



Notes

22

PRINCIPLES OF GENETICS

It is a common observation that seeds of mango trees germinate to grow into mango plants, and dogs give birth to puppies only and not into the young ones of any other animal. Humans give birth to human beings. The tendency of offsprings to inherit parental characteristics is termed as '**heredity**' and the study of science of heredity and the reasons governing the variation between the parents and their offsprings, is called '**Genetics**'. Genetics also seeks to answer questions like why two offspring of same parents look different, why some people have dark, and others have fair complexion. In other words, why is there **variation** among individuals of the same kind. This lesson deals with heredity and the reasons behind the variation among individuals of the same species. It also includes diagnostic techniques to find out the bases for types of sex determination, inheritance of blood groups in humans, hereditary disorders and gives an insight up the human genome as amniocentesis.



OBJECTIVES

After completing this lesson, you will be able to :

- *explain the terms heredity and Genetics;*
- *describe Mendel's experiments on garden pea and the principles derived;*
- *define the terms hybridization, alleles, trait, dominance, recessive, homozygous, heterozygous, genotype, phenotype;*
- *explain incomplete dominance, polygenic inheritance, pleiotropy and lethal genes with examples.*
- *explain the chromosome theory of heredity;*
- *define and give examples of linkage, crossing over and criss-cross inheritance;*
- *explain sex determination in honey bees, birds and humans.*
- *justify mitochondrial inheritance as a case of maternal inheritance;*
- *describe the human karyotype;*
- *list and describe the causes and symptoms of some common genetic disorders e.g. Colour blindness, haemophilia, Down's syndrome, Turner's syndrome, Klinefelter's syndrome;*

- describe the inheritance of Rh factor and explain its significance during pregnancy;
- explain inheritance of human blood groups;
- explain the diagnostic technique of amniocentesis and give its significance;
- give a brief idea of genomics and human genome.

22.1 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 from parents to children and so on generation after generation. This inheritance of genes is termed '**heredity**' the study of reasons of heredity is '**Genetics**'. New individuals develop features according to the genes inherited by them from their parents.

The transmission of characters from one generation to the next, that is from parents to offsprings 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 their offsprings or between offsprings 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 the time of 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 paternal and maternal chromosomes with different gene combinations. Such a source of variation which is most common is called genetic **recombination**.

Heritable Variations generally arise because of mutation and recombination.

22.2 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 on plant breeding and published his results in 1865. His work, however, was independently 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**.



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22.2.1 Mendel's Experiments

Mendel designed his experiments in such a way 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, that is anthers of flowers borne on dwarf plants were removed and their stigmas were dusted by pollen from flowers of tall plants.

In the following spring, seeds from the new plants were collected and sown. Mendel 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. 22.1).

In this way he tried to cross pea plants differing in seven such contrasting characters or **traits**. These were 1. red flowered and white flowered plants; 2. axillary flowered (flower arising in the axil of the leaf) and terminal flowered (flower arising at tip of stalk); 3. yellow seeded versus green seeded; 4. round seeded versus 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 these contrasting characters existed in varieties that were 'self pollinating' so that generation after generation they expressed only one type of feature (Fig. 22.2).

Crosses involving plants differing in the inheritance of **one contrasting feature only** are called **monohybrid crosses**. Mendel also tried crosses involving two contrasting features, such as tall and red flowered plants crossed with dwarf and white flowered plants. Such crosses are termed **dihybrid crosses**.

22.2.2 Mendel's Principles (laws) of inheritance

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

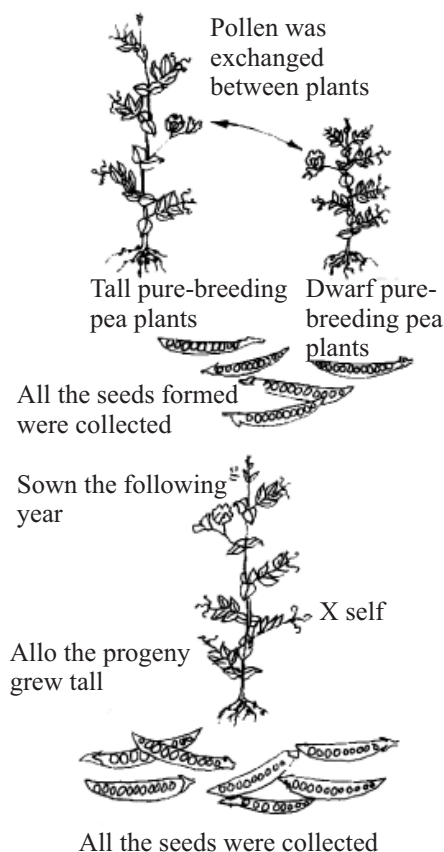


Fig. 22.1 Mendel's experiment with tall and dwarf plants.



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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.

Mendel's factors later came to be known as genes.

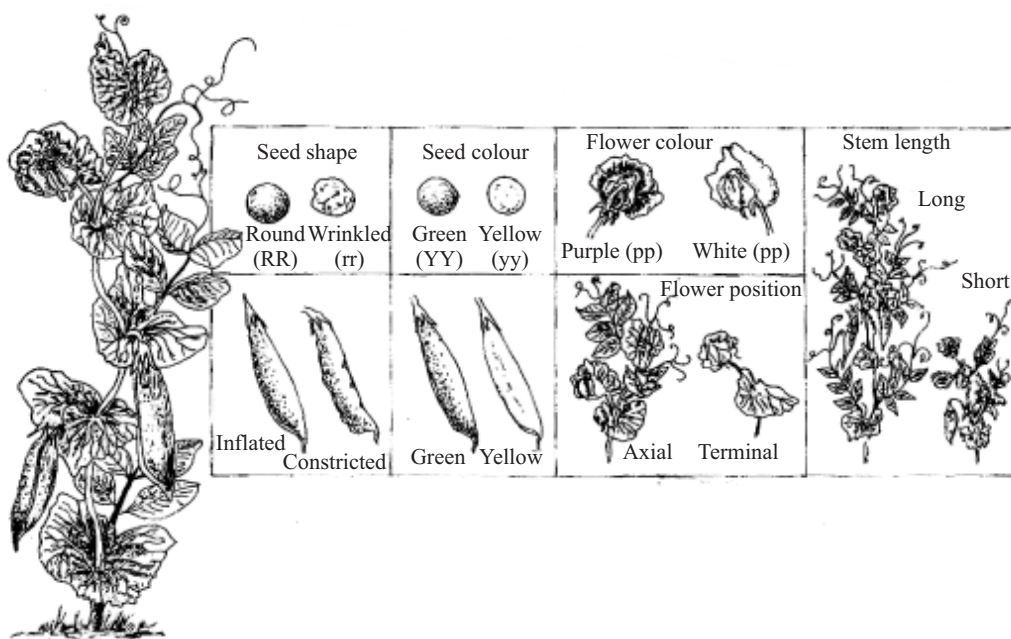


Fig. 22.2 Seven traits studied by Mendel

2. **Law of dominance.** During inheritance of many traits (e.g. eye colour, flower colour, seed shape) is controlled by **one pair of genes**. When the two genes of a pair are of the same kind (e.g. brown colour of eyes, red colour of flower) the condition is termed as **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 another member of the pair (termed its **homologue**) the condition is termed **heterozygous**. The factors or genes for red and white flower colour are alternative forms of the same gene, that is, the gene for flower colour. Such alternative forms of the same gene are termed as **Alleles**.

The second law of inheritance maintains that when the two genes of a pair, represent contrasting characters **the expression of one is dominant over that of the other**. Thus if both genes of an allele are for tallness (represented as TT)



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that is **homozygous** or one gene is for tallness and another for dwarfness (Tt), that is **heterozygous**, the pea plants will be tall. The opposite of dominant gene is termed **recessive gene**. The recessive feature (e.g. dwarfness of the plant) is expressed only when both the genes of allele are in the homozygous condition (tt). The law of dominance was found to be true in both monohybrid and dihybrid crosses in cases of all the seven 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. 22.3) i.e. the segregation of one pair of factors is independent of the segregation of the factors belonging to any other pair of factors or allelic pair.

	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	Tttr Tall white
tR	TtRR Tall red	TtRr Tall red	ttRR Dwarf red	ttRr Dwarf red
tr	TtRr Tall red	Tttr Tall white	ttRr Dwarf red	tttr Dwarf red

Fig. 22.3 Dihybrid phenotypic ratio

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

In Fig. 22.3 results show independent assortment in two pairs of genes. R stands for red flower colour, r for white flower colour, T for tallness and t for dwarfness. You would have noticed that the composition of genes termed **genotype** controls the outside expression which we can 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 and polygenic inheritance.



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22.2.3 Reasons for Mendel's success

1. **Mendel succeeded in postulating laws of inheritance because of his choice of experimental plant** garden pea which has a short life cycle, has self pollinated bisexual flowers so that cross-pollination is not allowed and the true breeding behaviour of parents could be maintained. Because of the property of self pollination in garden pea plants, a large number of pure line of plants with several pairs of contrasting characters could be obtained in the same field.
2. **His selection of traits :** All the seven pairs of contrasting characters of pea plants considered by Mendel in his experiments showed complete dominance that helped Mendel to postulate the law of dominance and the law of segregation.
3. The factors for all the seven traits selected by Mendel for his experiments were either present on separate homologous chromosomes or if they were present on the same chromosome, they were apart so that the factors segregated independently & were not inherited together so that Mendel failed to discover linkage and crossing over.
4. **Mendel's methodology:** His technique of experimentation also helped him in discovering the Laws of Heredity :
 - (i) Homozygous pure line plants with contrasting characters were crossed.
 - (ii) Self pollination was prevented by removing stamens to bring about cross-pollination between the desired parents.
 - (iii) Female plants were dusted with pollen grains from another plant with the contrasting feature and were tied in a bag to prevent any further pollination.
 - (iv) Seeds were collected from plants of different generations and sown in time.
 - (v) The results of different generations were maintained, and analysed statistically, by counting the individuals exhibiting different traits.
 - (vi) He considered the inheritance of one character at a time, then he considered inheritance involving individuals differing in two contrasting characters.
 - (vii) He performed reciprocal crosses and test crosses to confirm the results. (see section 22.3 for definition of these terms), and formulated the basic laws of heredity.

22.3 IMPORTANT TERMS IN GENETICS

- **Factor :** The unit of inheritance and expression of a particular character is controlled by inheritable units called factor (gene) which are present in pairs in parental cells and singly in the gametes.
- **Gene :** A segment of DNA molecule which determines the unit of inheritance and expression of a particular character.



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- **Alleles or Allelomorphs :** Two or more alternative forms of a gene are called **alleles**. For example in pea plant, the gene for producing seed shape may occur in two alternative forms: smooth (S) and wrinkled (s). Genes for smooth wrinkled seeds are alleles of each other, and occupy same locus on homologous chromosomes.
- **Trait :** is the morphologically or physiologically visible character, e.g. colour of flower, and shape of seed.
- **Dominant trait :** Out of the two alleles or allelomorphs of a trait, the one which expresses itself in a heterozygous organism in the F_1 hybrid is called the dominant trait (dominant allele) and the one that remains masked in F_1 individual but gets expressed in the next generation (F_2), is called **recessive**. Thus, if the allelic combination in an organism is Tt, and T (tallness) expresses itself but t (dwarfness) cannot, so T is the dominant allele, and tallness is dominant on dwarfness represented by “t”.
- **Recessive trait :** Out of the two alleles for a trait, the one which is suppressed (does not express) in the F_1 hybrid is called the recessive trait (recessive allele). But the Recessive allele does express itself only in the homozygous state (e.g. tt).
- **Genotype :** A class of individuals recognised based on its genetic constitution and breeding behaviour is called the genotype, e.g., the genotype of pure smooth seeded parent pea plant is SS and it will always breed true for smooth-seeded character, but plants having Ss on selfing would give rise to a population represented by 3 : 1 ratio for smooth seeded plants and wrinkled seeded plants.
- **Phenotype :** A class of individuals recognised based on outward appearance of a trait in an individual is the phenotype, e.g. Smooth-seeded shape or wrinkled shape of seeds represent two different phenotypes.
- **Homozygous :** An individual possessing identical alleles for a trait is termed homozygous e.g. SS is homozygous condition for smooth seeded character in garden-pea.
- **Heterozygous :** An individual with dissimilar alleles for a trait is termed heterozygous for e.g. Ss represents the heterozygous condition for smooth seeded character in garden pea.
- **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 pollination 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 is the

Monohybrid. The phenotypic ratio of 3 dominants : 1 recessive obtained in the F_2 generation from the monohybrid crosses by Mendel was mentioned as 3:1 **monohybrid ratio**.

- **Dihybrid cross :** The cross in which two parents differing in two pairs of contrasting characters are considered simultaneously for the inheritance pattern is called dihybrid cross. The phenotypic ratio obtained in the F_2 generation from a dihybrid cross is called Mendelian dihybrid ratio (9 : 3 : 3 : 1), and the F_1 -individual is called dihybrid (Ss Tt).
- **Hybridisation :** Crossing organisms belonging to different species for getting desirable qualities in the offspring.
- **Test cross :** is the 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 between its different genotypes and phenotypes.
- **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.



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INTEXT QUESTIONS 22.1

1. Name the founder of genetics and state why he is called so.
.....
2. State one difference between
 - (i) homozygous and heterozygous individuals
.....
 - (ii) dominant and recessive traits
.....
 - (iii) genotype and phenotype
.....
 - (iv) monohybrid and dihybrid crosses.
.....
3. Define heredity and Genetics.
.....
4. Give the monohybrid and dihybrid phenotypic ratios for Mendelian inheritance.
.....
5. Mention two sources of variation.
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22.3 DEVIATION FROM MENDEL'S LAWS INCOMPLETE DOMINANCE

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

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 the same, that is, 1 : 2 : 1.

Multiple alleles and codominance

Height and flower colour in peas and eye colour of humans have only two **alleles** (T and t; R and r; B and b (alleles for Brown blue eyes in humans)). Most genes, however, may have more than two alleles or **multiple alleles**, controlling the same Trait. An example of multiple alleles is inheritance of blood group in man.

The four blood groups of humans are determined by combination of different alleles. 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 have the blood group AB as both the genes I^A and I^B are **co-dominant**. The gene i^o when homozygous ($i^o i^o$) gives the blood group O. Genotype and phenotype of blood groups in humans are given in Table 22.1.

Table 22.1 Genotypes and Phenotypes of human blood groups

Genotype	Blood group
$I^A I^A$ and $I^A i^o$	A
$I^B I^B$ and $I^B i^o$	B
$I^A I^B$	AB
$i^o i^o$	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 (y) which is also responsible for killing the mouse in homozygous (yy) condition at the zygotic stage indicating thereby that the mice

homozygous for dominant “Y” allele (that is, true breeding for yellow oat colour) are never borne. Such a combination of genes (y) are termed **lethal genes**, and the phenomenon is called **lethality**. Some lethal genes kill an individual only in the homozygous condition and are **recessive lethals**.

Pleiotropy

While a gene may have multiple alleles and thus give multiple genotypes, one gene may control several phenotypes. 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 vestigial wings and curled abdomen.

Polygenic or quantitative inheritance

When a trait (feature or character) is controlled by a single gene representing an allelic pair it is termed **monogenic inheritance**. However, many traits or features are controlled by a number of different genes present at different loci on the same chromosome or different chromosomes. For example, the height and 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 different loci on many chromosomes. Each of these genes has equal contribution and cumulative 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 an inheritance controlled by many genes having additive or cumulative effect in terms of expression of the phenotypic character, is termed as **quantitative inheritance** or **polygenic** (poly meaning or due to many genes) **inheritance**.



INTEXT QUESTIONS 22.2

1. Define :

- (i) An allele
- (ii) Codominance
- (iii) Polygenes
- (iv) Lethal genes

2. Name the kind of inheritance in terms of expression of

- (i) blood groups of humans
-
-
-



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(ii) wheat kernel colour

(iii) human skin colour?

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

22.4 CHROMOSOMAL THEORY OF INHERITANCE

Sutton and Boveri in **1902** observed that

Chromosomes from two parents come together in the zygote as a result of the fusion of two gametes and again separate out during meiosis at the time of formation of gametes. You have already learnt that chromosomes are filamentous bodies present in the nucleus and seen only during cell division. Gametes have half (n) number of chromosomes or are haploid and zygote is diploid or has (2n) or double the number of chromosomes when compared to chromosome number in the gametes.

The observations proved that there is a remarkable similarity between the behaviour of Mendelian factors or genes 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 or 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 genes controlling the same set of characters) 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 Mendelian 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 specific sets of segments of DNA are the genes**.

22.5 LINKAGE AND CROSSING OVER

Bateson and Punnett performed a dihybrid cross with true breeding varieties of sweet pea (*Lathyrus sativus*) and instead of 9 : 3 : 3 : 1 ratio in F₂ generation they got the ratio 7 : 1 : 1 : 7. It means that the characters controlled by the two genes chosen for the experiment do not follow the principle of independent assortment as postulated by Mendel. Instead they tend to be inherited together or are **linked** together. Thus genes present on the same chromosome tend to be inherited together and are said to be **linked**. This phenomenon is called **linkage**.

All the genes present on the same pair of chromosomes and with a tendency to be inherited together forms a linkage group.

In the above experiment some recombinant type of individuals 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 the chromosomes of a homologous pair (Fig. 22.4).

Crossing over occurs during prophase I at meiosis I of the time of gamete formation. The point where crossing over occurs is called **chiasma**. (plural : chiasmata) See Fig. 22.3. 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. 22.4. The figure 22.4 shows linked genes of the parents and **recombinants** due to crossing over.

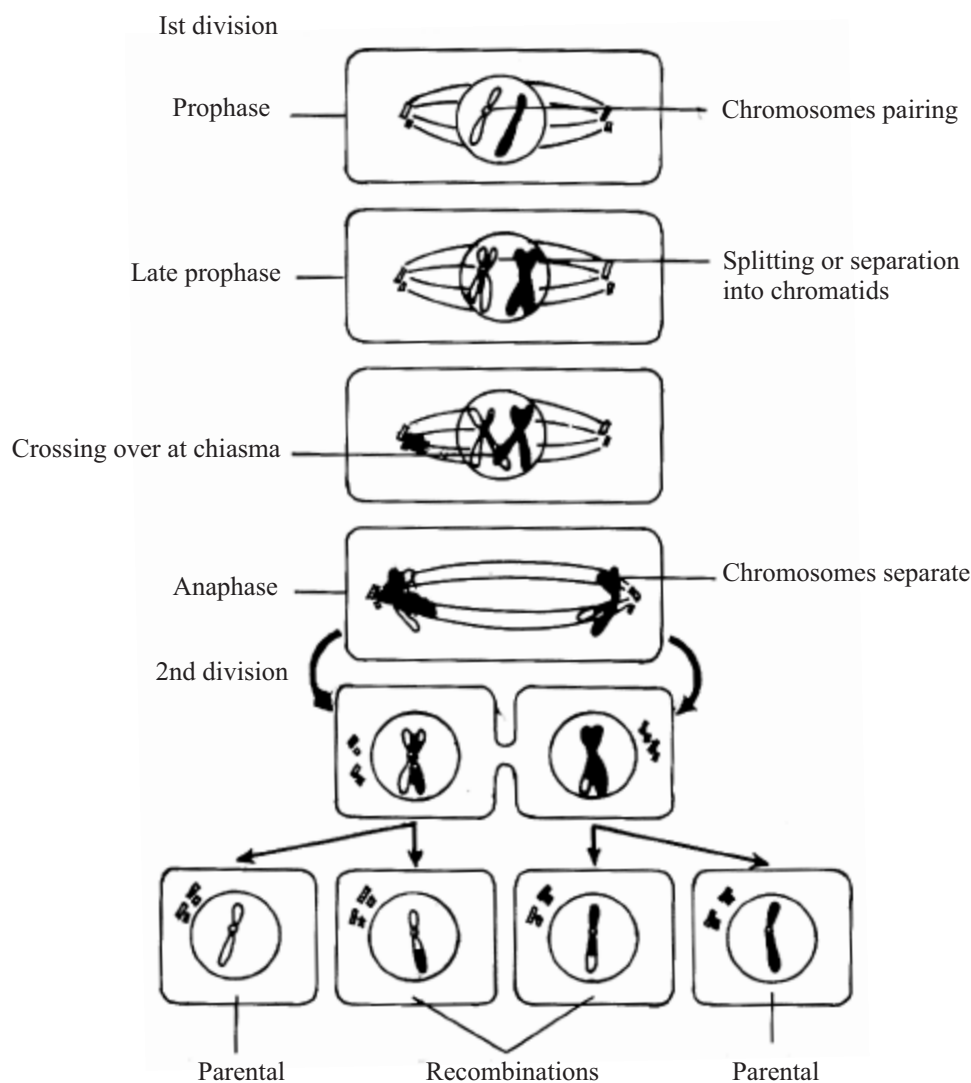


Fig. 22.4 Schematic diagram showing recombination by crossing over



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22.6 CHROMOSOMES AND SEX DETERMINATION

Sex of the unborn individuals is determined in different ways in different kinds of organisms. You will learn about sex determination in humans, birds and honey bees in this section.

In some diploid organisms, specific chromosomes have a role in sex determination. Such chromosomes 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 gametes (containing X). For example : all eggs of the human female contain an X chromosome and autosomes. So human female is termed as homogametic.

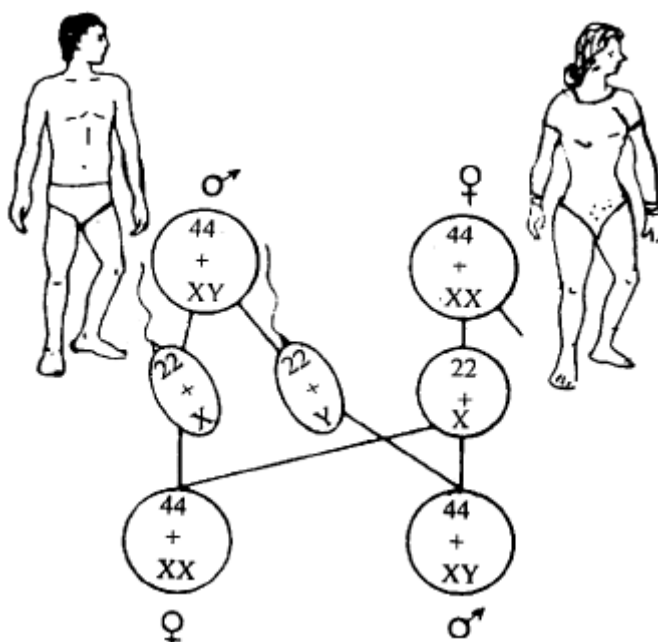


Fig. 22.5 Chromosomal basis of sex determination in humans.

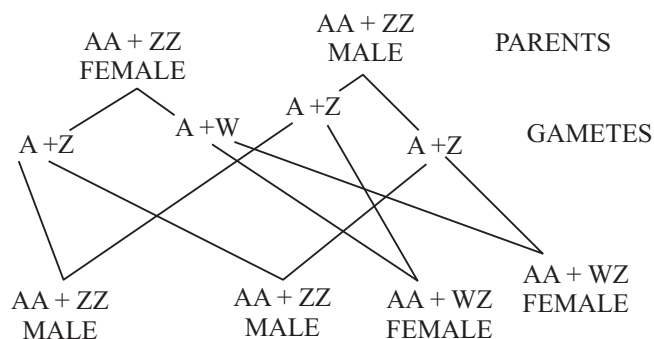
22.6.1 Sex Determination in Human

- Sex chromosomes in males are morphologically dissimilar (i.e. XY). Such individuals produce two types of gametes (one containing X and the other containing Y) and are called heterogametic. 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 girl is born, and if human egg is fertilized by a sperm having “Y” chromosome, a boy is born (Fig. 22.6). 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.

22.6.2 Sex Determination in Birds

You have just studied the XX-XY type of sex determination in humans. This type of sex determination is found in other mammals and most insects. However, the

method of sex determination in birds is a little different. In birds both sexes (male and female) possess two sex chromosomes but unlike human beings the female has the heteromorphic morphologically different sex chromosomes (ZW) while the males bear homomorphic (condition, the sex chromosomes (ZZ)). Thus, the females are heterogametic and produce two types of eggs: A+Z and A+W ('A' stands for autosomes). The male gamete is only of one type: A+Z. This type of sex determination is called ZW-ZZ type or WZ-ZZ type of sex determination. The letters Z and W are used to distinguish these types of sex chromosomes from X and Y chromosomes found in the X-Y type of sex determination.



ZW - ZZ Type of Sex Determination in birds AA - Autosomes

22.6.3 Sex Determination in Honey Bees

Honey bees have a unique method of sex determination. In honey bees, fertilised eggs emerge as females and unfertilised eggs develop into males. Since fertilised eggs and also females are diploid and unfertilised eggs and males haploid, sex determination in honey bees is referred to as **haplodiploidy**-sometimes also called **arrhenotoky**.

The sex is determined by the number of sets of chromosomes an individual receives. The male, which is called a drone, is produced from **unfertilized haploid eggs**. And thus, male honeybees contain a single set of chromosomes. The female honeybees, which are worker bees and queen bees, are produced from fertilized eggs and therefore are diploid. They contain two sets of chromosomes. In this case, only females are produced by sexual reproduction.

It is very interesting in honey bees that males have no father and cannot have sons but have a grandfather and can have grandsons.

22.7 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 criss-cross inheritance as shown in Fig. 22.6.



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When a male has a defective sex linked gene located on **X** chromosome he transmits the defective **X** chromosome to his **daughter only during reproduction**. The female who has this gene transmits it to her son 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 the genetic defect due to the presence of faulty gene on the single X-chromosome while females are only **carriers** of these defective genes as they have the other X which masks the effect of faulty gene. The trait shows up in females only both X chromosomes from mother and father have faulty gene.

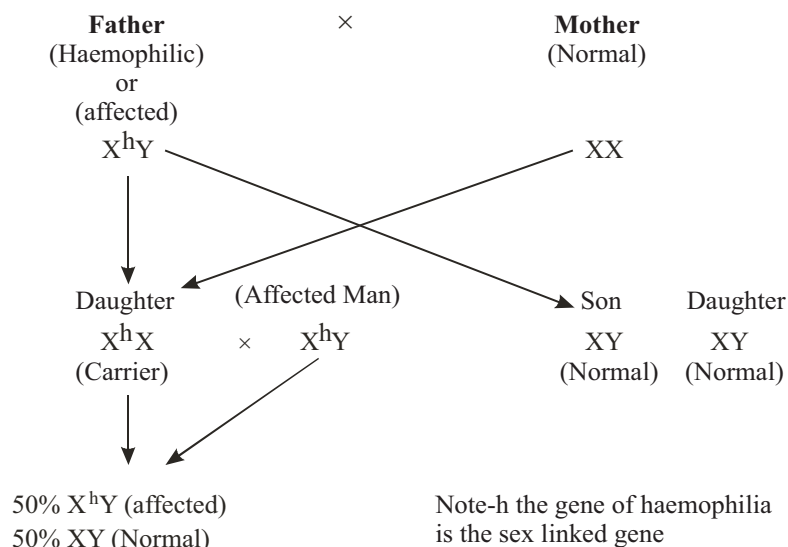


Fig. 22.6 Criss cross inheritance or X-linked sex linked inheritance

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**.

Criss 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 to mask the effect of the faulty gene. See Fig. 22.7 (a), and (b).



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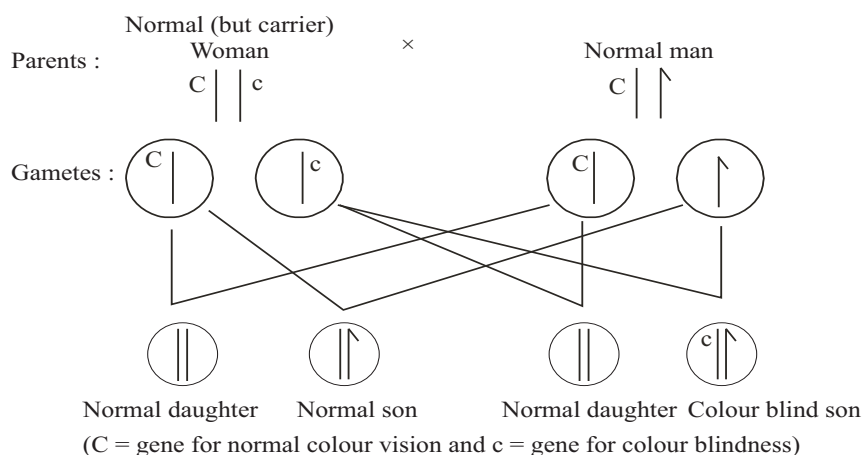


Fig. 22.7 (a) Progeny of carrier female for colour blindness and a man with normal colour vision.

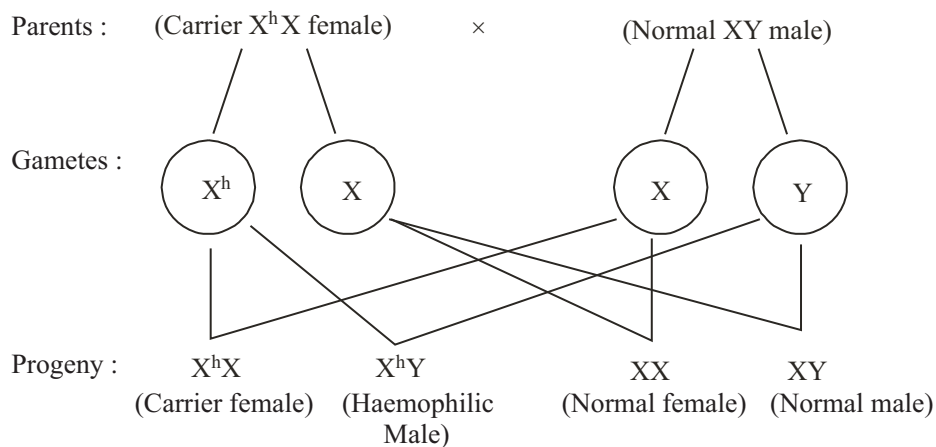


Fig. 22.7 (b) Progeny of a haemophilic carrier female and normal male



INTEXT QUESTIONS 22.3

- What are genes and where are they located?
.....
- State the names of the scientists who proposed the chromosomal theory of inheritance.
.....
- Define (i) linkage and (ii) crossing over.
.....
- When does gene exchange through chiasma formation occur between homologous chromosomes?
.....



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5. Why is the human female called the homogametic sex?
.....
6. A colour blind man married a normal woman whose father and mother both had normal colour vision. Will any of their sons be colour blind? If not why not.
.....
7. With the help of flow chart explain the difference in sex determination in birds and mammals.
.....
8. Name an insect in which all males are produced parthenogenetically.
.....
9. In honey bees “males have no father and cannot have sons but have a grandfather”. Justify the statement.
.....
10. Which sex in birds is heterogametic?
.....
11. Why is sex determination in honeybees called haplodiploidy
.....

22.8 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 that genes are present on the chromosomes present in the nucleus. Since mitochondria come into the zygote from the egg, inheritance of mitochondrial 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.

22.9 HUMAN KARYOTYPE

Human karyotype is the arrangement of human chromosomes in seven groups according to the types of chromosomes and their size. It is prepared by arranging chromosomes seen at mitotic metaphase in descending order with the longest pair of chromosomes drawn first, and the sex chromosomes are drawn the last :

- (i) Total no. of chromosomes or $2n = 46$ (23 pairs).
- (ii) Number of autosomes = 44 (22 pairs).
- (iii) Sex chromosomes $2 = X$ and Y
- (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. 22.8.

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

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

Normal female has 22 pairs of autosomes + two X chromosomes

Presence of Y is necessary for maleness.

Absence of Y chromosomes, makes the individual a female with some defective characters.

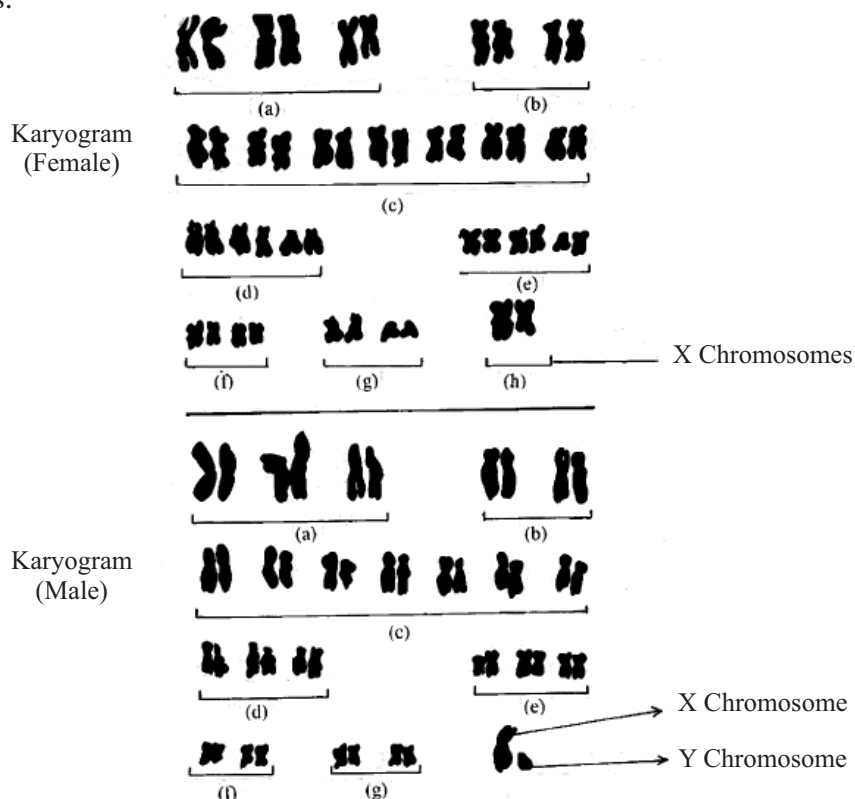


Fig. 22.8 Karyotype showing chromosomes of normal male. Female has the same autosomes but two X-chromosomes instead of XY

22.10 CHROMOSOMAL ABNORMALITIES AND GENETIC DISORDERS IN HUMANS

Any change from the normal number or structure of chromosomes causes abnormalities. Following are some examples of human genetic disorders :

1. Mongolism or Down's syndrome

The individual has 47 chromosomes because of one extra chromosome in the 21st pair (Trisomy of chromosome 21). The outcome of this defect are the following characters or features :

- mentally retarded
- have a thick tongue
- and a drooping (false expression of pleasure) face. Fig. 22.9.



Notes

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

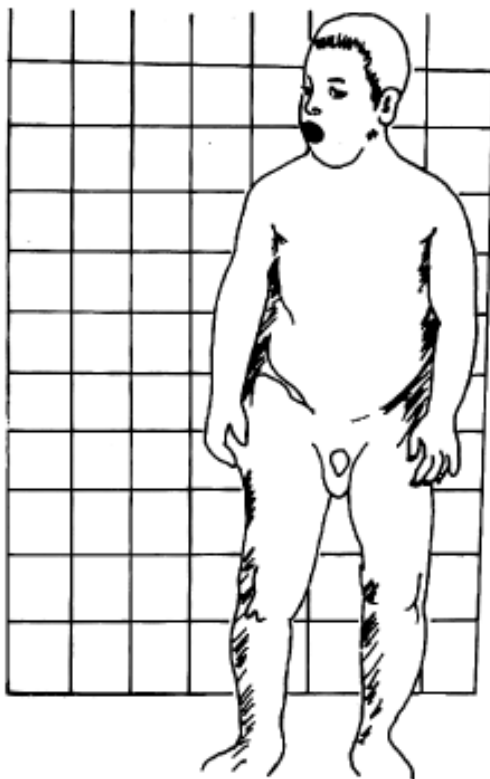


Fig. 22.9 Mongolism or Down's syndrome

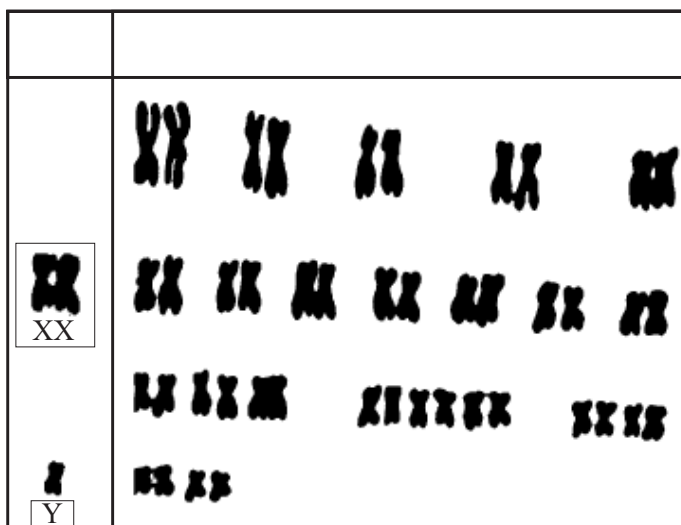


Fig. 22.10 Man showing Klinefelter's syndrome

2. Klinefelter's syndrome

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

- Tall, mentally retarded male;
- Sterile and shows breast development or **gynaecomastia** (gynae : female; massere : mammary glands). Fig. 22.10.

3. Turner's syndrome

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

- Mentally retarded
- web like skin on neck.
- incompletely developed breasts. Fig. 22.11.

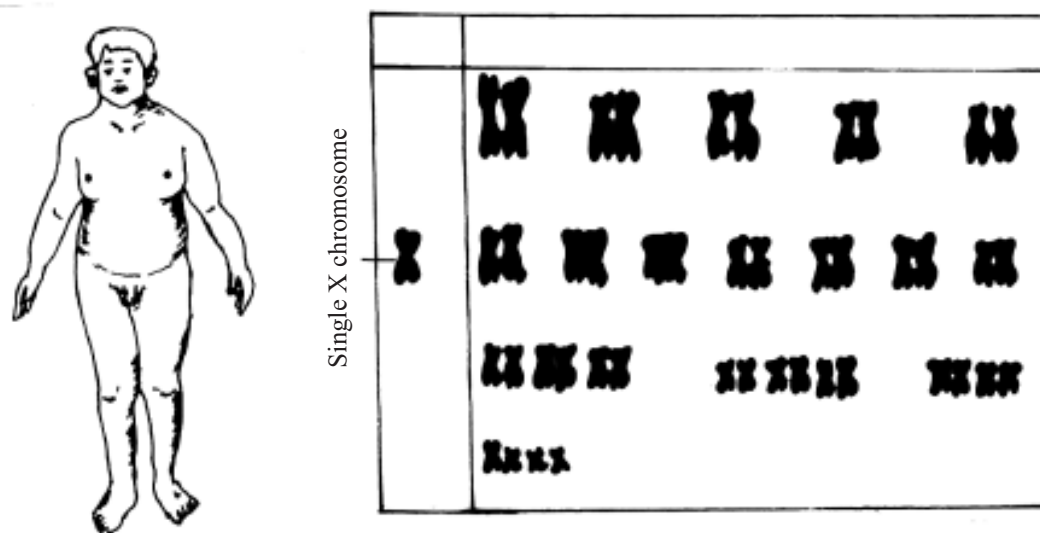
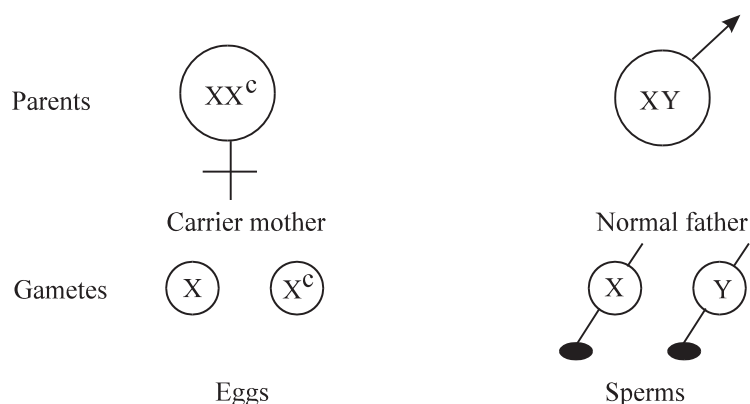


Fig. 22.11 A woman suffering from Turner's syndrome.

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

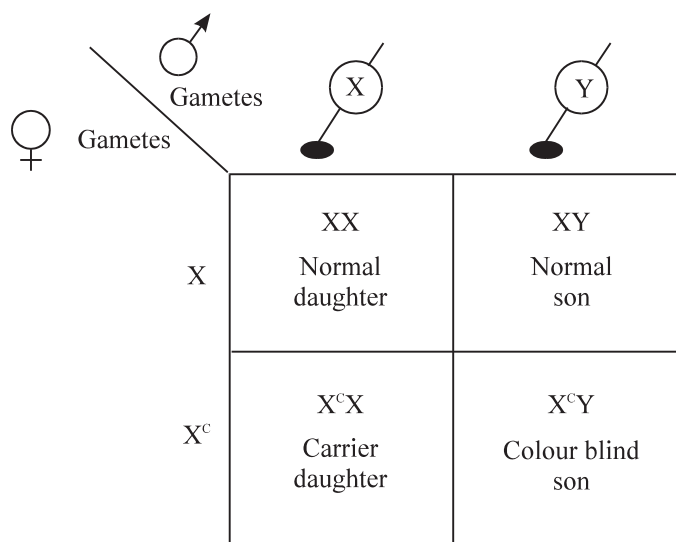
Both these defects are sex linked disorders. (See figures 22.6 and 22.7)





Notes

The inheritance is as follows :



Note : X = normal allele; X^c = recessive mutant

Fig. 22.12 Inheritance of colour blindness

See also 22.7

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. In a carrier daughter the defective gene received from the mother is masked chromosome received from normal father by normal allele on the other X (Fig. 22.12).

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 an autosomal disorder in which normal haemoglobin is not synthesised. So, frequent blood transfusions are required for survival.

The defective gene is recessive and present on an autosome in the 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 autosomal 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. For heterozygous individuals with sickle

cell gene, it is a boon in disguise against malaria for children with one defective haemoglobin gene can survive as they are less affected by malarial because the malarial parasite cannot thrive inside the defective RBCs.

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 woman whose husband is Rh + may bear a child who may have inherited the Rh + gene from the father. If the foetal blood of the Rh + foetus enters mother's body stream, her immune system produces antibodies against Rh antigen which may cause minor problems in first pregnancy. Antibodies remain in the mother's blood and in the subsequent pregnancies, the mother's antibodies against foetal Rh antigen may enter the foetal blood stream and destroy its red blood corpuscles causing severe anemia which may even be fatal (erythroblastosis foetalis) for the foetus.

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. 22.13)

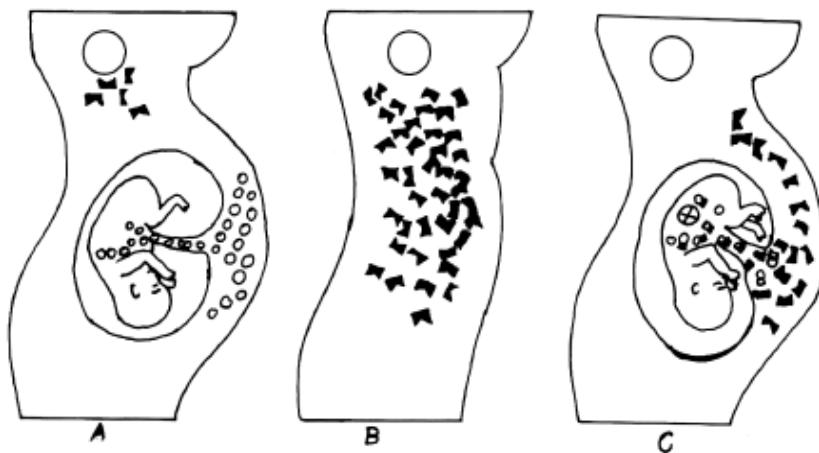


Fig. 22.13 The mechanism of Rh inheritance

- (a) shows the first pregnancy where the mother is Rh (-) and foetus Rh (+). Antigens (empty circles) of the foetus stimulate the production of anti bodies (black blocks) in the mother's blood.
- (b) shows the retention of anti bodies 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 mother's body destroy the infant's red blood cells.

22.11 AMNIOCENTESIS

Amniocentesis is a technique by which hereditary disorders due to defects in genes can be detected. In this technique (Fig. 22.14)



Notes



Notes

- (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.

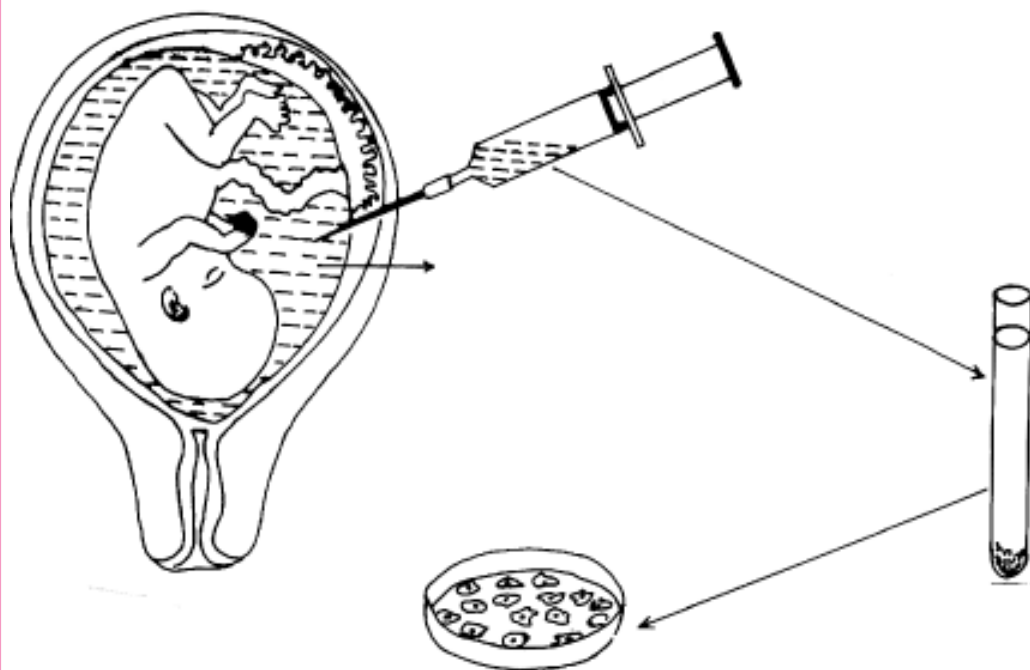


Fig. 22.14 Techniques of Amniocentesis

22.12 THE HUMAN GENOME

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

You have just read about genetic disorders and today there is hope for cure through gene therapy. This is because in 2003, most of the genes on human chromosomes have been mapped or located on the 23(n) chromosomes. The genes responsible for inheritance of various structural features, that control various enzymes that catalyse the various biochemical reactions in the body, and genes responsible for genetic disorders have been located. **Genome means genes of a particular organism on its haploid set of chromosomes and study of genome is Genomics.** 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 in 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 box) 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.



Notes



INTEXT QUESTIONS 22.4

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 syndrome and in Mongolism.
.....



WHAT YOU HAVE LEARNT

- Heredity means the transmission of characters from parents to offsprings.
- Variation pertains to differences between siblings or members of same species.
- Mendel was the first to explain that heredity involves transmission of certain factors from reproductive cells of parents to offsprings.
- Hugo de Vries, Correns and Tschermach rediscovered Mendel's Laws of inheritance nearly 35 years after Mendel's death.
- Mendel selected seven varieties of garden pea differing in seven pairs of contrasting characters.
- According to his 'law of segregation' the factors segregate at the time of gamete formation, and come together after fertilization.
- Mendel's 'law of dominance' states when partents differing in a pair of contrasting characters are crossed, the factor that expresses itself in the F-1 is called **dominant**, and the factor which is masked by **dominant** factor, is called **recessive**.
- Law of independent assortment states that the inheritance of factors controlling one character does not depend on inheritance of any other factor controlling any other character.



Notes

- There are deviations from Mendelian inheritance and these patterns of inheritance are incomplete dominance, codominance, multiple alleles, polygenic inheritance and pleiotropy.
- Sutton and Boveri (1902) proposed the chromosome theory of heredity. It states that Mendelian factors or genes are located on chromosomes.
- Genes are 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 are responsible for homogametic female and XY for heterogametic male. In birds it is the opposite—male is ZZ or homogametic and female is ZW or heterogametic.
- In honey bees, males arise from unfertilised eggs and are therefore, haploid or with half the number of chromosomes while females develop from fertilised eggs and are diploid.
- Human males inherit an X chromosome from female parent and Y from the male parents. Y chromosome bears genes for maleness.
- Females receive two X chromosomes one each from either of the two parents.
- Any change in normal number and structure of chromosomes of an individual causes abnormalities.
- A normal karyotype shows 23 pairs of human chromosomes bearing thousands of genes, controlling different characters.
- Down's syndrome patients have 47 chromosomes exhibiting 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 and sex-linked disorders.
- Thalassemia and Sickle cell anaemia are due to a single autosomal 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 is a technique for detecting genetic disorder in foetus.



TERMINAL EXERCISES

1. State the three Mendel's laws of inheritance. Which one of these laws is universal?



Notes

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 chromosome theory of inheritance.
7. Work out the following crosses and mention the phenotypic 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 criss-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?
11. In what way is chromosomal sex determination of humans different from that of birds?
12. From which kind of eggs do males and females of honeybees emerge.

**ANSWERS TO INTEXT QUESTIONS**

- 22.1** 1. Gregor John Mendel, was the first to suggest principles underlying heredity
2. (i) homozygous = bearing identical alleles controlling a trait;
heterozygous = bearing dissimilar alleles controlling 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, represented with the help of symbols.

Phenotype = class of individuals recognised based on externally/ internally visible characters.



Notes

(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 : is the study of transmission of characters from one generation to next generation.

Variation: Differences between individuals of same species.

4. Monohybrid ratio = 3 : 1, Dihybrid ratio = 9 : 3 : 3 : 1.
5. Mutation, Recombination.

- 22.2**
1. (i) Alleles are diffrentent forms of a gene.
(ii) Both alleles express as dominant phenotype.
(iii) Many genes controlling same trait.
(iv) Presence of which kind of genes 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

- 22.3**
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 chromocomes is a recessive gene so it gets marked by renual gene from mother.
 7. Female
 8. Because males develop from unfertilised or haploid eggs and females from fertilised or diploid eggs.

- 22.4**
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