coding sequences and 20,000 to 25,000 genes.

INTEXT QUESTIONS 21.4

1. Why is mitochondrial inheritance treated as a case of maternal inheritance?

   .....

2. Into how many groups have human chromosomes been grouped in the human karyotype?

   .....

3. State the chromosomal abnormality in Klinefelters, Turners and in Mongolism.

   .....

WHAT YOU HAVE LEARNT

- Heredity means the transmission of characters from parents to offspring.
- Variation pertains to difference between siblings or members of same species.
- Mendel was the first to explain that heredity involves transmission of units from reproductive cells of parents to offspring.
- Hugo de Vries, Correns and Tschermak rediscovered the Mendel's Laws of inheritance.
- Mendel selected seven varieties of garden pea differing in seven pairs of characters.
- According to his 'law of segregation' the factors segregate at the time of gamete formation.
- Mendel's 'law of dominance' states that one factor dominates the other contrasting factor in phenotypic expression.
- Law of independent assortment states that the inheritance of one character does not depend on inheritance of any other character.
- There are deviations from Mendelian inheritance and these patterns of inheritance are incomplete dominance, codominanace, multiple alleles, polygenetic inheritance and pleiotropy.

- Sutton and Boveri (1902) proposed the chromosomal theory of heredity. It states that Mendelian factors or genes are located on chromosomes.
- Gene located on chromosomes in a linear fashion and are held together in linkage group. Linked genes get segregated through chiasma formation or crossing over.
- Organisms with separate sexes have a pair of sex chromosomes called sex chromosomes in humans XX for female and XY for male.
- Males inherit an X chromosome from female parent and Y from the male parents. Y chromosome bears genes for maleness.
- Female receive two chromosomes one each from either of the two parents.
- Any change from normal number & structure of chromosomes causes abnormalities.
- A normal karyotype shows the 23 pairs of humans chromosomes and thousands genes.
- Down's syndrome patients have 47 chromosomes (tri-somy of chromosome 21).
- Klinefelter's syndrome patient has 44 autosomes and XXY.
- Turner's syndrome, has 44 autosomes + XO
- Colour blindness and Haemophilia are X-linked disorders.
- Thallesemia and Sickle cell anaemia are due to a single defective gene.
- Rh +ve foetus in a Rh negative mother poses problems in which antibodies are produced in mother's blood against antigens of the foetus.
- The human genome has been mapped.
- Amniocentesis a technique for detecting genetic disorder in foetus.

TERMINAL EXERCISES

1. State the three Mendel's laws of inheritance. Which one of these laws in universal?
2. Consider a hypothetical case of a cross between a tall plant (TT) and a dwarf plant (tt). Work out the phenotypic and genotypic ratios of the F2 progeny if the cross were to show
   (a) dominance (b) incomplete dominance
3. What will be the blood group of the progeny of parents with AB and O groups.
4. Write notes on :
   (a) recessive lethal genes (b) pleiotropy
   (c) linkage groups (d) mitochondrial inheritance
   (e) human karyotype (f) human genome

5. Why do we find so many different complexions among humans?
6. State the chromosomal theory of inheritance.
7. Work out the following crosses and mention the phenotype ratio of their progeny.
   (a) A colour blind man marries a carrier woman
   (b) A man with normal colour vision marries a carrier woman.
8. Why is X-linked inheritance termed cris-cross inheritance?
9. Give an account of genetic disorders caused by abnormal chromosomal number.
10. What is amniocentesis? How and for what is it carried out?

ANSWER TO INTEXT QUESTIONS

21.1 1. Gregor Johann Mendel, first to suggest principles underlying heredity

2. (i) homozygous = bearing identical alleles of a trait; heterozygous = bearing dissimilar alleles of a trait.
   (ii) Dominant allele = expressing in both heterozygous and homozygous conditions.
   Recessive = expressing only in homozygous condition.
   (iii) Genotype = genetic constitution of an individual
   Phenotype = outward expression of a gene
   (iv) monohybrid = cross between two parents differing in a single pair of contrasting character; dihybrid corss = cross of two parents differing in two pairs of contrasting characters.
3. Heredity : transmission of characters from one generation to next generation.
   Variation: Differences between individuals of same species.
4. Monohybrid ration = 3 : 1, Dihybrid ration = 9 : 3 : 3 : 1.
5. Mutation, Recombination.

21.2 1. (i) Alleles are diffrerent forms of a gene.
   (ii) Both alleles express as dominant phenotype.
   (iii) Many genes controlling same trait.
   (iv) whose presence in an individual proves to be fatal.

2. (i) Codominance and multiple alleles

   (ii) Incomplete dominance

   (iii) Polygenic inheritance

   (iv) Polygenic inheritance

3. 1 : 2 : 1

21.3 1. Genes are segments of DNA. They are located in chromosomes.

2. Sutton and Boveri

3. (i) Linkage is the tendency of genes residing on the same chromosome to be inherited together.

   (ii) Breakage and exchange of genes between two chromatids of a homologous pair is termed crossing over.

4. During prophase I of meiosis

5. Human female produces only one kind of gametes (homo = same)

6. No. Because gene for color blindness on X chrosomes is a recessive gene so gets marked by renual gene from mother.

21.4 1. Because mitochondia are inherited from the mother through the ovum.

2. Seven

3. Kline felter : 2n = 47; XXY

   Turner : 2n = 45; XO

   Mongolism : 2n = 47; Trisomy of chromosome 21