11. Chromosomal Basis of Inheritance

11.0 : Introduction

Q.1. Define the terms.

Ans:i) Heredity : The inheritance or transmission of characters from one generation to the next generation is called heredity.

ii) Variations : The differences existing between parents and their offsprings along with their siblings are called variations.

iii) Genetics : The scientific study of mechanism of heredity and causes of variation is called genetics.

Q.2. What is plasmid ?

Ans: Circular DNA found on prokaryotic cell and capable of independent existence is called plasmid.

11.1 : The Chromosomal Theory

Q.3. Who had proposed the chromosomal theory of inheritance?

Ans: Chromosomal theory of inheritance was proposed by Sutton and Boveri.

Q.4. Explain chromosomal theory of inheritance.

OR

Give important points of the chromosomal theory of inheritance.

Ans: Chromosomal theory of inheritance was proposed by Sutton and Boveri. This theory includes following points:

It states that :

i) Gametes (sperm and egg) carry all the hereditary characters. They are the link between parents and offspring.

ii) Nucleus of gametes contains chromosomes which carry all the hereditary characters.

iii) Chromosomes are found in pairs in somatic or diploid cells.

iv) During gamete formation homologous chromosomes pair and segregate or separate independently at meiosis. Thus each gamete contains only one chromosome of a pair.

v) During fertilization, the union of sperm and egg restores the diploid number of chromosomes

11.2 : Chromosomes

Q.5. Define Chromosome.

Ans: At the time of cell division, the chromatin threads condense to form distinct threads called chromosomes.

Q.6. How many chromosomes does each human diploid cell have ?

Ans: A human diploid cell has 46 chromosomes or 23 pairs of chromosomes.

Q.7. How many autosomes are there in a human haploid cell like sperm or ovum?

Ans: In a human haploid cell like sperm or ovum there are 22 autosomes.

Q.8. What do you mean by homologous chromosomes ?

Ans: Chromosomes which are morphologically and genetically identical are called homologous chromosomes.

Q.9. How many sex chromosomes are there in a human male diploid cell ?

Ans: In a human male diploid cell there are two sex chromosomes the, X chromosome and the Y chromosome.

Q.10. How many sex chromosomes are there in a human female diploid cell?

Ans: In a human female diploid cell there are two sex chromosomes, they are two copies of the X chromosome (XX).

Q.11. In which phase of cell division chromosomes appear short and thick? Why ?

Ans: In metaphase chromosomes appear short and thick because they get condensed during metaphase.

Q.12.Explain the concept of chromosome.

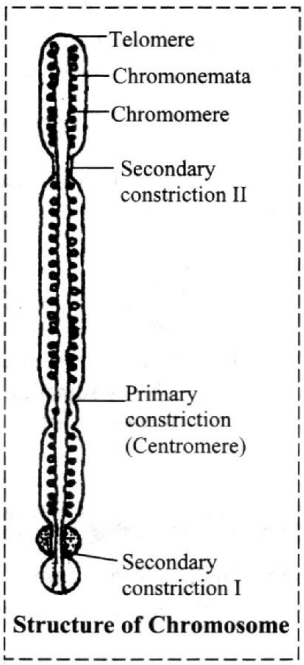
Ans: Concept of chromosome:

i) The chromosomes are filamentous bodies inside the nucleus.
ii) Chromosomes are made up of two identical halves called chromatids.
iii) They become distinct during cell division.
iv) Chromosomes are capable of self duplication.
v) They play vital role in heredity, mutation, variation, evolution etc.
vi) The number of chromosomes is constant and specific for every species.
vii) e.g. man in each diploid cell has 46 Chromosomes, Ascaris has 2 chromosomes, Frog has 26, Pigeon has 80 and Chimpanzee has 48.

Q.13.With help of neat, labelled diagram, describe structure of chromosome.

Ans:Structure:

i) A metaphasic chromosome has two identical halves called sister chromatids.
ii) Each chromatid is inturn made up of sub-chromatids called chromonemata (singular- chromonema).
iii) The chromatids lie side by side and are held together at one point called the centromere. The centromere is also called the primary constriction.
iv) During cell division the spindle fibres are attached at the centromere. Besides the primary constriction, additional narrow areas called secondary constrictions are present.
vi) Some of the secondary constrictions are called nucleolar organizers because they are necessary for formation of nucleolus.
vii) The part of the chromosome beyond the nucleolar organizer is short, spherical and is called satellite.
viii) The tip of the chromosome is called telomere.
ix) It has a unique property in that it prevents the ends of the chromosomes from sticking together but attaches to the nuclear envelope.
x) The surface of a chromosome bears number of small swellings called Structure of Chromosome chromomeres. Target Publications Pvt. Ltd.



Structure of Chromosome

Q.14.Which secondary constrictions are called nucleolar organizers?

Ans: Secondary constrictions which help in formation of nucleolus are called as nucleolar organizers.

Q.15.What is telomere ?

Ans: Tip of the chromosome is called telomere.

Q.16.Describe various types of chromosomes.

Ans: Chromosomes can be classified into different types according to :

A) Characters they govern
B) Position of the centromere

A) Types of chromosomes based on characters they govern: (A1) sex chromosomes & (A2) autosomes. (A1) Sex chromosomes

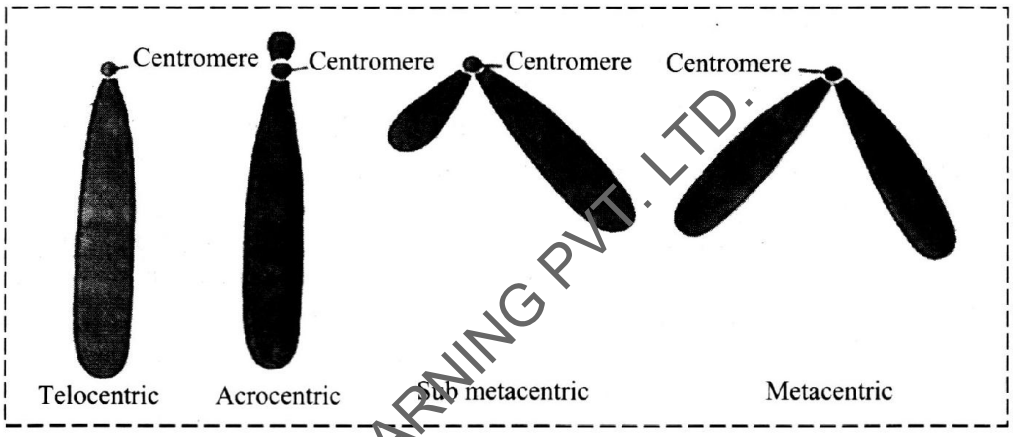
i) These are usually one pair in a diploid cell.
ii) They may be homologous X and X or heterologous i.e. X and Y. Homologous sex chromosomes are morphologically and functionally similar but Heterosomes or Heterologous sex chromosomes are morphologically and functionally different.
iii) They control the characters of sex (development of gonads)

(A2) Autosomes

i) Autosomes are chromosomes that are not involved in determining the sex of an organism.
ii) They are also responsible for the transmission of somatic characters from one generation to the preceding generation.
iii) In humans, there are 22 pairs of autosomes in both sexes.

B) Position of the centromere

There are four types of chromosomes with regard to position of centromere.



(B1) Metacentric :
Centromere is at **middle** of chromosome and hence arms are equal.
Chromosome looks **V-shaped** in anaphase.

(B2) Submetacentric :
Centromere is not in the center but near centre of chromosomes and therefore arms are unequal in length. The chromosome appears **J or L shaped** in anaphase.

(B3) Acrocentric :
Centromere is near one end of the chromosome and hence arms are very unequal.

(B4) Telocentric :
Centromere is at extreme tip of the chromosome. The arm is only on one side. Chromosome remains rod shaped in even anaphase. Chromosome appears **'i' shaped**

Q.17.Describe the structure and types of chromosomes.
Ans: *Refer Q. 13 and Q. 16.*

Q.18.Mention different types of chromosomes according to position of centromere.
Ans: *Refer Q. 16.*

Q.19. Enlist functions of chromosomes.
Ans:i) Chromosomes carry genes which determine hereditary characters.
ii) Genes on a chromosome are carried together as a linkage group to the next generation.
iii) Qualitative and quantitative changes in chromosomes are called mutations which leads to 'increased' or 'decreased' reproductive fitness of individuals and population.
iv) They help in cell division and cell growth.
v) They control cell metabolism by directing the synthesis of enzymes. (protein synthesis).
vi) Sex chromosomes are responsible for determination of sex.

Q.20. Give the significance of "X" chromosome.
Ans:i) It is one of the sex chromosomes responsible for sex determination.
ii) It determines femaleness in human beings.
iii) It determines maleness in birds.
iv) It contains more genetic information and hence controls more functions.
v) It controls certain sex linked characters which shows "criss- cross" inheritance.

Q.21. Give the significance of "Y" chromosome.
Ans:i) It is one of the sex chromosomes responsible for sex determination.
ii) In humans, it is responsible for maleness.
iii) In birds Y chromosome is responsible for femaleness.
iv) In humans, it carries holandric genes. It expresses only in males.
v) It contains less genetic information and hence controls less functions.

Q.22. Distinguish between Xand Y Chromosome. **[Mar 2014]**
Ans:

No.	X Chromosome	Y Chromosome
i)	These chromosomes are metacentric, hence appear X shaped.	These chromosomes are acrocentric, hence appear Y shaped.
ii)	They are straight rod like and larger than 'V' chromosomes.	They are straight rod like or hooked and bend at tip.
iii)	It contains large amount of euchromatin and small amount of heterochromatin.	It contains large amount of heterochromatin and small amount of euchromatin.
iv)	Found in both male and females.	Found only in males.
v)	Non-homologous part shows more genes than Y chromosome.	Non-homologous part contains few genes as compared to X chromosome.
vi)	They are responsible for X-linked character.	They are responsible for Y-linked character.
vii)	Genes present on X chromosome show criss-cross inheritance.	Genes present on Y chromosome shows straight inheritance.

11.3 : Linkage and Crossing Over

Q.23.Define linkage.

Ans: The tendency of the genes on the same chromosome to link together is called linkage.

Q.24.What are linked genes ?

Ans: Genes that are located on same chromosome and linked together are called linked gene.

Q.25. Write a note on linkage.

OR

What is linkage? Explain the types of linkages.

Ans : Linkage: It may be defined "as the tendency of genes to remain together and pass on to next generation together". The genes remain tied together through several generations. The alleles which are situated on the same homologous chromosomes are said to be linked but these transmit together during the formation of gametes.

Types of linkages:

There are two types of linkage:

i) Complete linkage **ii) Incomplete linkage.**

i) Complete linkage :

a) Linkage is said to be complete when the linked genes do not allow for any crossing over, but are always inherited together only parental combinations of genes/traits are observed in the progeny.

ii) Incomplete linkage :

a) Linkage is incomplete when the linked genes allow for crossing over at time (but not always); so, the frequency of recombination between such genes in a test cross progeny is less than 50 percent

Q.26. What is synapsis ?

Ans: The homologous chromosomes come close to each other and form a pair. This pairing is called synapsis.

Q.27. How can a pair of linked genes be identified ?

Ans: Linked genes are found on one pair of chromosomes forming one linkage group.

Q.28. Distinguish between Incomplete sex linked gene and complete sex linked gene.

Ans:

No.	Incomplete sex linked gene	Complete sex linked gene
i)	These genes are located on homologous region of X and Y chromosomes.	These are located on non-homologous region of X and Y chromosomes.
ii)	They do not inherit together.	They inherit together.
iii)	Crossing over may occur in this region.	Crossing over does not occur in this region.
iv)	e.g. Nephritis, total colour blindness, Retinitis pigmentosa.	e.g. Red green, Colour blindness, haemophilia, myopia.

Q.29.Which process is antagonistic to linkage?

Ans: Crossing over is the process antagonistic to linkage.

Q.30.Explain Morgan's experiment of linkage and recombination.

Ans:i) Morgan carried out dihybrid crosses in *Drosophila* to study sex linked gene.

ii) He hybridised yellow bodied, white eyed female to brown bodied red eyed males.

iii) He intercrossed their F_1 progeny.

iv) He observed that two genes did not segregate independently of each other and he got the 9 : 3 : 3 :1 ratio.

v) Morgan concluded that genes were located on X chromosome. When two genes in a dihybrid cross were located on the same chromosome, the parental gene combinations were much higher than nonparental type.

vi) He also concluded that when genes are grouped on the same chromosome, some genes were tightly linked while others are loosely linked.

vii) Genes white (w) and yellow (y) were tightly linked and showed 1.3% recombination.

viii) Cross between white (w) and miniature wing (m) showed 37.2% recombination.

Q.31.Describe two methods by which parental genes may form new combination.

Ans: New combinations are produced due to :

i) Crossing over : It is the exchange of genetic segments with adjacent paternal and maternal chromatids.

ii) Mutation: These are the sudden inheritable changes.

Q.32. In which stage of cell division, crossing over takes place ?

Ans: In prophase -I of meiosis I, crossing over occurs.

Q.33.What is crossing over ?

Ans: Crossing over can be defined as the reciprocal exchange of equivalent segments between non-sister chromatids of homologus chromosomes.

Q.34.Explain the mechanism of crossing over.

OR

Write a note on crossing over.

Ans: i) There are four steps

a) Synapsis b) Tetrad formation c) Crossing over d) Disjunction

ii) Crossing over involves breaking and rejoining of DNA.

iii) During synapsis, a characteristic ladder like scaffold called synaptonemal complex aligns the DNA molecules of two homologus chromosomes side by side.

iv) Consequently DNA strand of one chromatid is able to pair with the corresponding DNA strand of other chromatid, where base sequences match.

v) Breaking and synthesis of one DNA strand using strand of the other chromatid DNA as the template results in crossing over.

11.4 : Linked Inheritance

Q.35.What are sex-linked genes ?

Ans: Genes located on non-homologous region of sex chromosomes but not involved in sex determination are called sex linked genes.

Q.36.What is sex-linkage?

Ans: The phenomenon of inheritance of sex linked gene is called sex linkage.

Q.37.Why do sex linked traits appear more in males than in females? [Mar 2014]

Ans: Sex linked traits appear more in males than in females because in females two recessive sex linked genes are required for expression of gene but in males single recessive gene can express itself.

Q.38.Mention any two sex-linked disorders.

Ans: i) Colourblindness and 2. Haemophilia

Q.39.Write a note on sex-linked inheritance.

Ans: i) Sex linked inheritance means inheritance of characters whose genes are located on the sex chromosomes.

ii) There are two types: X-linked or Sex-linked genes and V-linked or Holandric genes.

iii) **X-linked or Sex-linked genes:** Genes located on non-homologous region of X-chromosome only are called X-linked or sex-linked genes. The X-linked genes have no corresponding allele in Ychromosome. In females, two recessive sex-linked genes are required for the expression of sex-linked genes and in males, single recessive gene can express itself.

iv) **Y-linked or Holandric genes:** Genes located on non-homologous region of Y-chromosome only are called V-linked or holandric genes. They inherit along with V-chromosome expressed only in males. e.g. Hypertrichosis.

Q.40. The haemophilic male never passes the gene for haemophilia to his son. Why ?

Ans:i) Haemophilic gene is present on X-chromosome.

ii) In haemophilic male genetic constitution is X^hY.

iii) In X-linked disease, in males only one haemophilic gene on X-chromosome is sufficient to produce disease. So the male having haemophilic gene will be suffering from disease.

iv) Males suffering from this defect may bleed to death due to small cut and rarely survive to attain marriageable age. This disease is expressed in males but is transmitted by females.

Q.41. Explain the pattern of inheritance of haemophilia in humans. Why is the possibility of a human female becoming a haemophilic extremely rare ? Explain.

OR

Why is haemophilia generaUy observed in human males? Explain.

OR

What is haemophilia? Explain its inheritance with the help of suitable charts.

Ans:i) **Haemophilia :** It is a Mendelian disorder caused by a recessive allele in humans.

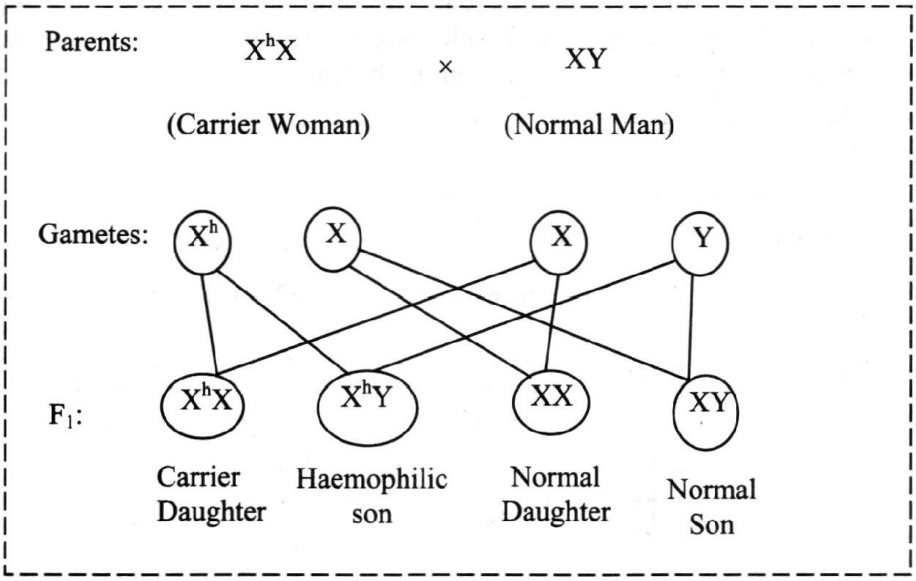
ii) Haemophilic person shows deficiency of clotting factors in blood so minor injuries cause continous bleeding.

iii) The genes for this disease (haemophilia) are located on X-chromosomes.

iv) The males usually suffer from this disease because the recessive gene is present on X-chromosomes. The corresponding gene on Y is not present. So the males suffer from this disease even when one gene of haemophilia is present.

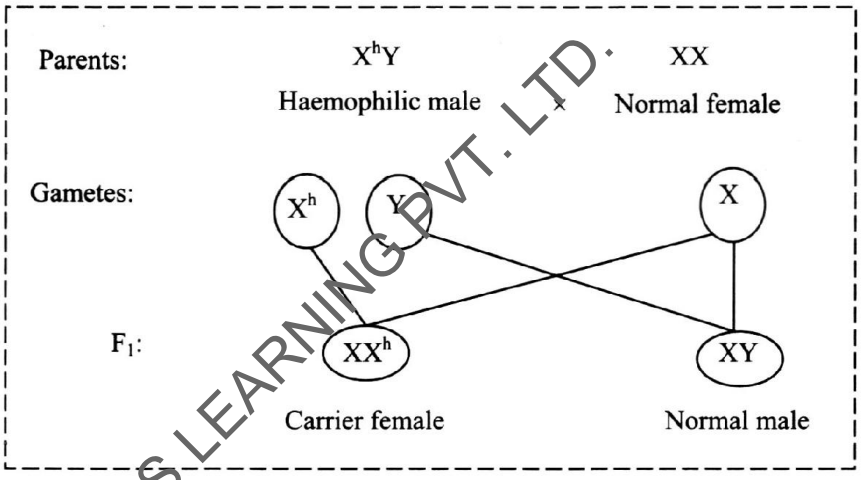
v) The females usually do not suffer from this disease as the recessive gene is present on one of the two X's. They become haemophilic when both the X-chromosomes have recessive genes.

i) When carrier woman marries normal man :

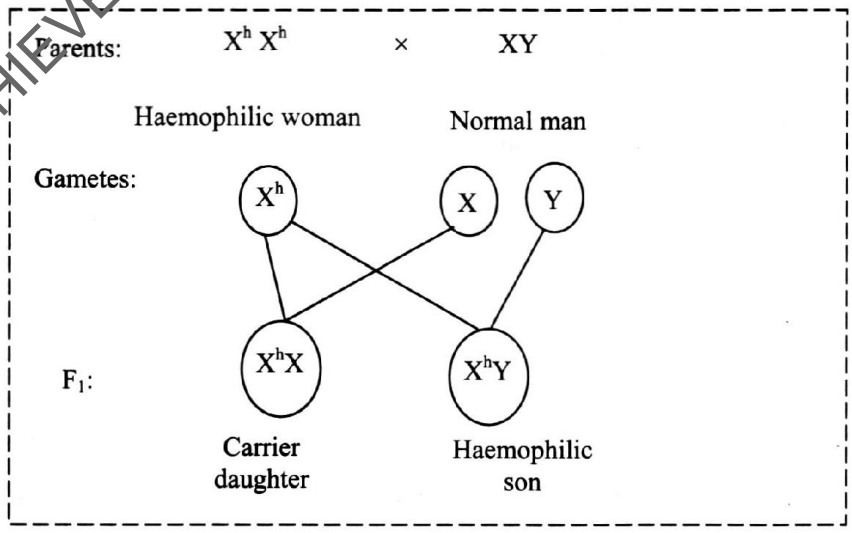


ii) If a haemophilic male (X^hY) marries a female with normal clotting of blood (XX) then all the

offsprings will have normal clotting of blood. The sons will have normal clotting of blood but daughters will be carriers for the disease. The carriers have normal clotting of blood.

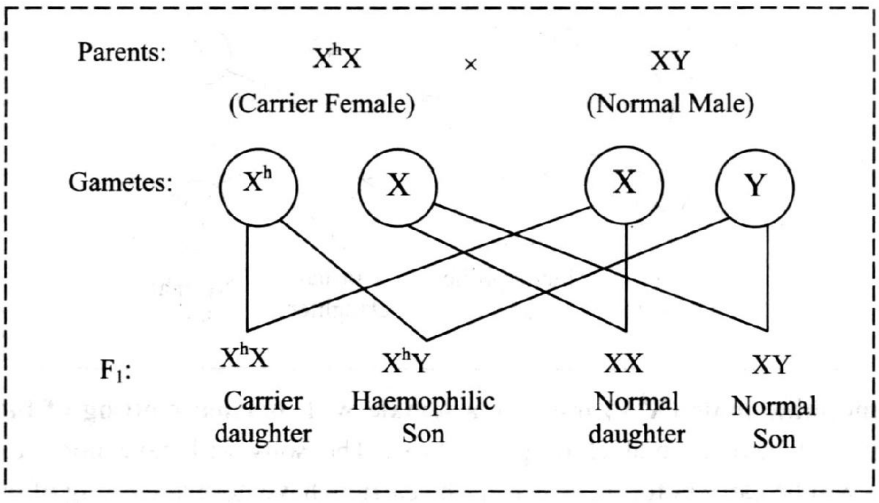


iii) When haemophilic woman marries normal man :



Q.42. A haemophilic son was born to normal parents. Give the genotypes of the parents.

Ans: Heterozygous female (carrier) for haemophilic can transmit the disease to her sons. The genotypes of parents will be X^hX and XY.In this case the female parent will carry gene for haemophilia(X^hX) but will not express disease in her due to presence of another X chromosome. She will be carrier of the disease. Thus genotype of haemophilic son whose mother is carrier of haemophilia and father is normal, can be represented as follows :



Q.43.Why the son of a carrier mother and a normal father may be sufferer of haemophilia whereas the son of haemophilic father and normal mother would not be? Under what circumstances it is possible for both the father and the son to be the sufferer from this trait ?

Ans: Due to heterozygous nature of males, son receives one X-chromosome from mother. So, in first case son will be normal. Son of a carrier mother and normal father may be sufferer of haemophilia due to possible genetic combination of Xhy (Xh from mother and Y from father). When father is haemophilic and mother is haemophilic carrier, then son will be haemophilic.

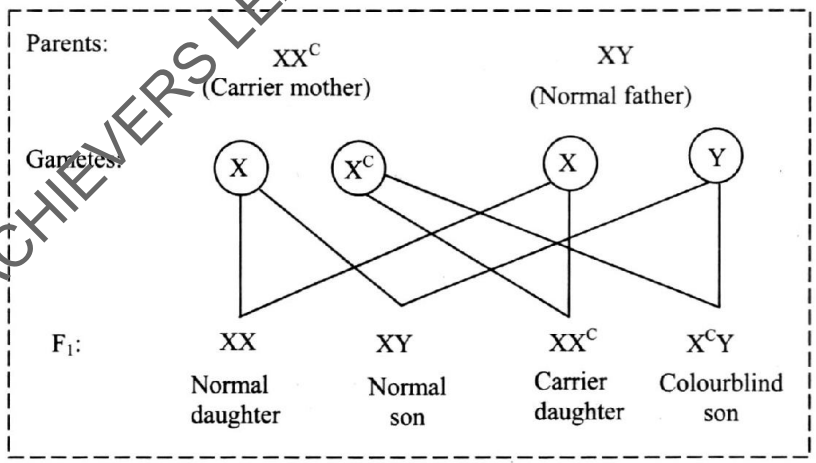
Q.44.What is criss-cross inheritance?

Ans: The inheritance of characters from the father to his grandson through his daughter is called criss-cross inheritance.

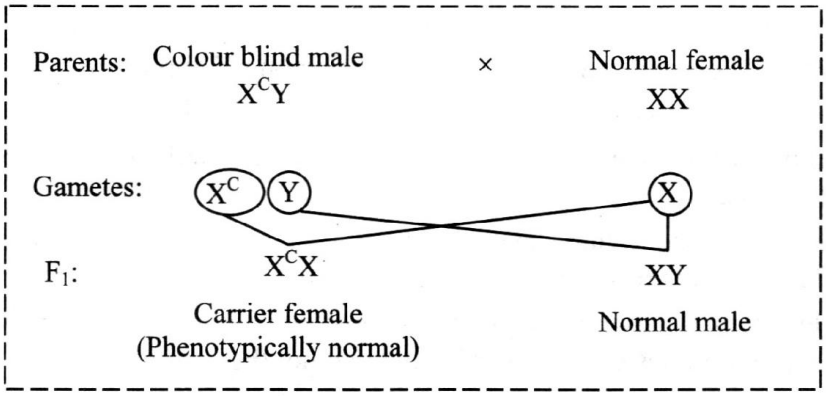
Q.45. What is colour blindness? Explain its inheritance with the help of suitable charts.

Ans:i) Colour blindness is a defect in which person cannot differentiate between colours.

ii) Colour blindness genes are found on X chromosome. When a normal man marries a carrier colour blind woman, one of their sons may be colour blind.



iii) If a colour blind male (X^CY) marries a female with normal vision (XX) then all the offspring will have normal vision. The sons will have normal vision but daughters will be carriers for the disease. The carriers have normal vision.



Q46.What is sex linkage ? Explain the inheritance of colour blindness and haemophilia with suitable charts.

Ans: *Refer Q.36, 41 and 45.* **[Mar 2013]**

11.5 : Sex determination

In human being

Q.47. Define the terms homogametic and heterogametic.

Ans: i) Homogametic: It means production of gametes containing only one kind of sex chromosome, as in the human female.

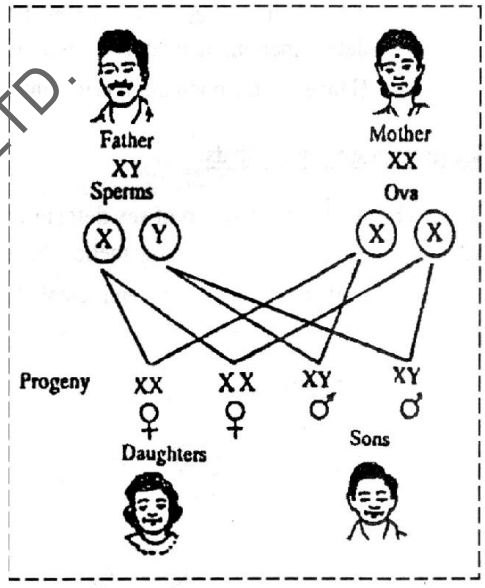
ii) Heterogametic: Producing dissimilar gametes, such as those of human males, who produce two types of spermatozoa, one bearing the X-chromosome and the other bearing the Y-chromosome.

Q.48.Write a note on sex determination in human.

Ans:Sex Determination in human :

Human diploid cell has 46 chromosomes i.e. 23 pairs in each cell. Out of these, 22 pairs of chromosomes are called autosomes. Autosomes determine all body characters like colour of hair, skin, colour of eyes, height etc. Out of 23 pairs, one pair of chromosomes is called sex chromosomes. They are X and Y chromosomes. Every individual gets one set of chromosomes from his mother and one from his father.

A human **male thus has 44 +XY** chromosomes whereas Progeny **a female has 44 + XX** chromosomes. During gamete formation; meiosis or reduction division takes place and a gamete gets only one set of chromosomes and thus it is haploid. e.g. Female gamete (ovum) 22 + X Male gamete (sperm) 22 + X or 22 + Y. When the male and female gametes unite to form a zygote the chromosomes again become diploid. Thus the offspring gets the same number of chromosomes as his parents. Since which sperm (X or Y) fertilizes Sex of baby is determined by the nature of sperm (X or Y) that fertilizes ovum. Thus in human, male determines sex of the baby.



Q.49. Write a note on social significance of sex determination.

Ans:i) Women produce ova, each ovum contains 23 chromosomes: 22 autosomes and one X chromosome. That is all ova are of only one type so far as numbers and types of chromosomes they bear.

ii) A male produces sperms. They are of two types

a) X sperm has 23 chromosomes: 22 autosomes and one X chromosome.

b) Y sperm has 23 chromosomes: 22 auto somes and one Y chromosome.

iii) If X sperm fertilizes the ovum, zygote gets 44 autosomes and two X chromosomes. This zygote grows into a female baby.

iv) If Y sperm fertilizes the ovum, zygote gets 44 autosomes, one X chromosome and one Y chromosome. This zygote grows into a male baby.

v) Which sperm fertilizes the ovum decides the fate of zygote, whether to grow into a male or female baby. Thus father determines sex of his baby and and not the mother.

vi) In our society, females are blamed for giving birth to a baby girl. However, sex determination shows that, it is the male who is responsible for the sex of the foetus.

Q.50. Explain PNDT Act of 1996. Why it was required ?

Ans:i) PNDT act is Prenatal Diagnostic Technique Act.

ii) Act was made because medical techniques were developed by which sex determination can be done.

iii) It was misused by the people for female foeticide.

Q.51. What is sex ratio ?

Ans: Sex ratio is the number of female per 1000 males.

Q.52. Why female population is declining in India ?

Ans:i) In India due to many practices such as Dowry male child is preferred.

ii) Due to this many girls are killed after birth. Besides this after development of prenatal sex determination technique many people does this test and if it is female child is born then it is aborted.

iii) Though it is illegal practice as PNDT act - 1996 has banned use of medical technique for sex determination, many doctors and their patients violate the law.

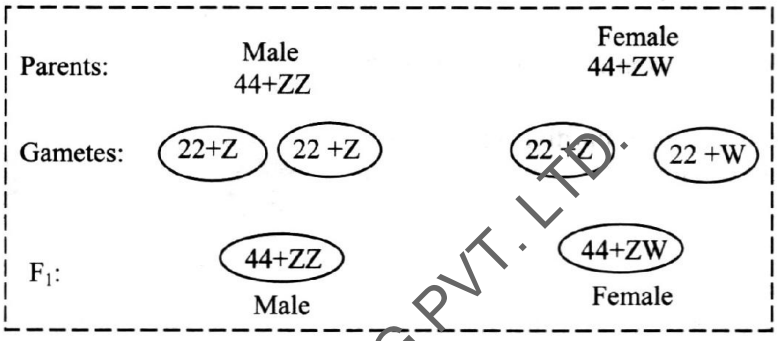
iv. Hence female population is declining in India.

<u>Sex determination in Birds</u>

Q.53. What is the mechanism of sex determination in birds ?

Ans:i) In birds male is homogametic where as female is heterogametic.

ii) Male has genotype ZZ and female has genotype ZW.



iii) Sperms carry Z chromosome but egg carries Z and W chromosome approximately in equal number.
iv) Sex of the zygote depends on which egg gets fertilized.

Q.54. Explain sex-determination in human being and birds.
Ans: *Refer Q. 48 & 53.*

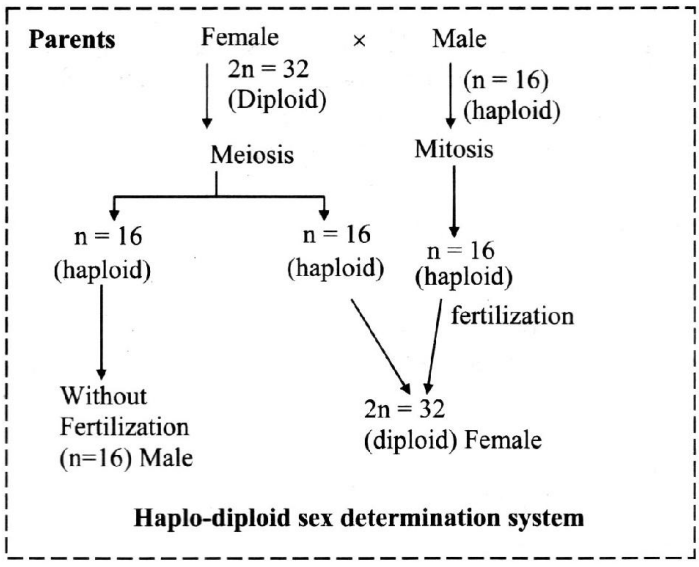
Sex determination in Honeybee

Q.55.With the help of a chart, explain the method of sex determination in honeybees. [Mar 2014]

OR

How sex is determined in honeybee ?

Ans:i) In honeybee, haplodiploid sex-determination system determines the sex of the offspring.
ii) In this system sex is determined by the number of sets of chromosomes an individual receives.
iii) An offspring developed from the union of sperm and an egg develops as a female and unfertilized egg develops into male.
iv) After copulation queen store sperms from a male in a seminal receptacle within her body and remain available for fertilization when eggs are laid by her.
v) When she lays egg she has control for release of stored sperm from within the organ.
vi) It she release sperm when an egg passes down the oviduct, the egg is fertilized and it develops into female. Unfertilized eggs develops into males i.e. drones.



Haplo-diploid sex determination system

11.6 : Mendelian Disorders

Q.56.Write a note on thalassemia.

Ans: i) Haemoglobin is a conjugated protein consisting of A and B chain. Each chain consists of α and β subunits.
ii) It is a group of autosomal inherited disorder characterized by decreased synthesis of either α or β globin chain of HbA.
iii) Beta and alpha thalassemia is caused by deficient synthesis of ~ and a subunits respectively.

iv) The a globin chain is coded by a gene on chromosome 16 and the gene for ~ globin chain is located on chromosome 11.
v) It results in deficiency of one chain and the relative excess of the other chain.
vi) Clinically, person suffers from anaemia (inability to synthesize Hb), Jaundice, variation in size and shape ofRBCs.
vii) Massive Blood transfusion is needed (Massive blood transfusion-the need to transfuse one or two times the patient's normal blood volume).
viii) In a normal adult, this is equivalent to 10-20 units. So thalassemia is a quantitative abnormalities of polypeptide globin chain synthesis.

11.7 : Chromosomal disorders

Q.57. What is syndrome ?
Ans: Appearance of different types of symptoms is called syndrome.

Q.58. Define aneuploidy.
Ans: Addition or deletion of one or two chromosomes in diploid chromosome number is called aneuploidy.

Q.59. Write a short note on - Down's syndrome.
Ans:i) Down's syndrome (trisomy 21) is caused due to aneuploidy i.e. addition or deletion of one or two chromosome in diploid chromosome number.
ii) It was first described by **John Langdon Down in 1866.**
iii) In this genetic disorder, the patient carries 2n + 1 (47) chromosomes, instead of normal 2n (46 chromosomes).
iv) The extra chromosome is present with 21SI chromosome pair, thus 21SI pair has three chromosomes instead of usually two chromosomes.
v) 2n + 1 condition is known as trisomy.
vi) Trisomy occurs due to non disjunction of chromosome during anaphase.
vii) Down's syndrome is characterised by :
a) The person has small, round head with flat back and straight and sparse hair.
b) The face is broad, flat and is moon like with standing eyes, flat nose and malformed ears.
c) They show prominent folding at the comer of the eyes.
d) They have small and arched palate, protruding tongue that causes the mouth to remain partially open.
e) The hands and feet are short and broad with short fmgers and toes. Fingers show many loops and tips.
f) Severe mental retardation is often seen.
g) Sexual maturity and fertility is rarely attained.
h) Person with Down's syndrome has short life.

Sex chromosomal abnormalities:

Q.60.Name two sex chromosomal abnormalities.
Ans: Klinefelter's syndrome and Turner's syndrome are two sex chromosomal abnormalities.

Q.61.Write a short note on - Turner's syndrome.
Ans: Turner's syndrome (Sterile female) :
i) It was first described by H.H. Turner.
ii) Turner's syndrome is caused by non disjunction of XX chromosomes due to monosomy i.e. loss of one chromosome from the pair i.e. 2n - 1.
iii) The sufferer possesses 45 chromosomes instead of 46.
iv) When an abnormal egg without any X chromosome is fertilized by a sperm with X chromosome, the resulting baby contains XO chromosomes.i.e. 44XO.
v) **Symptoms :**
a) The baby is sterile and short stature.

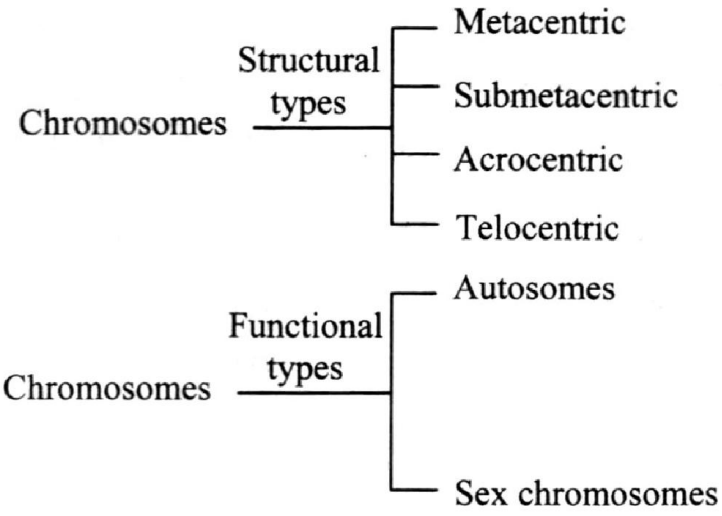
b) She is normal phenotypically but no menstruation at puberty.
c) Ovaries do not develop into a functional tissue.
d) Female sex hormones secretion is low.
e) The chest is broad and breasts are poorly developed.
f) She is mentally retarded.
g) Body height is less.
h) Webbing of skin at neck. i.e. folds of skin are seen.
i) Uterus is very small.

Q.62. Write a short note on - Klinefelter's syndrome.

Ans: Klinefelter's syndrome:

i) Klinefelter's syndrome is genetic abnormality in human male.
ii) It is caused by trisomy (2n + 1) due to chromosomal aberration.
iii) The individual has 47 chromosomes instead of 46 and chromosomal make up will be 44A + XXY.
iv) When abnormal egg with XX chromosomes is fertilized with normal Y sperm, the resulting baby gets XXY.
v) Symptoms:
a) They are sterile males.
b) Testes are small and there is no spermatogenesis.
c) Amount of male hormone secreted is low.
d) Breasts may be enlarged.
e) Late puberty with reduced secondary sexual characteristics.
f) Penis is normal-sized.
g) In Klinefelter's syndrome patients, Diabetes mellitus and goitre incidence are more common.
h) Also various cancers may be more prevalent.

Quick Review



Additional information

Sutton and Boveri : Proposed chromosomal theory of inheritance.

Hofmeister : Discovered chromosome.

Waldeyer : He coined the term chromosome

Bateson and Punnet (1905): Discovered linkage in sweet pea

Bateson → Father of modem genetics.

John Lagdon Down (1866): First discovered Down's syndrome.

Multiple Choice Question

1. Chromosomal theory of inheritance was proposed by
   a) Bridges b) Morgan
   c) Neil stevens d) Sutton and Boveri
2. Telomeres are
   a) ends of chromosome
   b) additional constriction of chromosome
   c) regions of highly folded DNA
   d) none of these
3. If the centromere is situated at the tip of the chromosome it is called
   a) Metacentric b) Acrocentric
   c) Telocentric d) Submetacentric
4. _____chromosome appears 'V' shaped during anaphase.
   a) Metacentric b) Acrocentric
   c) Telocentric d) Sub-metacentric
5. The sister chromatids are held together by
   a) Centrioles b) Chromomere
   c) Chromonemata d) Centromere
6. Chromosomes do not have well defined structure in
   a) interphase b) prophase
   c) metaphase d) telophase
7. Number of autosomes in human sperm are
   a) 23 b) 44
   c) 46 d) 22
8. A submetacentric chromosome has
   a) only one arm b) 'V' shaped arms
   c) 'L' shaped arms d) very unequal arms
9. Which of the following is not a part of the chromosome ?
   a) Centromere b) Spindle fibre
   c) Chromo mere d) Telomere
10. Shape of the chromosome is determined by the position of
    a) centromere b) chromomere
    c) telomere d) all of these
11. Two homologous chromosomes are connected at
    a) telomere b) chromomere
    c) centromere d) centriole
12. The pairing of homologous chromosomes is called **[Oct 2013]**
    a) crossing over b) terminalization
    c) synapsis d) bivalent
13. How many linkage groups are present in man ?
    a) 23 linkage groups
    b) 46 linkage groups
    c) 22 linkage groups
    d) 44 linkage groups
14. _____ are located on homologous region of X and Y chromosomes.
    a) Completely linked genes
    b) Incompletely linked genes
    c) Completely sex linked genes
    d) Incompletely sex linked genes
15. Crossing over takes place during which of the following stage?
    a) Diplotene b) Pachytene
    c) Leptotene d) Diakinesis
16. The genes present on non - homologous part of Y - chromosome are
    a) Holandric genes
    b) Sex - linked genes
    c) X - linked genes
    d) Gynaec genes
17. Which of the following trait is never observed in human females? [Mar 2014]
    a) Hypertrichosis
    b) Haemophilia
    c) Colour blindness
    d) Myopia
18. Colour blindness is a
    a) Deficiency disease
    b) X-linked disease
    c) Y-linked disease
    d) XY- linked disease
19. Sex-linked genes are present on
    a) Homologous region of sex chromosomes
    b) Non-homologous region of auto somes
    c) Homologous region of autosomes
    d) Non-homologous region of X chromosome
20. Which of the following is not X-linked disorder?
    a) Haemophilia b) Hypertrichosis
    c) Night blindness d) Myopia
21. Which of the following is also called bleeder's disease ?
    a) Anaemia b) Thrombocytopenia
    c) Polycythemia d) Haemophilia
22. Daughter of a colour-blind father and normal mother marries a colour blind person. Colourblindness in the progeny will be
    a) 50% sons and 50% daughters
    b) All sons and daughters
    c) All daughters
    d) All sons

23. A marriage between normal visioned man and colour blind woman will produce
a) normal son and carrier daughter
b) colour blind sons and carrier daughters
c) carrier sons and carrier daughters
d) colour blind sons and daughters normal.

24. Sex chromosomes in human male are
a) XX b) XO
c) XY d) XZ

25. Sex determination in human being is
a) XY-XX type
b) XX-XO type
c) XX-XY type
d) XO-XX typ-e

26. Sex of the child is determined by
a) female chromosome
b) male chromosome
c) size of the sperm
d) autosome

27. X -chromosome of father is transmitted to
a) son
b) daughter
c) grandson
d) none, of these

28. In human sex, determination factor is
a) Y Chromosome
b) X Chromosome
c) Both a) and b)
d) None of the above

29. The sex determination pattern in honeybee is called
a) female haploidy b) haplodiploidy
c) gametic diploidy d) gametogony

30. In Down's syndrome each cell has how many chromosomes?
a) 21st pair having one less
b) 23rd pair having one less
c) 45
d) 47

31. Genetic constitution of Klinefelter's syndrome is
a) 44 + XXY b) 44 + XO
c) 42 + XXXX d) 44 + XX

32. Turner's syndrome is
a) XO b) XY
c) XY d) XYY

33. The cause of Turner's syndrome in man is
a) incomplete sex linkage
b) sex-linked inheritance
c) autosomal abnormality
d) sex--chromosomal abnormality

34. XXY chromosome compliment is found in
a) Down's syndrome
b) Turner's syndrome
c) Klinefelter's syndrome
d) Edward's syndrome

35. The person with Turner's syndrome has
a) 45 autosomes and X sex chromosome
b) 44 autosomes and XYY sex chromosomes
c) 45 autosomes and X sex chromosomes
d) 44 autosomes and X sex chromosome

36. Webbed neck is characteristic of
a) XXY sex chromosomes
b) XY sex chromosomes
c) XXY sex chromosome
d) XO sex chromosomes

37. Which one of the following is sex chromosomal disorder ?
a) Colourblindness
b) Thalassemia
c) Turner's syndrome
d) Down's syndrome

Answer Keys

1.	d)	2.	a)	3.	c)	4.	a)	5.	d)	6.	a)	7.	d)	8.	d)	9.	b)	10.	a)
11.	c)	12.	c)	13.	a)	14.	d)	15.	b)	16.	a)	17.	a)	18.	b)	19.	d)	20.	b)
21.	d)	22.	a)	23.	b)	24.	c)	25.	c)	26.	b)	27.	b)	28.	a)	29.	b)	30.	d)
31.	a)	32.	a)	33.	d)	34.	c)	35.	d)	36.	d)	37.	c)						