CHAPTER / 06

Sex-Determination

Topics Covered

Sex-Determination
- Sex Chromosomes
- Genetic Mechanisms of Sex-Determination
- Sex Differentiation
- Environmental Factors in Sex-Determination

Sex-Linked Inheritance and Hereditary Disorders
- Sex-Linked Inheritance
- Inheritance of Sex-Linked Characters
- Mendelian Disorders in Humans
- Chromosomal Disorders in Humans

TOPIC ~01 Sex-Determination

The organisms that produce offsprings *via* sexual reproduction have two sexes (unisexual) but some exceptional cases include hermaphrodites (bisexual) in which both male and female structures are present and parthenogenesis, in which the female reproduces without fertilisation.

A biological system that determines the development of sexual characters in an organism constitutes the sex-determination system.

The sex-determination occurs in three steps
- **chromosomal sex-determination** by sex chromosomes
- **gonadal sex-determination** by the differentiation of gonads
- **phenotypic sex-determination** by sex hormones secreted from the gonads.

Mostly, the mechanism of sex-determination is genetic, i.e. controlled by chromosomes bearing genes which express their sexual morphology. In animals, this is often accompanied by chromosomal differences, generally through combinations of sex chromosomes during fertilisation.

Sex Chromosomes

These are those chromosomes which singly or in pair determine the sex of the individual in dioecious or unisexual organisms. These chromosomes are designated as 'X' and 'Y' or 'Z' or 'W'. Such chromosomes are morphologically distinguished from each other. The remaining chromosomes of the cell are known as autosomes and designated as 'A'.

Genetic Mechanisms of Sex-Determination

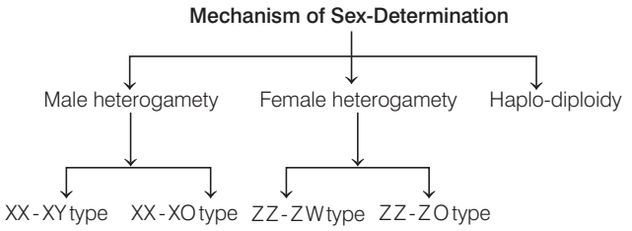
Various genetically controlled sex mechanisms include

- Chromosomal mechanism
- Haplo-diploid mechanism
- Genic-balance mechanism
- Single gene effect

1. Chromosomal Machanism of Sex-Determination

The sex in majority of the organisms is determined by the pair of sex chromosomes called X and Y-chromosome.

The various types of chromosomal sex-determination mechanism observed in different animals are as follows



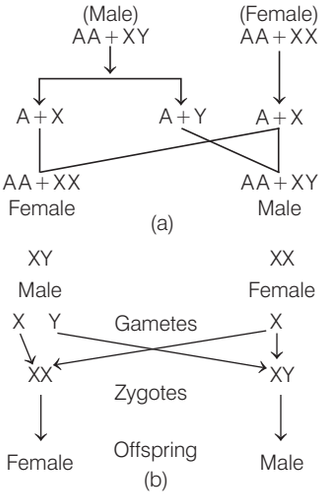
Mechanism of sex-determination

(i) XX-XY Type or Lygaeus Mechanism (Male Heterogamy)

This mechanism was first studied by **Wilson** and **Stevens** in the milkweed bug called *Lygaeus turcicus*.

It is present in certain insects like *Drosophila melanogaster* and mammals including **human beings**.

The males have autosomes plus XY-chromosomes and females have autosomes plus XX-chromosomes. The males produce two types of gametes containing X or Y sex chromosome (heterogametic) and females produce only one type of gametes with an X-chromosome (homogametic).

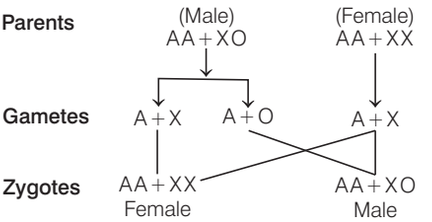


(a) Determination of sex in Drosophila
(b) Pattern of sex chromosomal inheritance in human

(ii) XX-XO Mechanism (Male Heterogamy)

In this pattern, the female has two X-chromosomes (called XX), while male has only one X-chromosome (called XO). The Y-chromosome is completely absent here and it is denoted by the letter O.

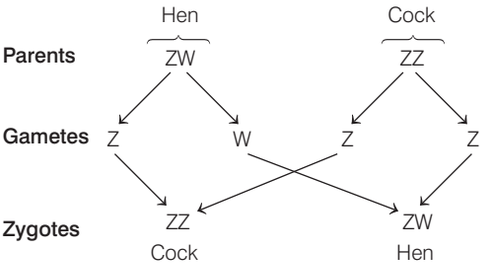
Thus, the presence of unpaired X-chromosome determines the masculine sex. The female produces only one type of eggs and male produces two types of sperms, i.e. 50% with one X-chromosome and 50% without any sex chromosome, e.g. grasshopper and bugs.



Determination of sex in grasshopper (XX–XO type)

(iii) ZZ-ZW Mechanism (Female Heterogamy)

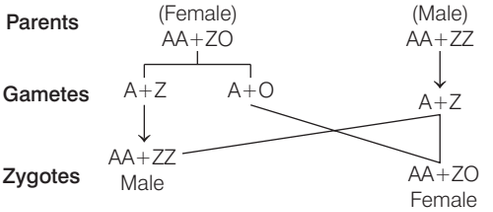
This mechanism of sex-determination is seen in birds, fowls and fishes. Females have one Z and one W-chromosome (i.e. heterogametic) along with autosomes whereas males have a pair of Z-chromosomes (i.e. homogametic).



Determination of sex (ZZ-ZW) in fowl and birds

(iv) ZZ-ZO Mechanism (Female Heterogamy)

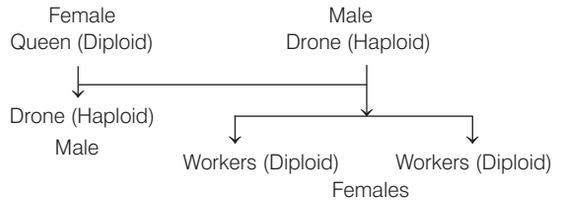
In this mechanism of sex-determination, the female is heterogametic (ZO) and male is homogametic (ZZ). It occurs in Lepidoptera, e.g. certain butterflies and moths.



Determination of heterogametic and homogametic female and male

2. Haplo-Diploidy Mechanism

In insects like honeybees, wasps, ants, etc., the sex chromosomes are not differentiated and sex is determined on the basis of ploidy of the individual. In honeybees, males (drones) are haploid (n =16) and develop from unfertilised eggs of females (Arrhenotoky) by mitosis like division. The females are diploid ($2n$ = 32), i.e. queen and workers, and develop from fertilised eggs. This method of sex-determination is referred to as haplo-diploidy method and the developmental process is also known as parthenogenesis.



Haplo-diploidy mechanism of the sex-determination in honeybee

3. Genic Balance Mechanism

Genic balance theory by **Bridges** proposed the sex-determination mechanism based on the ratio of number of X-chromosomes (X) and sets of autosomes (A).

The table given below describes about the phenotypic sex of *D. melanogaster* based on X/A values

X/A Ratios and Corresponding Sexual Phenotypes in *Drosophila*

Chromosome complement	Ratio= X/A	Sex
2A+XX	2/2=1.0	Female
3A +XXX	3/3 = 1.0	Triploid female
4A+XXXX	4/4=1.0	Tetraploid female
2A + XXX	3/2=1.50	Superfemale
3A +XX	2/3 = 0.67	Intersex
2A +XY	1/2 = 0.50	Male
3A +XY	1/3 = 0.33	Supermale

4. Single Gene Effect

In certain organisms-like *Drosophila*, human, *Asparagus* and several fishes, a single gene pair is responsible for the determination and expression of sex.

In *Drosophila*, the sex is expressed by a recessive gene called *tra* (transformer) present on the third autosome. Males and female members with dominant (tra^+) allele are mostly fertile. However, a normal female (i.e. AA + XX), having homozygous recessive *tra* alleles, develops into sterile male.

In humans, the Y-chromosome has a *sry* gene which influences the development of testis in males. Its absence results in development of ovaries in females.

Sex Differentiation

In vertebrates and some invertebrates, the embryo develops certain traits of opposite sex along with its own. This indicates that sex of an organism changes under specific conditions. This may happen as a result of hormones secreted from the gonads of such organisms.

Some examples of sex differentiation are given below

Sex Reversal

Artificial removal of gonads of either sex before puberry (castration or ovariectomy) results in the development of secondary sexual characters of the opposite sex. It is observed in fishes, amphibians, birds and some mammals, including humans.

Freemartin

It is an infertile female mammal with masculinised behaviour and non-functioning ovaries. Freemartinism is the normal outcome of mixed sex twins in all cattle species, i.e. it occurs when the twins of opposite sex are born, e.g. sheep, goats and pigs.

Mostly the male hormones are produced first. If both foetuses are of the same sex this is of no significance, but if they are of different sex, male hormones pass from the male twin to the female twin.

The male hormones then masculinise the female twin and the result is a freemartin or sterile masculine female.

Environmental Factors in Sex-Determination

In some lower animals, sex-determination is non-genetic and depends on the factors present in the external environment.

Different environmental factors responsible for sex-determination are given below

Chemotactic Sex-Determination

- It is seen in males of the marine worm *Bonellia*. These are small worms that live within the reproductive tract of the larger female.
- In *Bonellia*, the larvae of male and female are genetically hermaphrodite. A newly hatched worm if reared from a single cell kept in isolation, develops into a female. If the larvae are reared with mature females in water, they adhere to the proboscis and transform into males which eventually migrate into the female reproductive tract, where they become parasitic.
- It has been found that a chemotactic substance secreted by the proboscis of a mature female *Bonellia* induces the differentiation of larva into males.

Temperature Dependent Sex-Determination

- In some reptiles, the temperature at which the fertilised eggs are incubated, plays a major role in determining the sex of the offspring.
- Surprisingly high temperature during incubation have opposite effect on sex-determination in different species.
- In turtles, high incubation temperature (above 30°C) of eggs results in the production of female progeny whereas in the lizard and crocodiles, high incubation temperature results in the production of male offspring.
- At the lower temperature range between 22.5-27°C, male turtles are produced. This pattern is reversed in lizards and crocodiles.

PRACTICE QUESTIONS

Exams', Textbook's Other Imp. Questions

1 MARK *Questions*

Important Question

- *Choose the correct option*

Q.1 Y-chromosome is called **[Textbook]**

(a) sex chromosome
(b) androsome
(c) autosome
(d) gynosome

Ans (*a*) sex chromosome

Q.2 A fruitfly exhibiting both male and female trait is **[Textbook]**

(a) heterozygous
(b) gynandromorph
(c) hemizygous
(d) gynandev

Ans (*b*) gynandromorph

Q.3 Genes located on Y-chromosome are **[Textbook]**

(a) mutant genes
(b) autosomal genes
(c) holandric genes
(d) sex-linked genes

Ans (*c*) holandric genes

Q.4 Which gene is present in the Y-chromosome that codes for the protein TDF? **[Textbook]**

(a) *cry*
(b) *sry*
(c) *try*
(d) *tra*

Ans (*b*) *sry*

Q.5 When the ratio of X/A=0.67 in genic balance theory, which type of sex is expressed? **[Textbook]**

(a) Super female (b) Intersex
(c) Super male (d) Triploid female

Ans (*b*) Intersex

Q.6 Which type of sex-determination is found in *Bonellia*? **[Textbook]**

(a) Temperature dependent (b) Chemotactic
(c) Holandric (d) Pseudoautosomal

Ans (*b*) Chemotactic

Q.7 Number of Barr bodies present in Turner syndrome is **[Textbook]**

(a) 0 (b) 1
(c) 2 (d) Either (b) or (c)

Ans (*a*) 0

- *Fill up the blanks*

Q.8 Hypertrichosis of pinna is a ..... trait. **[Textbook]**

Ans holandric

- *Answer in one word or line*

Q.9 Who proposed the genic balance theory? **[Textbook]**

Ans Calvin Bridges

Q.10 Which protein is in *sry* gene of Y-chromosome? **[Textbook]**

Ans TDF (Testis Determining Factor)

Q.11 What is freemartin? **[Textbook]**

Ans Sterile female mammal with masculinised behaviour and non-functioning ovaries.

2½ MARK *Questions*

Exams' Question

Q.12 What is Barr body? **[Textbook, 2009]**

Ans A Barr body is a small darkly stained mass of X-chromosome, which is inactive and is found only in the female cells. Out of the two X-chromosomes in females, only one is functional and the other remains as Barr body.

Important Questions

- *Write brief notes on the following (within 50 words each)*

Q.13 Haplo-diploidy mechanism of sex-determination. **[Textbook]**

Ans Refer to text on page no. 87.

Q.14 Genic balance theory. **[Textbook]**

Ans Refer to text on page no. 87.

Q.15 Freemartin. **[Textbook]**

Ans Refer to text on page no. 87.

Q.16 Single gene effect. **[Textbook]**

Ans Refer to text on page no. 87.

Q.17 Sex reversal. **[Textbook]**

Ans Refer to text on page no. 87.

Q.18 Temperature dependent sex-determination. **[Textbook]**

Ans Refer to text on page no. 88.

Q.19 Chemotactic sex-determination. **[Textbook]**

Ans Refer to text on page no. 87.

3½ MARK *Questions*

Important Questions

Q.20 Differentiate between autosomes and allosomes. **[Textbook]**

Ans Differences between autosomes and allosomes are as follows

Autosomes	Allosomes
These are somatic chromosomes which control the body character or somatic characters.	These are sex chromosomes which determine the sex of an individual.
In humans, out of the total 23 pairs of chromosomes, 22 pairs are autosomes.	In humans, the 23rd pair of chromosome is called sex chromosome.

Q.21 Differentiate between X-chromosome and Y-chromosome. **[Textbook]**

Ans Differences between X and Y-chromosomes are as follows

X-chromosome	Y-chromosome
It is a sex chromosome that occurs in paired in the female and single in the male.	It is a sex chromosome that normally occurs in single form in the male only.
It contains gene for female sex-determination.	It contains gene for male sex-determination.
It does not contain the *sry* gene.	It contains the *sry* gene.

Q.22 Differentiate between superfemale and supermale. **[Textbook]**

Ans Differences between superfemales and supermales are as follows

Superfemales	Supermales
Such individuals have 47(44 + XXX), 48(44 + XXX) chromosomes.	Such individuals have 47(44+YYY) chromosomes.
These females have abnormal sexual development and are mentally retarded.	These males are characterised by abnormal height, mental retardation.

Q.23 Differentiate between sex differentiation and sex reversal. **[Textbook]**

Ans Differences between sex differentiation and sex reversal are as follows

Sex differentiation	Sex reversal
It is the process of differentiation of an undifferentiated zygote either into a male or into a female under the influence of hormones and embryonal conditions.	It is the phenomenon of development of secondary sexual characters of the opposite sex.
It is induced by specific genes by hormones and by anatomy.	It is induced by some chemicals.
Castration and ovariectomy does not influence the process of sex differentiation in animals.	Castration or ovariectomy results in sex reversal in animals.

Q.24 Differentiate between gynandromorph and freemartin. **[Textbook]**

Ans Differences between gynandromorph and freemartin are as follows

Gynandromorph	Freemartin
It is the phenomenon in which half of the body part exhibits female characters, while the other half part exhibits male characters, e.g. *Drosophila*.	In cattle, when twins of the opposite sex are born, the male is normal but the female is sterile with many male characters. Such sterile females are called freemartin,
These develop due to failure of segregation of X-chromosomes at cleavage.	These develop due to influence of male hormone.

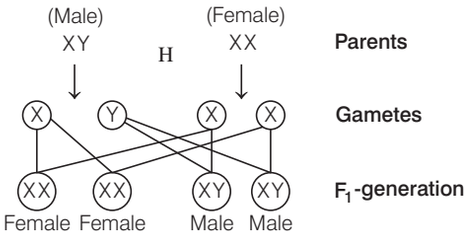
7 MARK *Questions*

Exams' Questions

Q.25 Describe the chromosomal basis of sex-determination in human, honeybee and bird. **[2018]**

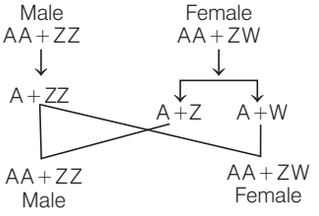
Ans Chromosomal basis of sex-determination in human, honeybee and birds is as follows

(i) **Sex-Determination in Humans**
In males (XY), 50% of sperms carry X-chromosomes and the remaining 50% carry Y-chromosomes. In females (XX), all ova carry X-chromosomes. The sex of an individual is determined by the type of sperms which fertilise the ovum. If the ovum is fertilised by Y-chromosome, the zygote (XY) develops into a male and if the ovum is fertilised by X-chromosome, zygote (XX) develops into a female.



(ii) **In Honeybee**
Haploid-diploid method In this method, the unfertilised egg develops into male (Arrhenotoky) while fertilised egg develops into female. This type of sex-determination is the characteristic feature of insects like honeybees, ants, etc.

(iii) **In Birds**
It is the ZZ-ZW type of sex-determination method commonly seen in birds, fowls and fishes. In this type, females carry one Z and one W-chromosome along with autosomes, whereas males have a pair of Z-chromosome.



Determination of sex in birds

Thus, the sex of an organism is determined by the type of ovum that is fertilised to produce offspring. In this type of sex-determination, total number of chromosomes is same in both males and females. But females produce two different types of gametes and are thus known as heterogametes.

Q.26 Discuss the chromosomal theory of sex-determination in animals. **[2016]**

Ans **Chromosomal Theory of Sex-Determination**
The five main methods of sex-determination by chromosomes are as follows

(i) **XX-XY method** In this mechanism, half of the male gametes carry X and the other half carry Y-chromosomes. The females have a pair of X-chromosomes, e.g. mammals (humans), *Drosophila melanogaster,* etc.

(ii) **XX-XO method** In this method, there is difference in number of chromosomes in two sexes with 'O' indicating the absence of X-chromosome. The female has a pair X-chromosomes and produces homogametic eggs while male has only one X-chromosome (XO) and produces two types of sperms, with X and without X.

(iii) **ZZ-ZW method** This type of sex-determination is seen commonly in birds, fowls and fishes. In this type, females have one Z and one W-chromosome along with autosomes (heterogametic), whereas males have a pair of Z-chromosomes (homogametic).

(iv) **ZO-ZZ method** In this type, the female is heterogametic having only one Z-chromosome, while the male is homogametic having a pair of Z-chromosomes, e.g. moths and certain butterflies.

(v) **Haploid-diploid method** In this method, the unfertilised egg develops into male while fertilised egg develops into female. This type of sex-determination is the characteristic feature of insects like honeybees, ants, etc.

Important Questions

Q.27 What is genic balance theory and explain its role in sex-determination?

Ans Refer to text on page no. 87.

TOPIC TEST 1

- *Choose the correct option (Q. 1 and 2)*

1. ZZ/ZW type of sex-determination is seen in
(a) peacock (b) snails
(c) cockroach (d) platypus

2. The non-homologous segment of Y-chromosome carries
(a) dominant genes (b) holandric genes
(c) recessive genes (d) None of these

[***Ans.*** 1. (a), 2. (b)]

3. Mary F Lyon discovered X-chromosome in male bug and described it as X-body. [***Ans.*** Barr body]

4. In grasshopper, the female is XX and the male is ..... . [***Ans.*** XO]

5. Name the environmental factor that determines sex in *Bonellia*. [***Ans.*** Chemotactic substance]

6. Differentiate between male and female heterogamy.

7. Explain why it is scientifically incorrect to blame the mother for bearing female child.

8. Explain the sex-determination mechanism in humans. How it is different in birds?

9. Which one of the two, sperm or ovum determines the sex of the offspring in fowl? Justify your answer.

10. Describe the different types of sex-determination in insects.

TOPIC ~02 Sex-Linked Inheritance and Chromosomal Disorders

Sex-Linked Inheritance

In addition to sex genes, organisms also contain genes to control other body characters and thus are called **sex-linked genes**. The somatic characters whose genes are located on sex chromosomes are known as **sex-linked characters**. The inheritance of a trait (phenotype) that is determined by a gene located on one of the sex chromosome is called **sex-linked inheritance**.

Sex-Linked Genes

The sex-linked genes are of the following types

(i) **X-linked genes** These are sex-linked genes which lie on X-chromosomes, e.g. genes for colour blindness and haemophilia. These X-linked traits have a unique mode of inheritance as females have two doses of X-linked genes, while males have only one. Thus, males are hemizygous for X-linked traits they possess only half the number of X-chromosomes a female possess. An X-linked gene can be dominant or recessive due to which a female can be a heterozygous carrier of X-linked trait.

(ii) **Y-linked genes** These are sex-linked genes, which are inherited straight from father to son or male to male, e.g. genes for hypertrichosis. Any gene which occurs exclusively on Y-chromosome is said to be **holandric** and it shows holandric inheritance pattern.

(iii) **Pseudoautosomal genes** These genes are located on homologous parts of both X and Y-chromosomes.

Inheritance of Sex-linked Characters

The alleles for sex-linked traits are recessive to their normal alleles. These alleles express themselves in males, i.e. in heterogametic condition whereas in females, they express themselves only in homozygous condition (X^CX^C).

A male transmits his (X-linked) traits to his grandson through her daughter whereas, a female passes her traits to granddaughter through her son. This pattern of inheritance where a trait skips a generation or *criss-crosses* the F_1-generation while passing the trait to F_2 is known as ***criss-cross*** **inheritance**.

Inheritance of Haemophilia

It is a sex-linked recessive disease, which is transmitted from an unaffected carrier female to some of the male offspring. Due to this, patient continues to bleed even during a minor injury because of defective blood coagulation and hence, it is also called as **bleeders disease.**

The gene for haemophilia is located on X-chromosome and it is recessive to its normal allele. In this disease, a single protein that is part of cascade of proteins involved in blood clotting is affected. The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and father should be haemophilic, e.g. females suffer from this disease only in homozygous condition, i.e. X^CX^C.

The haemophilic alleles show *criss-cross* inheritance and they follow Mendelian pattern of inheritance. The **family pedigree of Queen Victoria** (who was a carrier of haemophilia) shows a number of haemophilic individuals.

The inheritance is explained below

(a) **Normal mother and haemophilic father** All the daughters will be carrier of the recessive allele and all the sons will be normal.

(b) **Haemophilic mother and normal father** All the daughters will be carrier and the sons will haemophilic.

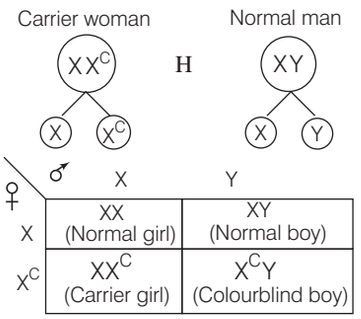
(c) **Carrier mother and normal father** Half of the daughters will be carrier and remaining will be normal. While half of the sons would be haemophilic and remaining will be normal.

(d) **Carrier mother and haemophilic father** Half of the daughters as well as son will be haemophilic while remaining half of the daughter will be carriers, on the other hand remaining half of the sons will be normal.

Inheritance of Red-Green Colour Blindness

It is a sex-linked recessive disorder, which results in defect in either red or green cone of eye. It does not mean the incapability to see any colour at all, infact it leads to the failure in discrimination between red and green colour. The gene for colour blindness is present on X-chromosome.

It is present mostly in males (X^CY) because of the presence of only one X-chromosome as compared to two chromosomes in females. A heterozygous female has normal vision, but is a carrier and passes on the disorder to some of her sons. Its inheritance pattern is similar to that of haemophilia.



Mendelian Disorders in Humans

Under normal conditions, a normal gene encodes normal proteins which then regulate normal physiological functions of our body. But when a gene is mutated, abnormal proteins are formed due to which body functions of our body gets disturbed.

The expression of these characters is known as **genetic disorders** or **syndrome**. These are inherited in simple Mendelian fashion.

For example, Phenylketonuria (PKU), an autosomal (somatic) recessive mutation on chromosome 12. Some other Mendelian genetic disorders are discussed below in detail.

Thalassemia

It is an autosomal recessive blood disease, which is caused due to reduced synthesis of either the α or β-chains of haemoglobin, as a result of mutation in the genes of α or β-chains.

Anaemia is the characteristic symptom of this disease. Depending upon the globin chain affected, thalassemia is classified into following types, i.e.

(i) **α-Thalassemia** is caused by defective α-chain. The α-globin is controlled by two genes present on chromosome 16, i.e. HBA1 and HBA2. α-thalassemia is of two types–haemoglobin H-disease and hydrops foetalis. The later is more severe as all the four globin genes are mutated and the defective alleles kill the foetus resulting in still birth or death soon after delivery.

Haemoglobin H-disease occurs when there are three defective alleles out of four α-globin genes.

(ii) **β-Thalassemia** is caused by decreased synthesis of β-globin chain. It is further classified into **thalassemia major** (Cooley's anaemia), i.e. when both the alleles for β-globin are defective or absent. It is more severe in comparison to the second type called **thalassemia intermedia**, i.e. when only one allele is defective in β-globin.

Symptoms, Diagnosis and Treatment

The common symptoms include fatigue, pale skin with severe anaemia, enlarged spleen, yellowish skin and dark urine. The disease is diagnosed by blood test and genetic analysis. There are two treatment options, i.e. blood transfusion and bone marrow transplantation.

Chromosomal Disorders in Humans

These genetic disorders are caused due to the absence or excess or abnormal arrangement of one or more chromosomes. The inheritance of these disorders is not like those of Mendelian traits.

Some of these disorders are as follows

1. Down's Syndrome (Mongolism)

This syndrome was previously called mongolism because the affected persons were of short stature. It was described by **J Langdon Down** in 1866 but its cause was found by Lejeune in 1959.

Genetic Basis

It occurs due to chromosomal aberration, known as **aneuploidy** (trisomy). The individuals suffering from Down's syndrome possesses an extrachromosome number 21. Both the chromosomes of 21 position passes into a single egg due to primary non-disjunction which may occur during meiosis-I or II in maturation phase of gametogenesis. Thus, the egg instead of possessing 23 chromosomes have 24 chromosomes and the offspring has 47 chromosomes (45 + XY in males, 45 +XX in females). It is also seen in chimpanzees and other related primates.

Symptoms

- Individuals are short statured with small, round head and furrowed tongue and partially open mouth.
- Palm is broad with characteristic simian palm crease, hyperflexible joints.
- Slow mental, physical and psychomotor development.

Diagnosis, Treatment and Prevention

Prenatal screening of the pregnant women is undertaken by ultrasonography. Also amniocentesis sampling is done to ensure the absence of this disorder.

2. Turner's Syndrome

This condition is characterised by one missing X-chromosome which result in 45 + XO chromosomal complement in affected person. It was first described by H.H. Turner in 1938.

Genetic Basis

It is a disorder which is caused due to chromosomal aberration, known as aneuploidy (monosomy). Due to absence of one of the X-chromosome, the condition is 45 with XO. Primary non-disjunction in either of meiotic divisions during gametogenesis results in this condition.

Symptoms

- Affected females are sterile as ovaries are rudimentary.
- Lack of secondary sexual characters and poor breast's development.
- Short stature, small uterus, puffy fingers, webbed neck, lowset ears, etc.

Diagnosis, Treatment and Prevention

Diagnosis is done by physical examination and genetic analysis. Turner syndrome affected subjects undergo hormonal therapy. Growth hormone injection in early childhood may increase the height by few inches. Oestrogen replacement therapy is undertaken at puberty to start the breast development. Oestrogen and progesterone are administered together a little later to initiate the monthly cycle. Turner's syndrome affected persons have a shorter life expectancy.

Superfemales or Poly X Females

These females have 47 (44 + XXX), 48 (44 + XXXX) or 49 (44 + XXXXX) chromosomes. Such individuals show abnormal sexual development and mental retardation.

Supermales or Jacobs Syndrome

These individuals have 47(44 + XYY) chromosomes. Males have characteristic abnormal tall height, criminal bent of mind and mental retardation. Due to overproduction of male sex hormone, they are more aggressive than normal males.

3. Klinefelter's Syndrome

HF Klinefelter first described this condition in 1942. This genetic disorder occurs due to the presence of an additional copy of the X-chromosome. It is also known as trisomy of X-chromosome. Its estimated birth frequency is 1/500 live male births.

Genetic Basis

The union of an abnormal XX-egg with a normal Y-sperm or a normal X-egg with an abnormal XY-sperms results in the karyotype of 47, XXY in males or 47, XXX in females.

The abnormal eggs and sperms are formed due to the primary non-disjunction of X and Y-chromosomes during the maturation phase of gametogenesis. Although the usual karyotype of this condition is 47 + XXY but sometimes more complex karyotypes also occurs, e.g. XXXY, XXXXY, XXXXXY, XXXXYY, etc.

Symptoms

- Individuals have masculine development but feminine characters like development of breasts (gynaecomastia), etc.
- Poor beard growth and feminine pitched voice.
- Such unclear individuals are sterile.

Diagnosis, Treatment and Prevention

Chromosome complement examination is used to diagnose the syndrome. However, there is no treatment available for affected individuals.

PRACTICE QUESTIONS

Exams', Textbook's Other Imp. Questions

1 MARK *Questions*

Exams' Questions

- *Choose the correct option*

Q.1 What is the diploid chromosome number in a person suffering from Down syndrome? **[2019]**

(a) 45 (b) 46 (c) 47 (d) 48

Ans (c) 47

- *Answer the following questions in one word or one sentence*

Q.2 The genotype of a carrier haemophilia is $\underline{X^hX^h}$. **[2014]**

Ans XX^h

Q.3 In which chromosome, the gene for haemophilia is located? **[2009]**

(X-chromosome, Y-chromosome, autosome, Both X-chromosome and Y-chromosome)

Ans X-chromosome

Important Questions

- *Choose the correct option*

Q.4 Which one is a sex-linked disorder? **[Textbook]**

(a) Leukemia (b) Cancer
(c) Night blindness (d) Colour blindness

Ans (d) Colour blindness

Q.5 A haemophilic man marries a normal homozygous woman. What is the probability that their son will be haemophilic? **[Textbook]**

(a) 100% (b) 75% (c) 50% (d) 0%

Ans (d) 0%

Q.6 What is the probability that their daughter will be haemophilic? **[Textbook]**

(a) 100% (b) 75% (c) 50% (d) 0%

Ans (d) 0%

Q.7 The gene responsible for haemophilia is linked to which chromosome? **[Textbook]**

(a) X (b) Y
(c) Both X and Y (d) Autosome

Ans (a) X-chromosome

Q.8 Red-green colour blindness in man is **[Textbook]**

(a) sex-linked character
(b) sex-limited character
(c) sex influenced character
(d) sexual character

Ans (a) sex-linked character

Q.9 Sex-linked characters are **[Textbook]**

(a) dominant (b) recessive
(c) lethal (d) not inherited

Ans (b) recessive

Q.10 In birds, which type of chromosomal basis of sex-determination is present? **[Textbook]**

(a) XX-XY (b) XX-XO
(c) ZW-ZZ (d) ZZ-ZO

Ans (c) ZW-ZZ

Q.11 In a person with Turner syndrome, the number of X-chromosome is **[Textbook]**

(a) 1 (b) 2
(c) 3 (d) 0

Ans (a) 1

Q.12 A Down syndrome will be **[Textbook]**

(a) 45 + XX (b) 44 + XY
(c) 44 + XXY (d) 22 + XY

Ans (a) 45 + XX

Q.13 A colourblind person cannot distinguish **[Textbook]**

(a) all colours (b) green
(c) red (d) red and green

Ans (d) red and green

Q.14 Number of Barr bodies present in Turner's syndrome is **[Textbook]**

(a) 0 (b) 1
(c) 2 (d) Both (b) and (c)

Ans (a) 0

- *Answer the following by filling the blank*

Q.15 ..... is the karyotype of a male suffering from Klinefelter's syndrome.

Ans 47 + XXY.

• *Answer the following questions in one word or one sentence*

Q.16 Name two sex-linked diseases of human being. **[Textbook]**

Ans Haemophilia and colour blindness.

Q.17 How Down's syndrome is caused? **[Textbook]**

Ans Down's syndrome is caused due to the presence of an extrachromosome number 21, i.e. 21 trisomy.

Q.18 In which chromosome is the gene for haemophilia located? **[Textbook]**

Ans X-chromosome

Q.19 What is the chromosomal complement for Turner's syndrome? **[Textbook]**

Ans 44 + XO

Q.20 Which sex is usually a carrier? **[Textbook]**

Ans Female sex

Q.21 In which chromosome, the factors for haemophilia and colour blindness are found? **[Textbook]**

Ans X-chromosome

Q.22 What is the other name of Bleeder's disease? **[Textbook]**

Ans Haemophilia

Q.23 Which type of defect is found in thalassemia?

Ans The mutation or deletion of the genes controlling the formation of globin chains of haemoglobin results in an abnormal form of haemoglobin.

Q.24 Who first described Klinefelter's syndrome? **[Textbook]**

Ans H. F. Klinefelter in 1942.

Q.25 Which Mendelian disorder is known as royal disease? **[Textbook]**

Ans Haemophilia is known as royal disease.

2½ MARK *Questions*

Exams' Questions

Q.26 What is *criss-cross* inheritance? **[Textbook, 2016]**

Ans *Criss-cross* inheritance is defined as the inheritance of sex-linked characters transmitted from father to daughter and from mother to son who pass it on to grandsons and granddaughters, respectively. The trait is expressed only in alternate generations, e.g. red-green colour blindness, haemophilia, etc.

Q.27 Describe the symptoms of Down's syndrome. **[2015]**

Ans The symptoms of Down's syndrome are

(i) Short stature
(ii) Small round head
(iii) Furrowed tongue
(iv) Partially open mouth
(v) Broad palm with characteristic palm crease
(vi) Slow mental, physical and psychomotor development.

Q.28 Write short note on inheritance of colour blindness in man. **[2014]**

Or

Write short note on colour blindness. **[2009, 2008]**

Ans Colour blindness is a recessive sex-linked trait in which an individual fails to distinguish between red and green colours. The recessive allele is carried by X-chromosomes. In female, it appears only when both the sex chromosomes carry the gene (X^CX^C). The females function as carriers in the presence of a single recessive gene (XX^C) while in males the defect appears in the presence of a single recessive gene (X^CY) because Y-chromosome does not carry any gene for colour vision.

Q.29 Write short note on sex-linked inheritance. **[2002]**

Ans Sex chromosomes carry genes for both determination of sex and controlling body characters. The body characters whose genes are located on sex chromosomes are called sex-linked characters. The genes which control sex-linked characters are called sex-linked genes.

The mode of inheritance of sex-linked genes is called sex-linked inheritance.

Important Questions

Q.30 The human male never passes on the gene for haemophilia to his son. Why?

Ans The gene responsible for haemophilia is located on X-chromosome and males have only a single copy of X-chromosome with no alternative normal allele. Thus, the father passes only Y-chromosome to his son and not the haemophilic X-chromosome.

Therefore, in this way, the human male can never pass on the gene for haemophilia to his son.

Q.31 What are the various causes of human genetic disorders?

Ans The various causes of human genetic disorders are as follows

(i) **Mendelian disorders** are caused due to the alteration or mutation in as single gene.
(ii) **Chromosomal disorders** are caused due to the excess, absence or abnormal arrangement of one or more chromosomes.
(iii) Failure of segregation of chromatids during cell division also results in the gain or loss of a chromosome called **aneuploidy**.
(iv) Failure of cytokinesis after telophase stage results in an increase in a whole set of chromosomes called **polyploidy**.

3½ MARK *Questions*

Exams' Questions

Q.32 Write short note on Turner's syndrome. **[Textbook, 2019]**

Ans **Turner's syndrome** This condition is characterised by one missing X-chromosome which results in 45 + XO chromsomal complement in affected person. It is produced by the union of an abnormal O egg and a normal X sperm or a normal egg and an abnormal O sperm. The individual has 45 chromosome (4 + X) instead of 46. Such people are sterile females who have rudimentary ovaries undeveloped breasts, short stature, small uterus and abnormal intelligence. They moved ovulate or menstruate. One in every 3000 children is a victim.

Q.33 What is sex-linked inheritance? Discuss how sex-linked gene inheritance occurs in human, giving two examples. **[2019]**

Ans Refer to text on page no. 91.

Q.34 Write short notes on the following. **[Textbook, 2018]**
(a) Down's syndrome (b) Thalassemia

Ans (a) **Down's syndrome** It is associated with the trisomy of 21st chromosome. It was described by **J Langdon Down** in 1886. The person suffering from this have broad forehead, short and broad neck, short and stubby fingers, a simian line in the palm and a wide gap between first and second fingers in feet.

The person also suffers from severe mental retardation because of malformation of central nervous system. The patient has 47 chromosomes, (i.e. an extra 21st chromosome). Trisomy arises due to the non-disjunction (non-separation) of 21st chromosome during oogenesis. Thus, the egg has 24 chromosomes instead of 23. Fertilisation of such egg with a normal sperm results in trisomy of 21st chromosome.

(b) **Thalassemia** It is an inherited blood disorder in which an abnormal form of haemoglobin is formed in the body. Thalassemia commonly occurs in three forms, i.e. α-thalassemia, β-thalassemia and thalassemia minor, α-thalassemia is caused by a pair of defective alleles regulating the synthesis of α-haemoglobin. It is further classified into two types haemoglobin-H disease and hydrops fetalis.

The latter is more severe and expressed when all four globin genes are mutated. The affected babies are either stillborn or die shortly after birth.

Haemoglobin-H disease occurs when 3 out of 4 α-globin genes are mutated.

β-thalassemia has 2 subtypes Coeley's anaemia (thalassemia major) and thalassemia intermedia.

Thalassemia major is more severe while thalassemia intermedia is less severe. Thalassemia major is expressed when 2β-globin genes are absent are mutated. It occurs when body is unable to produce β-globin.

Important Questions

Q.35 Differentiate between Down's syndrome and Turner's syndrome. **[Textbook]**

Ans Differences between Down's syndrome and Turner's syndrome are as follows

Down's syndrome	**Turner's syndrome**
It occurs due to the presence of an additional copy of the chromosome number 21. This condition is called trisomy of 21 chromosome.	It is a disorder caused due to the absence of one of the X-chromosomes, i.e. 44 with XO.
It is an autosomal genetic disorder.	It is a sex-linked chromosomal genetic disorder.

Q.36 How would you distinguish between Turner's syndrome and Klinefelter's syndrome?

Ans Differences between Turner's and Klinefelter's syndromes are

Turner's syndrome	**Klinefelter's syndrome**
It occurs due to the absence of one of the X-chromosomes in females.	It occurs due to the presence of an additional copy of X-chromosome in males.
It occurs mainly in females.	It occurs mainly in males.
Chromosome complement is 44 + XO.	Chromosome complement is 44 + XXY.
Stature is short, chest is broad with undeveloped breasts.	Stature is long due to long limb bones, chest is broad with or without enlarged breasts.
The presence of reduced ovaries.	The presence of reduced testes.
The absence of menstrual cycle.	Sterility occurs.

7 MARK *Questions*

Exams' Questions

Q.37 What is sex-linked inheritance? Discuss the inheritance of haemophilia in man. **[2015, 2013]**

Ans **Sex-linked Inheritance** Sex chromosomes contain genes that control the sex of an organism.

In addition to sex genes, these organisms also contain the genes to control body characters in sex chromosomes called sex-linked genes.

Inheritance of sex-linked genes is called sex-linked inheritance.

There are three types of sex-linked genes

(i) **X-linked genes** Sex-linked genes lie on X-chromosomes, e.g. colour blindness and haemophilia in man, eye colour in *Drosophila*.

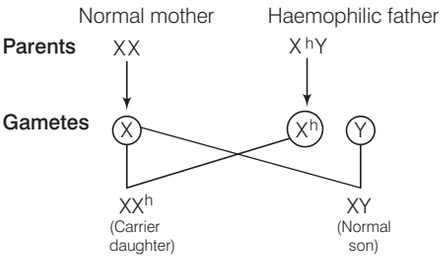
(ii) **Y-linked genes** (holandric genes) Sex-linked genes lie on Y-chromosomes only, e.g. hypertrichosis.

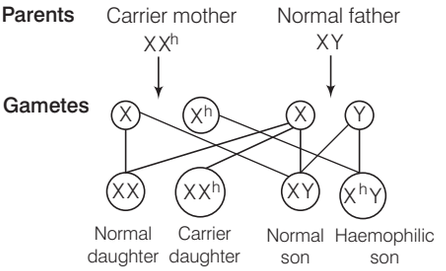
(iii) **X and Y-linked genes** Sex-linked genes lie on homologous part of the X and Y-chromosomes, e.g. total colour blindness and nephritis.

Haemophilia (Bleeder's disease) This is a sex-linked disease, where the patient will continue to bleed due to a minor cut in the body, as he or she cannot synthesise anti-haemophilic globulin (haemophilia-A) or plasma thromboplastin (haemophilia-B, also called Christmas disease) required for normal blood clotting.

It is caused by a recessive gene X^h located on the sex chromosome. Females have two X-chromosomes.For them to be haemophilic, it is necessary that each X-chromosome has a gene for haemophilia (X^hX^h).

The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least a carrier and father should be haemophilic.





Such females are never born as the combination of these two recessive genes is lethal (fatal).

A female having only one gene for haemophilia (XX^h), appears normal because its dominant allele in the X-chromosome produces the factor required for normal blood clotting. Such females are called **carriers**. Such heterozygous females may transmit this disease to her sons.

In males, a single gene for the defect is also to express itself as the Y-chromosome carries no gene for this trait.

Hence, haemophilia occurs only in males.

Q.38 What is sex-linked inheritance? Discuss this by taking colour blindness as an example.

[Textbook 2011]

Ans Refer to text on page no. 91 and 92.

Important Questions

Q.39 Discuss in detail about chromosomal disorders.

Ans Refer to text on page no. 92 and 93.

TOPIC TEST 2

1. A haemophilic man marries a normal homozygous woman. What is the probability that their son will be haemophilic?

(a) 100% (b) 50%

(c) 75% (d) 0% [***Ans.*** (d)]

2. An additional copy of X-chromosome causes..... .

[***Ans.*** Klinefelter's syndrome]

3. Name the scientist who discovered Down's syndrome.

[***Ans.*** J Langdon Down]

4. Give possible genotypes of the parents who can give birth to haemophilic daughter.

5. What do you understand by Y-linked genes?

6. What is thalassemia? Explain its types.

7. How chromosomal disorder differ from Mendelian disorder?

8. What are Mendelian disorders?

9. What are the causes and symptoms of Turner's syndrome in an individual?

10. Discuss the types of chromosomal disorders.

11. How are X-linked traits different from Y-linked traits?

12. How are genetic disorders broadly classified? Differentiate between them. Explain by giving one example for each.

Chapter Test

1 MARK *Questions*

- *Answer the following questions in one word or one sentence (Ques 1 to 3)*

1 What is the chromosomal complement of a female with Down's syndrome?

2 What would be the sex of the child developed from 44A + XX?

3 Name one autosomal recessive disease.
[**Ans.** 1. 45 + XX, 2. Female, 3. Thalassemia]

- *Choose the correct option (Ques 4 to 5)*

4 If father shows normal genotype and mother shows a carrier trait for haemophilia then
(a) all the female offspring will be normal
(b) all the female offspring will be carriers
(c) a male offspring has 50% chance of active disease
(d) a female offspring has probability of 50% to have active disease

5 The syndrome in humans in which individual contains the three sex chromosomes XXY is called
(a) Superfemale (b) Turner's syndrome (c) Down's syndrome (d) Klinefelter's syndrome
[**Ans.** 4. (a), 5. (d)]

- *Fill up the blanks (Ques 6 to 9)*

6 The gene for colour blindness is situated on ……chromosome.

7 The recessive X- linked disease for haemophilia shows characteristic ..... like colour blindness.

8 Humans like other mammals have a sex-determination mechanism that depends on the presence or absence of ..... .

9 Turner's syndrome is ..... disorder.
[**Ans.** 6. X, 7. *criss-cross* inheritance, 8. Y-chromosome, 9. chromosomal]

2½ MARK *Questions*

10 Give symptoms of the following disorders
(a) Colour blindness (b) Klinefelter's syndrome.

11 Write the sex chromosome constitution in male individual of human, bird, grasshopper and butterfly.

12 Describe sex-determination in fruitfly with example.

13 Why is the possibility of a human female suffering from haemophilia rare? Explain.

14 A colourblind child is born to a normal couple. Work out a cross to show how it is possible. Mention the sex of this child.

15 Why grasshopper and *Drosophila* show male heterogamety? Explain.

3½ MARK *Questions*

16 Differentiate between male heterozygosity and female heterozygosity.

17 Differentiate between genic balance theory and single gene effect of sex-determination.

7 MARK *Questions*

18 Differentiate between XX-XY mechanism and XX-XO mechanism.

19 Explain the causes, inheritance pattern and symptoms of any two Mendelian genetic disorders.