UNIT II

Chapter 4

PRINCIPLES OF INHERITANCE AND VARIATION

CHAPTER SNAPSHOT

Evaluation

1. Haemophilia is more common in males because it is a
(a) Recessive character carried by Y-chromosome
(b) Dominant character carried by Y-chromosome
(c) Dominant trait carried by X-chromosome
(d) Recessive trait carried by X-chromosome
[Ans. (d) Recessive trait carried by X-chromosome]

2. ABO blood group in man is controlled by
(a) Multiple alleles
(b) Lethal genes
(c) Sex linked genes
(d) Y-linked genes **[Ans. (a) Multiple alleles]**

3. Three children of a family have blood groups A, AB and B. What could be the genotypes of their parents?
(a) $I^A I^B$ and ii (b) $I^A I^o$ and $I^B I^o$
(c) $I^B I^B$ and $I^A I^A$ (d) $I^A I^A$ and ii
[Ans. (b) $I^A I^o$ and $I^B I^o$]

4. Which of the following is not correct?
(a) Three or more alleles of a trait in the population are called multiple alleles.
(b) A normal gene undergoes mutations to form many alleles
(c) Multiple alleles map at different loci of a chromosome
(d) A diploid organism has only two alleles out of many in the population.
[Ans. (c) Multiple alleles map at different loci of a chromosome]

5. Which of the following phenotypes in the progeny are possible from the parental combination AxB?
(a) A and B only (b) A,B and AB only
(c) AB only (d) A,B,AB and O
[Ans. (d) A,B,AB and O]

6. Which of the following phenotypes is not possible in the progeny of the parental genotypic combination $I^A I^O$ X $I^A I^B$?
(a) AB (b) O
(c) A (d) B **[Ans. (b) O]**

7. Which of the following is true about Rh factor in the offspring of a parental combination DdXDd (both Rh positive)?
(a) All will be Rh-positive
(b) Half will be Rh positive
(c) About ¾ will be Rh negative
(d) About one fourth will be Rh negative
[Ans. (d) About one fourth will be Rh negative]

8. What can be the blood group of offspring when both parents have AB blood group?
(a) AB only (b) A, B and AB
(c) A, B, AB and O (d) A and B only
[Ans. (b) A, B and AB]

9. If the childs blood group is 'O' and fathers blood group is 'A' and mother's blood group is 'B' the genotype of the parents will be
(a) $I^A I^A$ and $I^B I^o$ (b) $I^A I^o$ and $I^B I^o$
(c) $I^A I^o$ and $I^o I^o$. (d) $I^o I^o$ and $I^B I^B$
[Ans. (b) $I^A I^o$ and $I^B I^o$]

10. XO type of sex determination and XY type of sex determination are examples of.
(a) Male heterogamety
(b) Female heterogamety
(c) Male homogamety
(d) Both (b) and (c)
[Ans. (a) Male heterogamety]

11. In an accident there is great loss of blood and there is no time to analyse the blood group which blood can be safely transferred?
(a) 'O' and Rh negative
(b) 'O' and Rh positive
(c) 'B' and Rh negative
d) 'AB' and Rh positive
[Ans. (a) 'O' and Rh negative]

12. Father of a child is colourblind and mother is carrier for colourblindness, the probability of the child being colour blind is.
(a) 25% (b) 50%
(c) 100% (d) 75%
[Ans. (b) 50%]

13. A marriage between a colourblind man and a normal woman produces
(a) All carrier daughters and normal sons
(b) 50% carrier daughters, 50% normal daughters
(c) 50% colourblind sons, 50% normal sons
d) All carrier offsprings
[Ans. (a) All carrier daughters and normal sons]

14. Mangolism is a genetic disorder which is caused by the presence of an extra chromosome number.
(a) 20 (b) 21 (c) 4 (d) 23
[Ans. (b) 21]

15. Klinefelters' syndrome is characterized by a karyotype of
(a) XYY (b) XO
(c) XXX (d) XXY
[Ans. (d) XXY]

16. Females with Turners' syndrome have
(a) Small uterus
(b) Rudimentary ovaries
(c) Underdeveloped breasts
d) All of these **[Ans. (d) All of these]**

17. Pataus' syndrome is also referred to as
(a) 13-Trisomy (b) 18-Trisomy
(c) 21-Trisomy. (d) None of these
[Ans. (a) 13-Trisomy]

18. Who is the founder of Modern Eugenics movement?
(a) Mendel (b) Darwin
(c) Francis Galton (d) Karl pearson
[Ans. (c) Francis Galton]

19. Improvement of human race by encouraging the healthy persons to marry early and produce large number of children is called
(a) Positive eugenics (b) Negative eugenics
(c) Positive euthenics (d) Positive euphenics
[Ans. (a) Positive eugenics]

20. The _____deals with the control of several inherited human diseases especially inborn errors of metabolism.
(a) Euphenics (b) Eugenics
(c) Euthenics (d) All of these
[Ans. (a) Euphenics]

21. "Universal Donor" and "Universal Recipients" blood group are ___ and ___ respectively.
(a) AB, O (b) O, AB
(c) A, B (d) B, A
[Ans. (b) O, AB]

22. ZW-ZZ system of sex determination occurs in
(a) Fishes (b) Reptiles
(c) Birds (d) All of these
[Ans. (d) All of these]

23. Co-dominant blood group is
(a) A (b) AB (c) B (d) O
[Ans. (b) AB]

24. Which of the following is incorrect regarding ZW-ZZ type of sex determination?
(a) It occurs in birds and some reptiles
(b) Females are homogametic and males are heterogametic
(c) Male produce two types of gametes
(d) It occurs in gypsy moth
[Ans. (b) Females are homogametic and males are heterogametic]

25. What is haplodiploidy?
Ans. (i) It is a system of sex determination in which sex of the offspring is determined by the number of sets of chromosomes it receives.
(ii) **Eg.** Honey bees in which fertilized eggs develop into females (Queen or worker bees) and unfertilized eggs develop into males (drones) by parthenogenesis.
(iii) This means males have half the number of chromosomes (haploid - n) and females have double the number of chromosomes (diploid - $2n$). Hence the method is called haplodiploidy.

26. Distinguish between heterogametic and homogametic sex determination systems?
Ans.

Heterogametic sex determination	Homogametic sex determination
Sex chromosome may be disimilar or heter omorphic in the other sex.	Sex chromosome may be similar or homomorphic in one sex.
Heteromorphic individuals produce two types of gametes and are said to be heterogametic. **2 types** 1) Heterogametic males 2) Heterogametic females.	Individuals having homomorphic sex chromosomes provide only one type of gamete are said to be homogametic.
Eg. Males are the Heterogametic sex as they have one X and one Y chromosome (XY)	**Eg.** Females are the homogametic sex, because they have female 2 X's (XX) chromosomes.

27. What is Lyonisation?

Ans. *(i)* In the XY chromosomal system of sex determination, males have only one X chromosome, whereas females have two. In mammals the necessary dosage compensation is accomplished by the inactivation of one of the X chromosome in females so that both males and females have only one functional X chromosome per cell.

(ii) Mary Lyon suggested that Barr bodies represented an inactive chromosome, which in females becomes tightly coiled into a heterochromatin, a condensed and visible form of chromatin (Lyon's hypothesis). The number of Barr bodies observed in cell was one less than the number of X-Chromosome. XO females have no Barr body, whereas XXY males have one Barr body.

(iii) The discovery of X inactivator is attributed by British Genetician Mary Lyon and is called as Lyonisation.

28. What is criss-cross inheritance?

Ans. It is the transmission of a gene from mother to son or father to daughter. The character is inherited to the second generation through the carrier of first generation. **E.g.** Inheritance of gene causing haemophilia (x linked inheritance).

29. Why are sex linked recessive characters more common in the male human beings?

Ans. *(i)* Male human beings are hemizygous. Their sex chromosomes consist of one X and one Y chromosomes.

(ii) Sex linked character are inherited through genes in sex chromosomes since males inherit one allele only for sex linked character (one X and Y chromosomes) they express the trait commonly.

(iii) In female there are two x chromosome representing the sex chromosomes for each sex linked character. Therefore expression of the character depends on both the alleles. Sex linked inherited traits are more common in males than females because, males are hemizygous and therefore express the trait when they inherit one mutant allele.

30. What are holandric genes?

Ans. The genes present in the differential region of Y chromosome are called Y-linked or holandric genes. The Y- linked genes have no corresponding allele in X chromosome. **Eg:** Hypertrichosis

31. Mention the symptoms of Phenylketonuria.

Ans. It is characterized by severe mental retardation, light pigmentation of skin and hair. Phenylpyruvic acid is excreted in the urine.

32. Mention the symptoms of Downs syndrome.

Ans. It is characterized by severe mental retardation, defective development of the central nervous system, increased separation between the eyes, flattened nose, ears are malformed, mouth is constantly open and the tongue protrudes.

33. Differentiate Intersexes from Supersexes.

Ans. According to geneic balance theory (C. B. Bridges), the sex of an individual is determined by the ratio of X chromosome to that of its autosome sets. This ratio is termed sex index

$$\text{Sex index} = \frac{\text{Number of X Chromosomes}}{\text{Number of Sets of Autosomes}} \left(\frac{X}{A}\right)$$

Change in this ratio leads to a changed sex phenotype.

Super Sex	Inter Sex
Super sexes one of two type namely super females and super males.	These flies are intermediate between normal males and females.
Super females have a chromosome constitution 2A + XXX and the sex ratio is 1.5. These flies have an additional 'X' chromosome than the normal female and are known as metafemales. Super males have a chromosomal constitution 3A + XY and the sex ratio is 0.33. These flies have an additional sets of autosome than the normal male flies and are known as metamales.	Their chromosomal constitution is either 3A + XXY or 3A + XX.
These flies are very weak and sterile in nature.	They have a mixture of male and female sex characters and are sterile is nature.

34. Explain the genetic basis of ABO blood grouping man.

Ans. ABO system of blood grouping in humans is based on the chemical difference due to presence of antigens on the surface of the RBC and epithelial cells as follows:

Blood group	Antigen
A	Presence of A antigen
B	Presence of B antigen
O	absence of A and B antigen
AB	presence of A and B antigen

(i) Bernstein discovered that, the inheritance of different blood group in humans is determined by a number of multiple allelic series.

(ii) The three autosomal alleles located on chromosome 9 are concerned with determination of blood group.

(iii) The gene controlling blood type is labelled as 'L' or I. The gene (isoagglutination) I exists in three allelic forms I^A, I^B and I^o

(iv) I^A specifies A antigen, I^B allele determined B antigen and I^o allele specifies no antigen.

(v) Each allele (IA and IB) produces a transferase enzyme. IA allele produces N-acetyl galactose transferase and can add N-acetyl galactosamine (NAG) and IB allele encodes for the enzyme galactose transferase that adds galactose to the precursor (i.e. H substances) In the case of I^O/I^O allele no terminal transferase enzyme is produced and therefore called "null" allele and hence cannot add NAG or galactose to the precursor.

(vi) I^A and I^B are dominant to I^O but co – dominant to each other (I^A = I^B). The dominance hierarchy is given as (I^A =I^B>I^O).

(vii) A child receives one of three alleles from each parent giving rise to six positive genotypes and four possible blood type (phenotypes)

Hero types	Phenotype
I^AI^A I^AI^o	A group
I^BI^B I^BI^o	B group
I^A I^B	AB group
I^o I^o	O group

Genetic basis of the human ABO blood groups

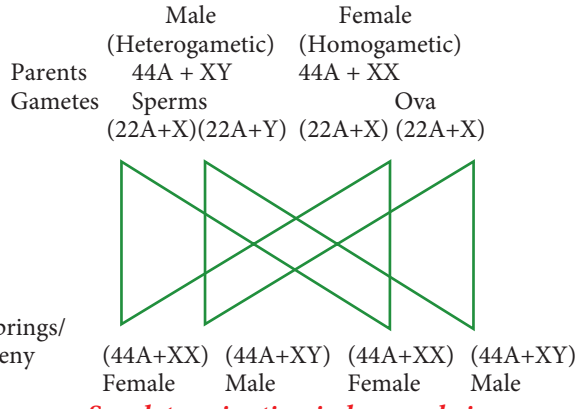
Genotype	ABO blood group phenotype	Antigens present on red blood cell	Antibodies present in blood plasma
I^AI^A	Type A	A	Anti -B
I^AI^o	Type A	A	Anti -B
I^BI^B	Type B	B	Anti -A
I^BI^o	Type B	B	Anti -A
I^AI^B	Type AB	A and B	Neither Anti - A nor Anti-B
I^oI^o	Type O	Neither A nor B	Anti -A and anti -B

35. How is sex determined in human beings?

Ans. In human beings the sex is determined by XX-XY type (Lygaeus type)

Genes determining sex in human beings are located on **two sex chromosomes, called allosomes**. In mammals, sex determination is associated with chromosomal differences between the two sexes, typically XX females and XY males. 23 pairs of human chromosomes include 22 pairs of autosomes (44A) and one pair of sex chromosomes (XX or XY). Females are homogametic producing only one type of gametes (egg), each containing one X chromosome while the males are heterogametic producing two types of sperms with X and Y chromosomes.

The sex of the embryo depends on the fertilizing sperm. An egg fertilized by an 'X' bearing sperm produces a female, if fertilized by a Y bearing sperm a male is produced.



Sex determination in human beings

Additional Questions

Choose the Correct Answer 1 Mark

I. Choose the correct options for the below Questions

1. The blood group _____ is called universal donor.
(a) A (b) AB
(c) B (d) O [*Ans.* (d) O]

2. The blood group _____ is called universal recipient.
(a) AB (b) O
(c) B (d) A
[*Ans.* (b) AB]

3. The ABO blood group was discovered by _____.
(a) Sturli (b) Decastelle
(c) Landsteiner
(d) Alexander wiener [*Ans.* (c) Landsteiner]

4. The inheritance of blood group is determined by multiple alleles as discovered by_____.
(a) Landsteiner (b) Bernstein
(c) Alexander castelle (d) lyon
[*Ans.* (b) Bernstein]

5. The _____ is called null allele.
(a) I^A (b) $I^O I^B$
(c) I^O (d) $I^B I^B$
[*Ans.* (c) I^O]

6. The secretors have the I allele in _____.
(a) tears (b) Gastric juice
(c) Saliva (d) All of these
[*Ans.* (d) all of these]

7. _____ proposed the existence of 8 alleles at a single Rh locus.
(a) Fischer (b) Landsteiner
(c) Bernstein (d) Wiener
[*Ans.* (d) Wiener]

8. XX – XO type of sex determination is in _____
(a) Cockroaches (b) *Drosophila*
(c) Humans (d) Moths
[*Ans.* (a) Cockroaches]

9. The lygaeus type (XX – XY) type of sex determination is seen in _____.
(a) Fishes (b) Chickens
(c) Human beings (d) Gypsy moth
[*Ans.* (c) Human beings]

10. The ZO – ZZ type of sex determination is seen is _____.
(a) moths (b) Reptiles
(c) Human beings (d) Bugs
[*Ans.* (a) moths]

11. The ZW – ZZ type of sex determination is seen is _____
(a) Butterflies (b) Drosophila
(c) Gypsy moth (d) Human being
[*Ans.* (c) Gypsy moth]

12. Sex index is applicable to _____
(a) Homogenetic condition
(b) Heterogametic condition
(c) Genic balance
(d) Gynandromorphs [*Ans.* (c) Genic balance]

13. X chromosomes was discovered by _____.
(a) Landsteiner (b) Henking
(c) Stevens (d) Bridges
[*Ans.* (b) Henking]

14. Y chromosomes was discovered by _____
(a) Stevens (b) Landsteiner
(c) Henking (d) Wiener
[*Ans.* (a) Stevens]

15. _____was first reported by John Cotto.
(a) Erythroblastosis foetalis
(b) Haemophilia
(c) Colour blindness
(d) Haplodiploidy [*Ans.* (b) Haemophilia]

16. Scientists who contributed to karyotyping ___
(a) Tjio and Levan (b) John Cotto
(c) Bridges (d) Wiener
[*Ans.* (a) Tjio and Levan]

17. Depending on position of centromere and relative length of two arms human chromosomes can be classified into ____ type.
(a) 2 (b) 3
(c) 4 (d) 5 [*Ans.* (c) 4]

5. Hemophilia follows _____ pattern of Inheritance. [Ans. criss – cross]

6. Cell division in cells can be arrested by use of a substance called _____ [Ans. colchicines]

7. For karyotype cells in _____ stages of cell division are photographed [Ans. metaphase]

8. Genetic disease in human can be detected by the technique _____. [Ans. karyotyping]

9. _____ is a family tree. [Ansz pedigree]

10. Absence of melanin results in a condition called _____. [Ans. Albinism]

11. In skin during pigment formation DOPA is converted into _____ [Ans. melanin]

12. The enzyme _____ converts DOPA to Melanin. [Ans. Tyrosinase]

13. Involuntary jerking of the body and degeneration of nervous system are symptoms of _____. [Ans. Huntington's chorea]

14. _____ is an example of Autosomal aneuploidy in humans. [Ans. Down's syndrome/Patau's syndrome]

15. _____ is an example of Allosomal abnormality in humans. [Ans. Klinefelter's syndrome / Turner's syndrome]

16. Shell coiling in Limnaea exhibits _____ inheritance. [Ans. Extra chromosomal / Cytoplasmic]

17. Kappa particles in paramecium were studied by _____. [Ans. Sonneborn]

18. The killer paramecia are said to posses _____. [Ans. kappa particle]

19. The kappa particles liberates a toxin called __ [Ans. paramecin]

20. The term _____ means well born. [Ans. Eugenics]

21. The term eugenics was coined by _____. [Ans. Francis Galton]

22. The term Euphenics was coined by _____. [Ans. Joshua Lederberg]

23. The science of improving human race by improving environmental conditions is called _____. [Ans. Euthenics]

Very Short Answers — 2 Marks

1. Mention two measures under negative eugenics.

Ans. (i) Sexual separation of the defectives.
(ii) Sterilization of the defectives
(iii) Control of immigration and
(iv) Regulation of marriages

2. Mention the symptoms seen in trisomy 13/ Pataus's syndrome.

Ans. It is characterized by multiple and severe body malformations as well as profound mental deficiency. Small head with small eyes, cleft palate, malformation of the brain and internal organs are some of the symptoms of this syndrome.

3. What is a syndrome?

Ans. Group of signs and symptoms that occur together and characterize a particular abnormality is called a syndrome. In humans, Down's syndrome, Turner's syndrome, Klinefelter's syndrome, Patau's syndrome are some of the examples of chromosomal disorders.

4. What is Lyon's hypothesis?

Ans. Mary Lyon suggested that Barr bodies represented an inactive chromosome, which in females becomes tightly coiled into a heterochromatin, a condensed and visible form of chromatin. The number of Barr bodies observed in cell was one less than the number of X-Chromosome. XO females have no Barr body, whereas XXY males have one Barr body.

5. What are Gynandromorphy?

Ans. These individuals have parts of their body expressing male characters and other parts of the body expressing female characters. The organism is made up of tissues of male and female genotype and represents a mosaic pattern.

6. How does hemophilia affect an individual?

Ans. A person with a recessive gene for haemophilia lacks a normal clotting substance (thromboplastin) in blood, hence minor injuries cause continuous bleeding, leading to death.

7. What is Rh factor or Rh antigen?

Ans. *(i)* Rh factor is a immunogenic D antigen of the Rh blood group system found on the surface of the erythrocytes of Rhesus monkey and human beings. In the blood it is inherited as a dominant trait.

(ii) An individual having D antigen are Rh D (Rh^+) and those without D antigen are Rh D negative (Rh^-).

SHORT ANSWERS — 3 Marks

1. Why are people with O blood group called as universal donors?

Ans. People with 'O' blood group lack both 'A' and 'B' antigens. Therefore when 'O' blood is donated to another individual, his blood will not produce any antibodies against it. Therefore people with 'O' blood group are called as universal donors.

2. 'AB' Blood group individuals are called universal recipients. Justify.

Ans. People with AB blood group have both 'A' antigen and B antigen in their RBCs. Therefore they can receive blood from 'A' group, 'B' group or 'O' group individuals their blood will not produce antibodies against any of them. Therefore people with 'AB' blood group are called as universal recipients.

3. What is null allele?

Ans. In ABO blood group, each allele (I^A and I^B) produces a transferase enzyme. I^A allele produces N-acetyl galactose transferase and can add N-acetyl galactosamine (NAG) and I^B allele encodes for the enzyme galactose transferase that adds galactose to the precursor (i.e. H substances) In the case of I^O/I^O allele no terminal transferase enzyme is produced and therefore called "null" allele and hence cannot add NAG or galactose to the precursor.

4. How can erythroblasts foetalis be prevented?

Ans. If the mother is Rh negative and foetus is Rh positive, anti D antibodies should be administered to the mother at 28th and 34th week of gestation as a prophylactic measure. If the Rh negative mother delivers Rh positive child then anti D antibodies should be administered to the mother soon after delivery. This develops passive immunity and prevents the formation of anti D antibodies in the mothers blood by destroying the Rh foetal RBC before the mother's immune system is sensitized. This has to be done whenever the woman attains pregnancy.

5. Draw a schematic representation to show ZW – ZZ type of sex determination.

Ans.

	♀	♂
P_1	AAZW	AAZZ
Gametes	AZ AW	AZ
F_1 Generation	AAZZ ♂	AAZW ♀

6. What is SRY?

Ans. SRY stands for sex determining region Y. It is a gene found in the euchromatin regions of the Non–combining region of Y chromosome. It codes for testes determining factor (TDF) present in testes of Males. This gene does not occur in X chromosome.

7. What is sex Index?

Ans. In *Drosophila* the sex is determined by the balance between the genes for femaleness located on the 'X' chromosome and those for maleness located on the 'autosomes' (A). Hence the sex of an individual is determined by the ratio of its X chromosome to that of the autosome A sets. This ratio is termed as sex index and is expressed as

$$\text{Sex Index} = \frac{\text{Number of X chromosomes (X)}}{\text{Number of sets of autosomes (A)}}$$

change in this ratio leads to a changed sex phenotype.

8. Females are carriers of Haemophilia. Give Reason.

Ans. Haemophilia is a disease caused by a recessive X – linked gene. If a normal female marries a colour blind male or vice versa, the gene is carried in the X – chromosome of the female (female has 2 X Chromosomes). If both the chromosomes carry the receive gene she becomes colour blind. If one X chromosome caries the recessive gene she may not suffer from the disease but will be a carrier carrying the gene to the next generation. Haemophilia follows the characteristics criss - cross pattern of inheritance. The females are carriers of the disease and world transmit the disease to 50% of their son's even if a male parent is normal.

20. What is euthenics?

Ans. The science of improvement of existing human race by improving the environmental conditions is called euthenics. It can be achieved by subjecting them to better nutrition, better unpolluted ecological conditions, better education and sufficient medical facilities.

21. What is erythoblastosis foetalis (or) Haemolytic disease of the new born (HDN)? (or) Write about the incompatiability of Rh-factor.

Ans. **(i)** Rh incompatability has great significance in child birth. If a woman is Rh negative and the man is Rh positive, the foetus may be Rh positive having inherited the factor from its father.

(ii) The Rh negative mother becomes sensitized by carrying Rh positive foetus within her body.

(iii) Due to damage of blood vessels, during child birth, the mother's immune system recognizes the Rh antigens and gets sensitized. The sensitized mother produces Rh antibodies.

(iv) The antibodies are IgG type which are small and can cross placenta and enter the foetal circulation. By the time the mother gets sensitized and produce anti 'D' antibodies, the child is delivered.

(v) Usually no effects are associated with exposure of the mother to Rh positive antigen during the first child birth, subsequent Rh positive children carried by the same mother, may be exposed to antibodies produced by the mother against Rh antigen, which are carried across the placenta into the foetal blood circulation.

(vi) This causes haemolysis of foetal RBCs resulting in haemolytic jaundice and anaemia. This condition is known as Erythoblastosis foetalis or Haemolytic disease of the new born (HDN).

22. Define Kin selection.

Ans. The mode of sex determination which facilitates the evolution of sociality in which only one diploid female becomes a queen and lays the eggs for the colony. All other females which are diploid having developed from fertilized eggs help to raise the queen's eggs and so contribute to the queen's reproductive success and indirectly to their own, a phenomenon known as Kin Selection. The queen constructs their social environment by releasing a hormone that suppresses fertility of the workers. **Eg.** Honey bees.

23. What is Sex-switch gene?

Ans. **(i)** A gene, sex–lethal (SxL) located on the X chromosome, in *Drosophila* directs female development.

(ii) This gene has two states of activity. When it is 'on' it directs female development and when it is 'off' maleness ensures.

(iii) Other genes located on the X chromosome and autosomes regulates sex switch gene.

(iv) However, the Y chromosome of *Drosophila* is required for male fertility.

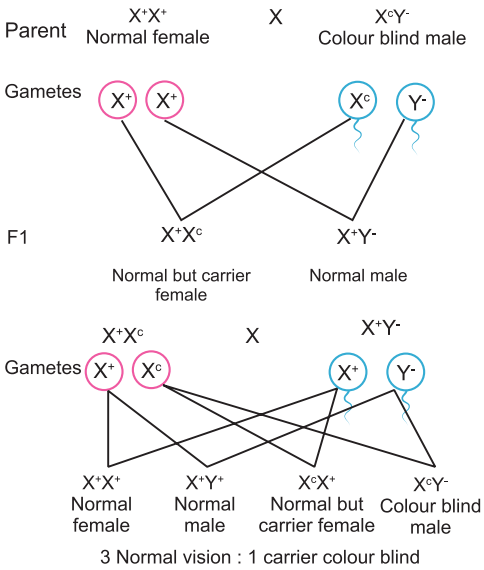
Long Answers — 5 Marks

1. Explain criss-cross pattern of inheritance with an example. (or) Explain Inheritance of colour blindness.

Ans. In human beings a dominant X – linked gene is necessary for the formation of colour sensitive cells, the cones. The recessive form of this gene is incapable of producing colour sensitive cone cells. Homozygous recessive females (X^cX^c) and hemizygous recessive males (X^cY) are unable to distinguish red and green colour. The inheritance of colour blindness can be studied in the following two types of marriages.

(i) Marriage between colour blind man and normal visioned woman:

A marriage between a colour blind man and a normal visioned woman will produce normal visioned male and female individuals in F1 generation but the females are carriers. The marriage between a F1 normal visioned carrier woman and a normal visioned male will produce one normal visioned female, one carrier female, one normal visioned male and one colour blind male. Colour blind trait is inherited from the male parent to his grandson through carrier daughter, which is an example of criss-cross pattern of inheritance.

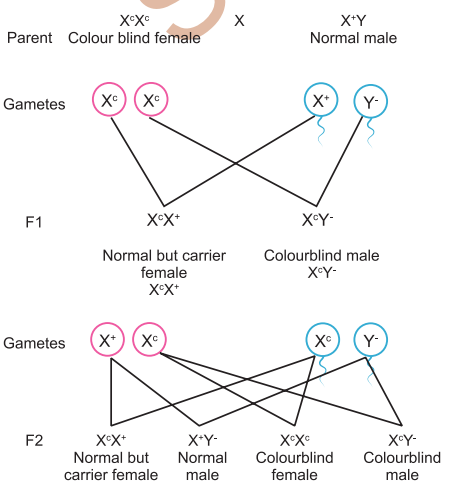


Marriage between colour blind man and normal visioned woman

(ii) **Marriage between normal visioned man and colour blind woman:**

(a) If a colour blind woman (X^cX^c) marries a normal visioned male (X^CY), all F_1 sons will be colourblind and daughters will be normal visioned but are carriers.

(b) Marriage between F_1 carrier female with a colour blind male will produce normal visioned carrier daughter, colourblind daughter, normal visioned son and a colourblind son in the F_2 generation.



Marriage between normal visioned man and colour blind woman

2. Write a note on thalassemia.

Ans. ***(i)*** Thalassemia is an autosomal recessive disorder. It is caused by gene mutation resulting in excessive destruction of RBC's due to the formation of abnormal haemoglobin molecules. Normally haemoglobin is composed of four polypeptide chains, two **alpha** and two **beta** globin chains. Thalassemia patients have defects in either the alpha or beta globin chain causing the production of abnormal haemoglobin molecules resulting in anaemia.

(ii) Thalassemia is classified into alpha and beta based on which chain of haemoglobin molecule is affected. It is controlled by two closely linked genes HBA1 and HBA2 on chromosome 16. Mutation or deletion of one or more of the four alpha gene alleles causes Alpha Thalassemia. In Beta Thalassemia, production of beta globin chain is affected. It is controlled by a single gene (HBB) on chromosome 11. It is the most common type of Thalassemia and is also known as Cooley's anaemia. In this disorder the alpha chain production is increased and damages the membranes of RBC.

3. Write a note on allosomal chromosomal abnormalities.

Ans. Mitotic or meiotic non-disjunction of sex chromosomes causes allosomal abnormalities. Several sex chromosomal abnormalities have been detected. **Eg.** Klinefelter's syndrome and Turner's syndrome.

(i) **Klinefelter's Syndrome (XXY Males)**

This genetic disorder is due to the presence of an additional copy of the X chromosome resulting in a karyotype of 47, XXY. Persons with this syndrome have 47 chromosomes (44AA+XXY). They are usually sterile males, tall, obese, with long limbs, high pitched voice, under developed genitalia and have feeble breast (gynaecomastia) development.

(ii) **Turner's Syndrome (XO Females)**

This genetic disorder is due to the loss of a X chromosome resulting in a karyotype

of 45, X. Persons with this syndrome have 45 chromosomes (44 autosomes and one X chromosome) (44AA+XO) and are sterile females. Low stature, webbed neck, under developed breast, rudimentary gonads lack of menstrual cycle during puberty, are the main symptoms of this syndrome.

4. Discuss the methods adopted for the improvement of human race.

Ans. The methods adopted for the improvement of human beings are

(i) Eugenics
(ii) Euthenics
(iii) Euphenics

Eugenics:

Application of the laws of genetics for the improvement of human race is called eugenics. The term eugenics means "well born" and was coined by Francis Galton in 1885. For the betterment of future generations it is necessary to increase the population of outstanding people and to decrease the population of abnormal and defective people by applying the principles of eugenics.

Two methods of Eugenics

(i) Constructive method or Positive eugenics
(ii) Restrictive method or Negative eugenics

(i) Positive eugenics: Positive eugenics attempts to increase consistently better or desirable germplasm and to preserve the best germplasm of the society. The desirable traits can be increased by adopting the following measures:

(a) Early marriage of those having desirable traits
(b) Subsiding the fit and establishing sperm and egg banks of precious germplasm
(c) Educating the basic principles of genetics and eugenics
(d) Improvement of environmentalconditions
(e) Promotion of genetic research

(ii) Negative eugenics: Negative Eugenics attempts to eliminate the defective germplasm of the society by adopting the following measures:

(a) Sexual separation of the defectives
(b) Sterilization of the defectives
(c) Control of immigration and
(d) Regulation of marriages

Euphenics:

The symptomatic treatment of genetic disease of man is called Euphenics or Medical engineering. In 1960, Joshua Lederberg coined the term Euphenics. It means normal appearing. It deals with the control of several inherited human diseases especially the inborn errors of metabolism. Eg. Phenylketonuria (PKU)

Euthenics:

The science of improvement of existing human race by improving the environmental conditions is called euthenics. It can be achieved by subjecting them to better nutrition, better unpolluted ecological conditions, better education and sufficient medical facilities.

5. Write a note on any 2 Mendelian disorders occurring in human beings.

Ans. The Mendelian disorders in human beings are

(a) Thalassemia
(b) Sickle cell anaemia
(c) Huntington chorea
(d) Phenylketonuria
(e) Albinism

Thalassemia:

(i) Thalassemia is an autosomal recessive disorder. It is caused by gene mutation resulting in excessive destruction of RBC's due to the formation of abnormal haemoglobin molecules. Normally haemoglobin is composed of four polypeptide chains, two alpha and two beta globin chains.
(ii) Thalassemia patients have defects in either the alpha or beta globin chain causing the production of abnormal haemoglobin molecules resulting in anaemia.
(iii) Thalassemia is classified into alpha and beta based on which chain of haemoglobin molecule is affected. It is controlled by two

closely linked genes HBA1 and HBA2 on chromosome 16. Mutation or deletion of one or more of the four alpha gene alleles causes Alpha Thalassemia. In Beta Thalassemia, production of beta globin chain is affected. It is controlled by a single gene (HBB) on chromosome 11. It is the most common type of Thalassemia and is also known as Cooley's anaemia. In this disorder the alpha chain production is increased and damages the membranes of RBC.

Huntington's chorea:

(i) It is inherited as an autosomal dominant lethal gene in man.

(ii) It is characterized by involuntary jerking of the body and progressive degeneration of the nervous system, accompanied by gradual mental and physical deterioration.

(iii) The patients with this disease usually die between the age of 35 and 40.

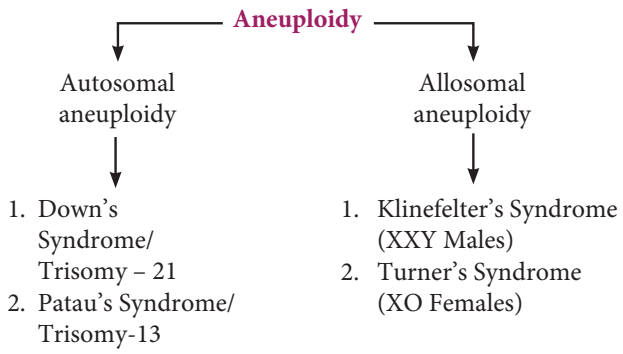
6. Write notes on chromosomal abnormalities in human beings. (or) What is aneuploidy? Explain it.

Ans. In human beings the diploid (2n) body cell has 46 chromosomes (23 pairs).

(i) chromosomal abnormalities are caused by errors is the number or structure of chromosome.

(ii) Failure of chromatids to segregate during cell division resulting in the gain or loss of of one or more choromosomes called aneuploidy. It is caused by the non-disjunction of chromosomes.

(iii) There abnormalities causes various syndromes in human beings. They are



1. Down's Syndrome/Trisomy – 21:

Trisomic condition of chromosome - 21 results in Down's syndrome. It is characterized by severe mental retardation, defective development of the central nervous system, increased separation between the eyes, flattened nose, ears are malformed, mouth is constantly open and the tongue protrudes.

2. Patau's Syndrome/Trisomy-13:

(i) Trisomic condition of chromosome 13 results in Patau's syndrome. Meiotic non disjunction is thought to be the cause for this chromosomal abnormality.

(ii) It is characterized by multiple and severe body malformations as well as profound mental deficiency. Small head with small eyes, cleft palate, malformation of the brain and internal organs are some of the symptoms of this syndrome.

b. Allosomal abnormalities in humanbeings:

Mitotic or meiotic non-disjunction of sex chromosomes causes allosomal abnormalities. Several sex chromosomal abnormalities have been detected. Eg. Klinefelter's syndrome and Turner's syndrome.

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