# 12th Zoology – Unit 2. Chapter 4 – Study Material

## CHAPTER 4 Principles of Inheritance and Variation

**1. heredity and variations**

- ✓ genetics is a branch of biology that deals with the study of heredity and variations.
- ✓ It describes how characteristics and features pass on from the parents to their offsprings in each successive generation.
- ✓ The unit of heredity is known as the gene. Gene is the inherited factor that determines the biological character of an organism.
- ✓ A variation is the degree by which the progeny differs from their parents.

2. **multiple allelism**.

✓ When three or more alleles of a gene that control a particular trait occupy the same locus on the homologous chromosome of an organism, they are called multiple alleles and their inheritance is called **multiple allelism**.

**3. ABO blood group**

✓ Karl Landsteiner discovered two kinds of antigens called antigen 'A' and antigen 'B' on the surface of RBC's of human blood.

✓ ased on the presence or absence of these antigens three kinds of blood groups, type 'A', type 'B', and type 'O' (universal donor)were recognized.

4. The fourth and the rarest blood group 'AB' (universal recipient) was discovered in 1902 by two of Landsteiner's students Von De Castelle and Sturli.**Genetic basis of the human ABO blood groups**

Table 4.1 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 |

5. **Rhesus or Rh – Factor**

✓ The Rh factor or Rh antigen is found on the surface of erythrocytes.

- ✓ It was discovered in 1940 by Karl Landsteiner and Alexander Wiener in the blood of rhesus monkey, *Macaca rhesus* and later in human beings.
- ✓ The term 'Rh factor' refers to "immunogenic D antigen of the Rh blood group system. An individual having D antigen are Rh D positive (Rh+) and those without D antigen are Rh D negative (Rh-)".

6. **Fisher and Race hypothesis:**

Rh factor involves three different pairs of alleles located on three different closely linked loci on the chromosome pair. This system is more commonly in use today, and uses the 'Cde' nomenclature.


7. **Fischer and Race hypothesis – Rh Blood Type - Homologous Chromosome pair (showing 3 loci and 2 alleles per locus)**
   - ✓ In the above Fig. 4.1, three pairs of Rh alleles (Cc, Dd and Ee) occur at 3 different loci on homologous chromosome pair-1.
   - ✓ The possible genotypes will be one C or c, one D or d, one E or e from each chromosome. For e.g. CDE/cde; CdE/cDe; cde/cde; CDe/CdE etc.,
   - ✓ All genotypes carrying a dominant 'D' allele will produce Rh+positive phenotype and double recessive genotype 'dd' will give rise to Rh-negative phenotype.

8. **Wiener Hypothesis**
   - ✓ Wiener proposed the existence of eight alleles (R1, R2, R0, Rz, r, r1, r11, ry) at a single Rh locus.
   - ✓ All genotypes carrying a dominant 'R allele' (R1, R2 ,R0 ,Rz) will produce Rh+positive phenotype and double recessive genotypes (rr, rr1, rr11, rry) will give rise to Rh-negative phenotype.
9. **Erythoblastosis foetalis**
   - ✓ 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.

- ✓ 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).**

**10.autosomes.**

- ✓ Sex chromosomes determine the sex of the individual in dioecious or unisexual organisms.
- ✓ The chromosomes other than the sex chromosomes of an
- ✓ individual are called autosomes.

11.**Y CHROMOSOME**

- ✓ The human Y chromosome is only 60 Mb in size with 60 functional genes whereas X chromosomes are 165 Mb in size with about 1,000 genes.

12.**Prevention of Eryhroblastosis foetalis**

- ✓ 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.

13.**Heterogametic Sex Determination:**

- ✓ In heterogametic sex determination one of the sexes produces similar gametes and the other sex produces dissimilar gametes.
- ✓ The sex of the offspring is determined at the time of fertilization

14.**Heterogametic Males**

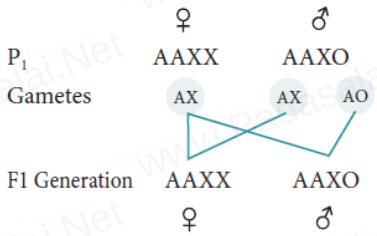
- ✓ In this method of sex determination the males are heterogametic producing dissimilar gametes while females are homogametic producing similar gametes.
- ✓ It is of two kinds XX-XO type and XX-XY type.

**15.XX-XO Type**

**Type of sex determination**

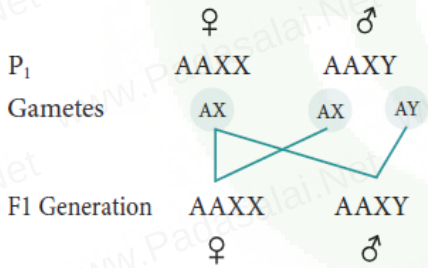
- ✓ This method of sex determination is seen in bugs, some insects such as cockroaches and grasshoppers.
- ✓ The female with two X chromosomes are homogametic (XX) while the males with only one X chromosomeare heterogametic (XO).

- ✓ The presence of an unpaired X chromosomes determines the male sex.
- ✓ The males with unpaired 'X' chromosome produce two types of sperms, one half with X chromosome and other half without X chromosome.
- ✓ The sex of the offspring depends upon the sperm that fertilizes the egg




16. **XX-XY type (Lygaeus Type) type of sex determination**

- ✓ This method of sex determination is seen in human beings and in *Drosophila*.
- ✓ The females are homogametic with XX chromosome, while the males are heterogametic with X and Y chromosome.
- ✓ Homogametic females produce only one kind of egg, each with one X chromosome, while the heterogametic males produce two kinds of sperms some with X chromosome and some with Y chromosome.
- ✓ 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

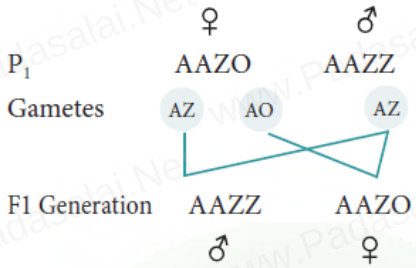



17. **Heterogametic Females**

- ✓ In this method of sex determination, the homogametic male possesses two 'X' chromosomes as in certain insects and certain vertebrates like fishes, reptiles and birds producing a single type of gamete; while females produce dissimilar gametes.
- ✓ The female sex consists of a single 'X' chromosome or one 'X' and one 'Y' chromosome. Thus the females are heterogametic and produce two types of eggs.
- ✓ To avoid confusion with the XX-XO and XX-XY types of sex determination, the alphabets 'Z' and 'W' are used here instead of X and Y respectively.
- ✓ Heterogametic females are of two types, ZO-ZZ type and ZW-ZZ type.

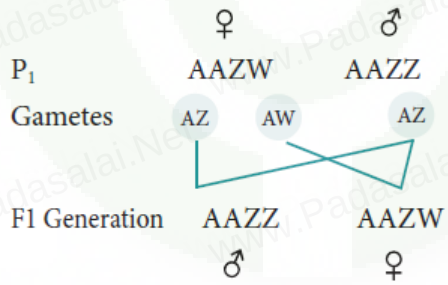
18. **ZO-ZZ Type**
**type of sex determination**

- ✓ This method of sex determination is seen in certain moths, butterflies and domestic chickens.
- ✓ In this type, the female possesses single 'Z' chromosome in its body cells and is heterogametic (ZO) producing two kinds of eggs some with 'Z' chromosome and some without 'Z' chromosome, while the male possesses two 'Z' chromosomes and is homogametic (ZZ).



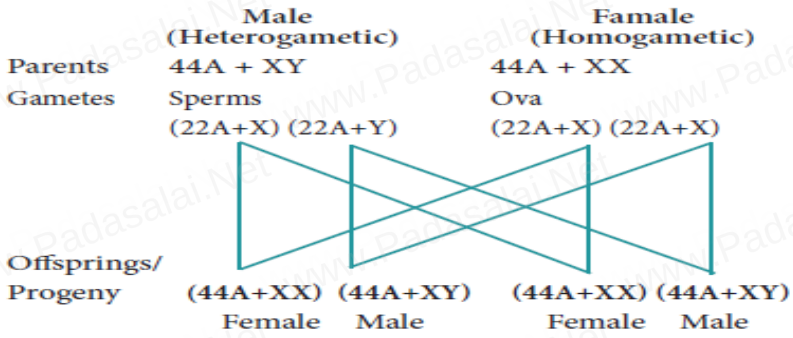

19. **ZW-ZZ type**

- ✓ This method of sex determination occurs in certain insects (gypsy moth) and in vertebrates such as fishes, reptiles and birds.
- ✓ In this method the female has one 'Z' and one 'W' chromosome (ZW) producing two types of eggs, some carrying the Z chromosomes and some carry the W chromosome.
- ✓ The male sex has two 'Z' chromosomes and is homogametic (ZZ) producing a single type of sperm




20. **Sex determintion in human beings**

- ✓ 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.
- ✓ An independently evolved XX: XY system of sex chromosomes also exist in *Drosophila*




21.**Barr body**.

- ✓ In 1949, Barr and Bertram first observed a condensed body in the nerve cells of female cat which was absent in the male.
- ✓ This condensed body was called sex chromatin by them and was later referred as **Barr body**.

22.**Kin Selection**

- ✓ 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.

**23.haplodiploidy mechanism**

- ✓ In hymenopteran insects such as honeybees, ants and wasps a mechanism of sex determination called haplodiploidy mechanism of sex determination is common

**24.haplodiplody**

- ✓ Fertilized eggs develop into females (Queen or Worker) and unfertilized eggs develop into males (drones) by parthenogenesis.
- ✓ It means that the males have half the number of chromosomes (haploid) and the females have double the number (diploid), hence the name haplodiplody for this system of sex determination.

25. **Colour blindness**

- ❖ 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 (XcXc) and hemizygous recessive males (XcY) are unable to distinguish red and green colour.

- ❖ The inheritance of colour blindness can be studied in the following two types of marriages.
- ❖

26. **Karyotyping**

- ❖ Karyotyping is a technique through which a complete set of chromosomes is separated from a cell and the chromosomes are arranged in pairs.
- ❖ An idiogram refers to a diagrammatic representation of chromosomes.

27.**karyotype**

- ❖ The individual chromosomes are cut from the photograph and are arranged in an orderly fashion in homologous pairs.
- ❖ This arrangement is called a **karyotype**.

28.**Applications of Karyotyping:**

- ❖ It helps in gender identification.
- ❖ It is used to detect the chromosomal aberrations like deletion, duplication, translocation, nondisjunction of chromosomes.
- ❖ It helps to identify the abnormalities of chromosomes like aneuploidy.
- ❖ It is also used in predicting the evolutionary relationships between species.
- ❖ Genetic diseases in human beings can be detected by this technique.

29.**Human Karyotype**

Depending upon the position of the centromere and relative length of two arms, human chromosomes are of three types:

Metacentric, sub metacentric and acrocentric. The photograph of chromosomes are arranged in the order of descending length in groups from A to G

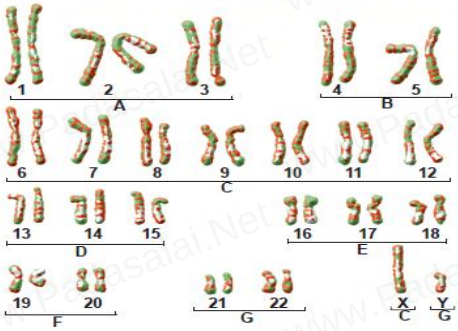



Fig. 4.9 - Human karyotype (male)

30.**Pedigree Analysis**

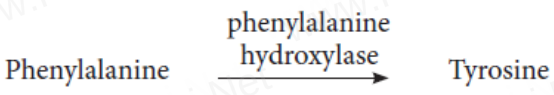
- ❖ Pedigree is a "family tree", drawn with standard genetic symbols, showing the inheritance pathway for specific phenotypic characters
- ❖ Pedigree analysis is the study of traits as they have appeared in a given family line for several past generations

31.**Thalassemia**

- ❖ 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.
  - 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.

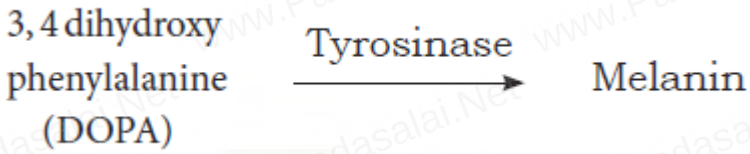
32.**Phenylketonuria**

- ❖ It is an inborn error of **Phenylalanine** metabolism caused due to a pair of autosomal recessive genes. It is caused due to mutation in the gene PAH (phenylalanine hydroxylase gene) located on chromosome 12 for the hepatic enzyme "phenylalanine hydroxylase" This enzyme is essential for the conversion of phenylalanine to tyrosine.
- ❖ Affected individual lacks this enzyme, so phenylalanine accumulates and gets converted to phenylpyruvic acid and other derivatives.
- ❖ It is characterized by severe mental retardation, light pigmentation of skin and hair. Phenylpyruvic acid is excreted in the urine.



33.**Albinism**

- ❖ Albinism is an inborn error of metabolism, caused due to an autosomal recessive gene. Melanin pigment is responsible for skin colour.
- ❖ Absence of melanin results in a condition called albinism. A person with the recessive allele lacks the tyrosinase enzyme system, which is required for the conversion of dihydroxyphenyl alanine (DOPA) into melanin pigment inside the melanocytes.
- ❖ In an albino, melanocytes are present in normal numbers in their skin, hair, iris, etc., but lack melanin pigment.




34.**Huntington's chorea**

- ❖ It is inherited as an autosomal dominant lethal gene in man.
- ❖ It is characterized by involuntary jerking of the body and progressive degeneration of the nervous system,
- ❖ accompanied by gradual mental and physical deterioration. The patients with this disease usually die between the age of 35 and 40.

35.**Chromosomal Abnormalities**

- ❖ Each human diploid (2n) body cell has 46 chromosomes (23 pairs). Chromosomal disorders are caused by errors in the number or structure of chromosomes.
- ❖ Chromosomal anomalies usually occur when there is an error in cell division. Failure of chromatids to segregate during cell division resulting in the gain or loss of one or more chromosomes is called aneuploidy.
- ❖ It is caused by non-disjunction of chromosomes. 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.

**36.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.

## 37.Patau's Syndrome/Trisomy-13

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

## 38.Allosomal abnormalities in human beings

- 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.

## 39.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

## 40.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.