ST. LAWRENCE HIGH SCHOOL

A JESUIT CHRISTIAN MINORITY INSTITUTION

STUDY MATERIAL -7

Class: XII **Sub: BIOLOGICAL SCIENCE** **Date: 29.06.2020**

Topic - Ch 5 – (Part III) Principles of Inheritance and variations

---

PEDIGREE ANALYSIS

Definition: It is a chart of the family history , showing the inheritance of a particular trait (character) is known as pedigree chart.

- It is the analysis of a person for a particular trait ,taking into consideration ,its occurrence in the family and available ancestry.
- **The person or individual ,for whom the analysis is made , is called propositus.**
- For pedigree analysis,the whole available record of the family history of the propositus is required.
- The investigator tries to trace the history of the character by going through the history of the family.
- Pedigree analysis is helpful in:
  - i) Tracing the first appearance of the character in the family.
  - ii) Nature of the character.
  - iii) Genetic counseling regarding the occurrence of the disease.

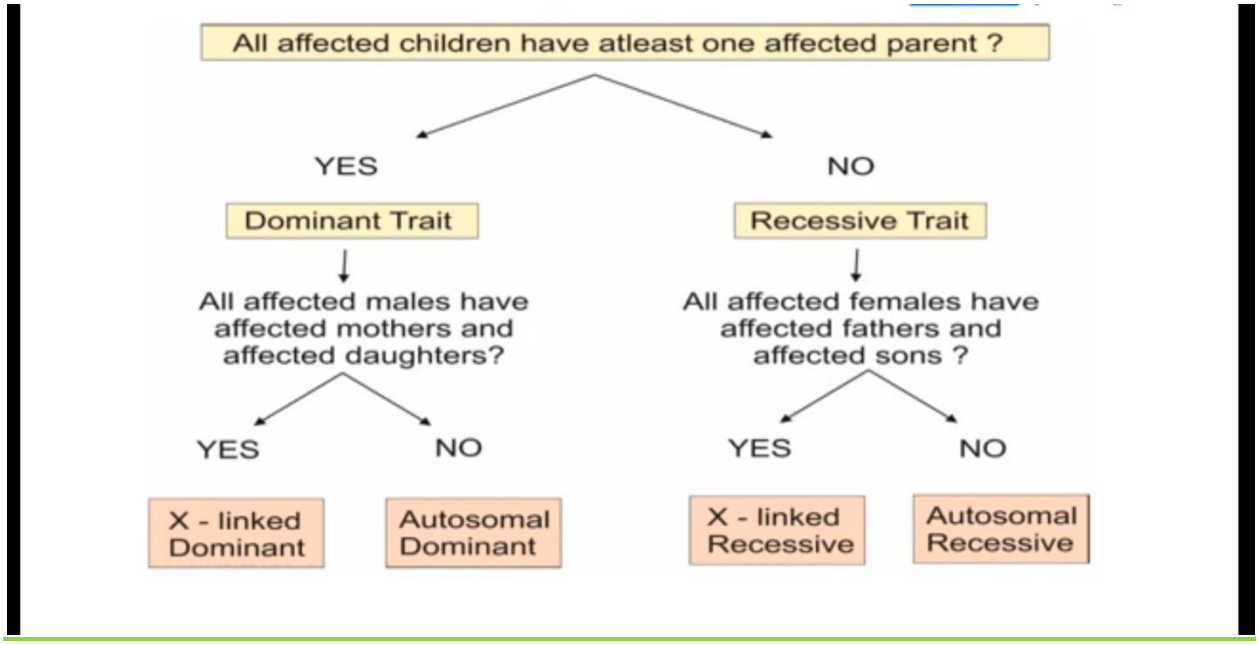
How to analyse :

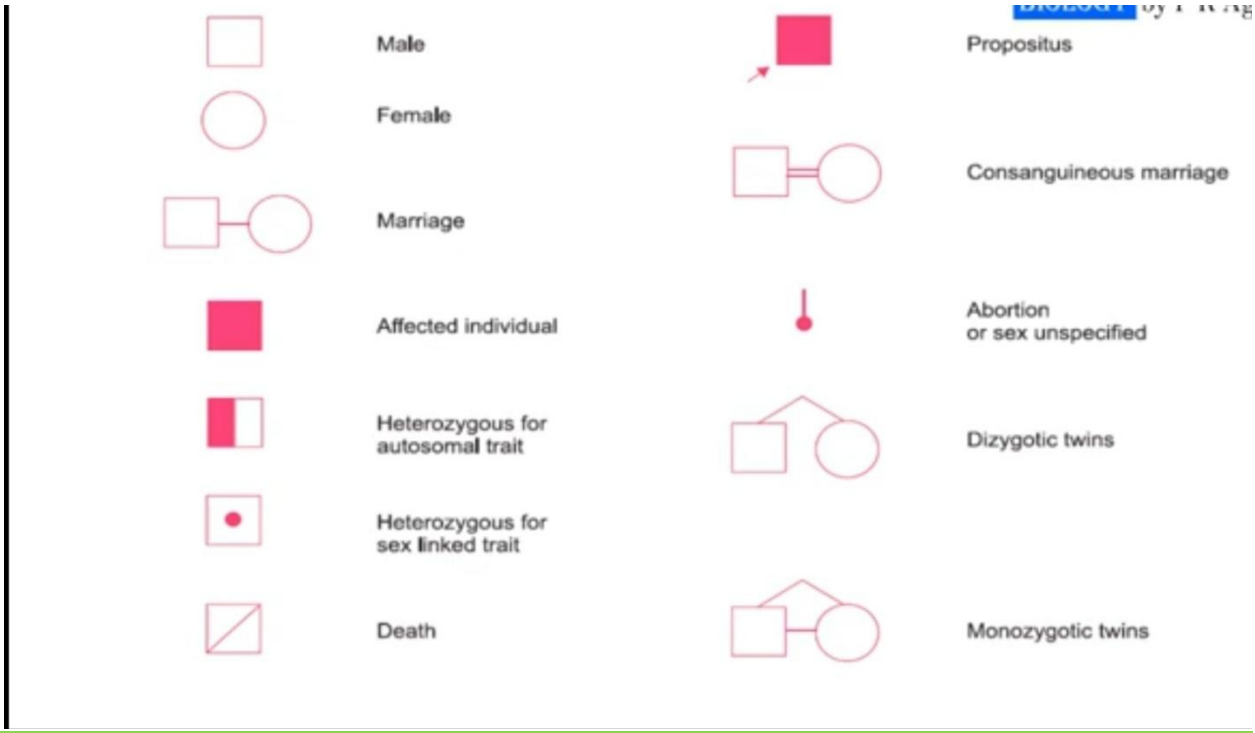
There are four different types of inheritances that are considered in a pedigree chart.These are:

- Autosomal Dominant
- Autosomal recessive
- Sex linked dominant
- Sex linked recessive

Some tips for solving the pedigree:

1) If a disease is absent in parents ,but present in children ,it is a recessive disease.
2) Dominant trait never skips a generation.
3) When father transfers a disease to his son ,it is always autosomal.
4) When parents and children are usually affected it is usually a dominant trait.



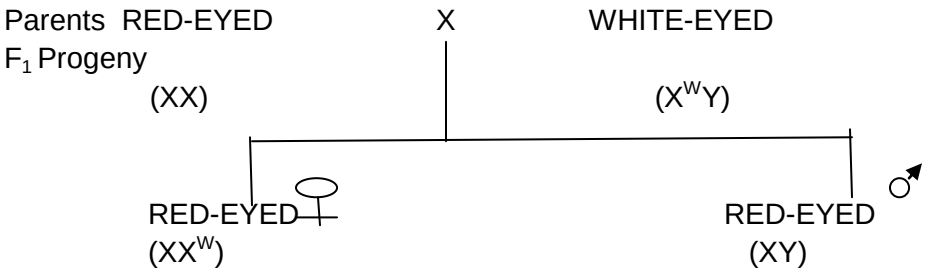


SYMBOLS FOR PEDIGREE ANALYSIS

SEX-LINKAGE OR SEX-LINKED INHERITANCE

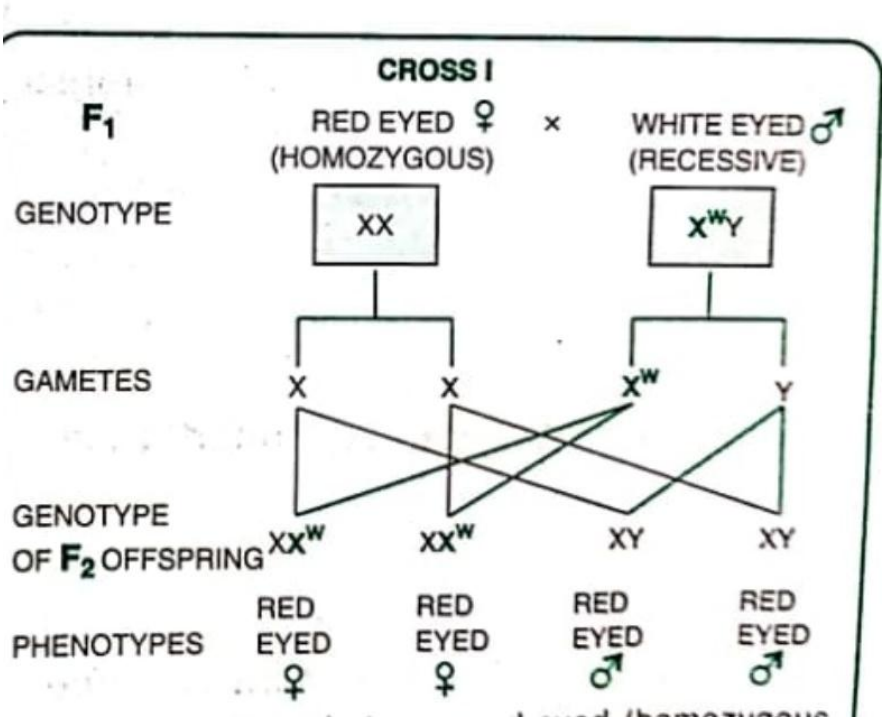
The sex chromosome or X-chromosome carries genes for sex determination and also some genes for some somatic characters. The genes for somatic characters present on sex chromosomes are called sex-linked genes. The characters which are controlled by the sex-linked inheritance or sex-linkage. The inheritance of sex-linked characters is linked to the sex. It was discovered by Morgan in *Drosophila melanogaster* when he found sudden appearance of a white-eyed *Drosophila* and followed its inheritance.

Cross I: Between Red-eyed Female and White-eyed Male : Morgan (1910) noted a white-eyed male in a culture of red-eyed *Drosophila*. This white-eyed male was mated with a homozygous red-eyed female *Drosophila*. **All males and females of F_1 progeny were red-eyed**.



When these F_1 males and females were allowed to interbreed, the F_2 progeny consisted of

1. **FEMALES** :
   All RED-EYED — HOMOZYGOUS (XX) = 25%
   HETEROZYGOUS (XX^W) = 25%
2. **MALES** — RED-EYED (XY) = ¼ 25%
   WHITE-EYED (X^WY) = ¼ = 25%



A cross between red eyed (homozygous female) and white eyed male

Cross II : Between F_1 Red-eyed Female (F and a white-eyed Male : Morgan made a cross between F_1 females (heterozygous) and white-eyed males. This is a test cross. The F_2 results were white and red-eyed females and red and white-eyed males in equal proportion.

1. FEMALES : WHITE-EYED (X^WX^W) = 25%
   RED-EYED (X^WX) = 25%
2. MALES WHTIE-EYED (X^WY) = 25%
   RED-EYED (XY) = 25%

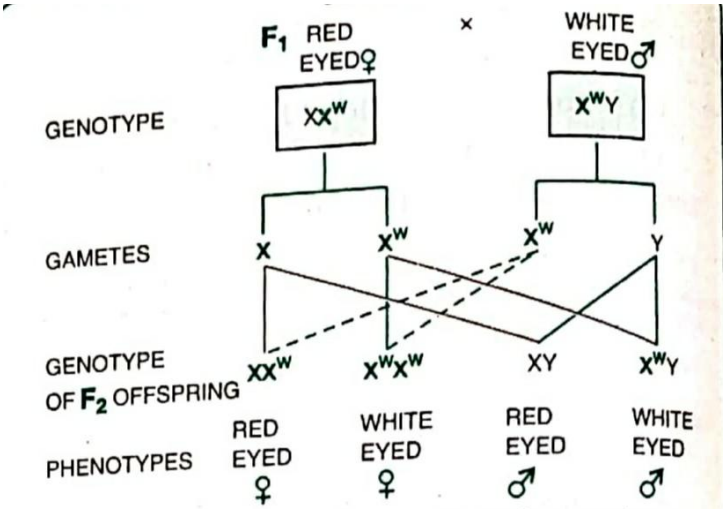


Fig. 10.28 Testcross of F_1 female and recessive male (The F_2 progeny has both red and white-eyed males and females in equal proportion)

CROSS II

Cross III : Between F_2 White-eyed Female and Red-eyed Male : Morgan then made a cross between F_2 white-eyed female (X^WX^W) and red-eyed male (XY). In F_2 generation, all females were red-eyed (X^WX) and all males were white-eyed (X^WY).

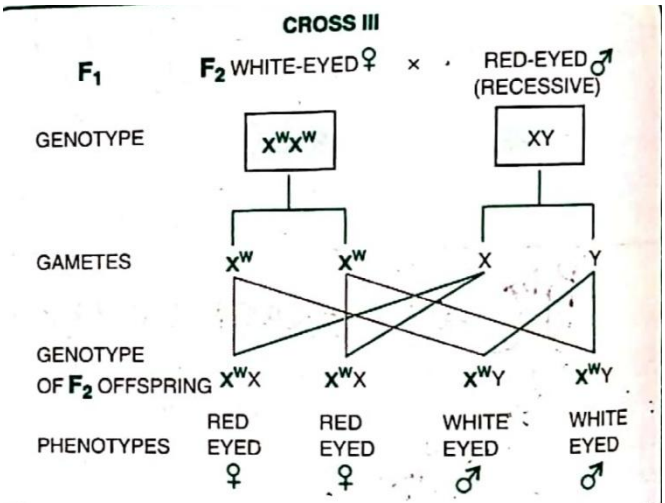


Fig. 10.29 A reciprocal cross between white-eyed female and red-eyed male produces males all white-eyed and females all red-eyed (but heterozygous)

Criss-Cross Inheritance.

Cross I and Cross III are reciprocal. According to Mendel's laws, the results of reciprocal crosses are expected to be identical but it is not so in these cases. Morgan observed that in first cross, only red eyes appeared in both female and male Drosophila, but in cross III all the males were white-eyed.

From all the three crosses, it is evident that

- Gene for the eye colour is located on the X-chromosome.
- Male transmits its X-chromosome to his daughters and female transmits its X-chromosome to both sons and daughters. This is called **criss-cross inheritance.**

Significance of Criss-cross Inheritance

- Criss-cross inheritance helps in establishing relationship between genes and sex chromosomes. It provides evidence that sex-linked genes are located on X-chromosome.
- It helps in understanding mechanism of inheritance of sex-linked disorders.

Characteristics of Sex-linked Inheritance

- The sex-linked genes (gene for eye colour) are located on X-chromosome. They are also called X-linked genes. Y-chromosome is without gene for eye colour.
- Female has XX-chromosome and therefore two genes for each sex-linked character (eye colour)
- Male has XY-sex chromosomes. Y is without corresponding allele of eye colour gene. Therefore, male possesses only one gene for eye colour.
- Male produces two types of male gametes. Half of them have X-chromosome (with sex-linked gene) and other half Y-chromosome (without sex-linked gene). The female produces only one type fo eggs, all with one X-chromosome and with X-linked genes.
- Female offspring of F1 generation receive one X-chromosome from mother and other X-chromosome from father, i.e., they have one gene for eye colour from mother and one gene for eye colour from father.
- The male offspring receive X-chromosome from mother and Y-chromosome from father. Therefore, males receive eye colour gene from mother only.
- Female transmits one X-chromosome to its male and one X-chromosome to its female offspring. If red-eyed female is heterozygous, it produces both red and white-eyed male offspring in equal proportion, because 50% of them receive w gene and other 50% receive W gene from mother.
- Red-eyed male offspring of F_1 generation transmit X-chromosome to F_2 females and Y-chromosome to F_2 males. Therefore, all F_2 females receive W gene from males and are all red-eyed.
- The sex-linked character (white eye) being recessive is not expressed in heterozygous condition in F_1 and F_2 female flies. It means a white-eyed female possess gene for white eye colour (w) on both its X-chromosomes.
- The sex-linked characters are passed on from male parent to grandsons (F_2 males) through F_1 females. The inheritance of recessive sex-linked character from male to F_2 males (grandsons) through F_1 female is called criss-cross inheritance.

Sex-limited Characters

The genes for sex-limited characters are present in both sexes but they are expressed only in one sex. For example, milk producing genes are functional only in female mammals. Hair on the pinna of ear develop only in males.

Sex-influenced Characters

Sex influenced characters develop due to the effect of sex hormones. For example, gene for baldness functions as an autosomal dominant trait in males and as an autosomal recessive in females.

MUTATIONS

Definition

Mutations are changes in the hereditary material. These introduce variations in the morphology. Physiology and behaviour among individuals of a population or species.

Based on the nature of genetic material, mutations are classified into three categories:

- Changes in the structure of genes : Gene Mutations
- Changes in the structure of chromosomes : Chromosomal Mutations
- Changes in the number of chromosomes : Genomic Mutations

(a) Gene Mutations or Point Mutations (Micromutations)

Gene mutations or point mutations are changes in the fine structure of genes. Since genes are DNA segments, the gene mutations include changes in the number or arrangement of nucleotides. These are known as intragenic mutations. Though, these mutations involve minor changes in DNA (one or few nucleotides), they may modify the information conveyed by the genes. Lethal genes, albino character, chlorophyll deficiencies, sickle cell anaemia, etc. are produced by gene mutations.

Muton : Muton is the smallest portion of a gene that can mutate or change. The smallest mutton is a single nitrogenous base. Thus, a gene has almost as many mutons as is the number of nitrogenous bases (or nucleotides).

Causes and Types of Gene Mutations

The changes in the sequence of nitrogenous bases may be due to the following changes.

(i) **Changes in Number of Nucleotides in a DNA Segment (Cistron) or Frame Shift Mutation**: These are caused either by deletion or addition (insertion) of one or more nucleotides in a DNA segment.

Since genetic code is commaless, both addition as well as deletion of nucleotides shift the reading frame of codons from the site of change onward. Therefore, these mutations are known as **frame shift mutations.** These mutations change the reading frame of codons and the sequence of amino acids in a protein from the point of deletion or insertioin. The new polypeptide chain may be entirely different from the normal one. It may be modified, deleterious or inactive. For example, human hereditary disease muscular dystrophy is caused by frame shift mutation which leads to premature termination of translation of protein dystrophin.

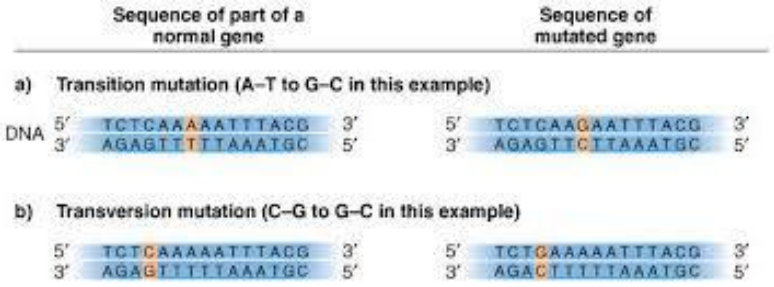
(ii) **Replacement of a Base Pair (nucleotide pair) in a DNA Segment or Cistron or the Substitution Mutation** : These are caused either by : (a) Transition or (b) Transversion.

(a) **Transition** : In transition, a purine is replaced by another purine and a pyrimidine is replaced by another pyrimidine, i.e., A = T is replaced by G = C or vice versa.

(b) **Transversion** : In transversion, a purine is replaced by a pyrimidine or a pyrimidine by a purine, i.e., C=G is replaced by G = C or A = T is replaced T = A.

The substitution mutation alters a single codon, which replaces a single amino acid in a polypeptide chain. Even this could change nature of the protein and character associated with it as in case of sickle cell anaemia.

Gene mutations occurring due to change in a single nucleotide are called **point mutations**. Gene mutations which involve change of more than one nucleotides are called **gross mutations**. Most mutations occur due to errors in DNA replication or transcription. Therefore, they are called copy error mutations.



(b) **Chromosomal Mutations or Chromosomal Aberrations (Macromutations)**

These mutations include changes in the number of genes in a chromosome or in the arrangement of genes

(i) **Changes in the Number of Genes in a Chromosome** : The changes are :

(ai) **Deficiency or deletion** : It involves loss of one or a block of genes caused due to loss of chromosome segment.

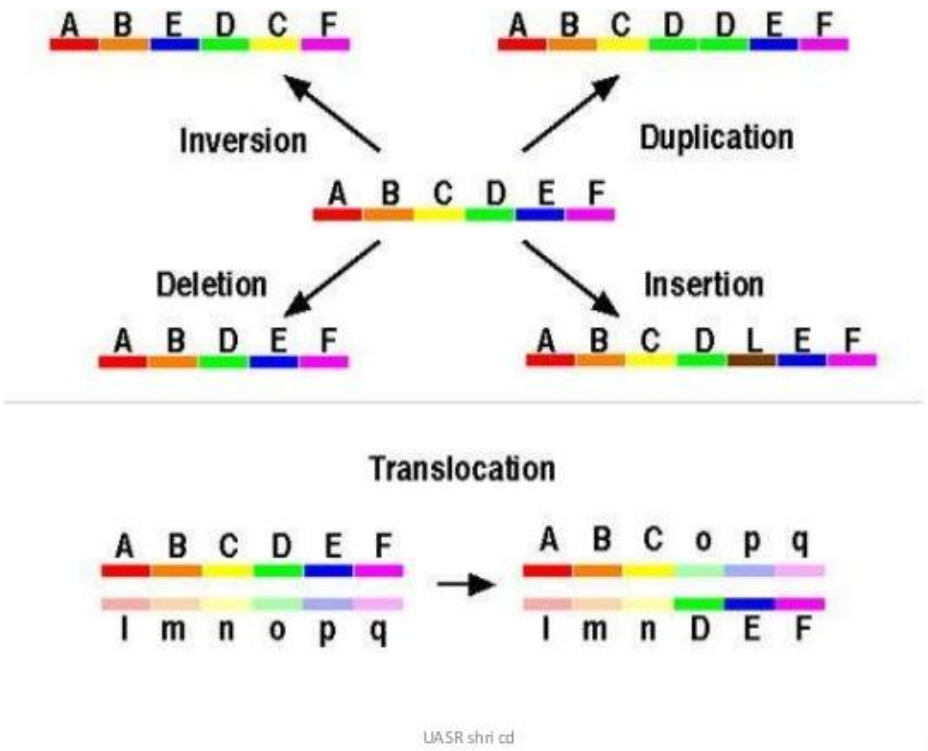
(aii) **Duplication** : It involves addition of one or more genes. As a result, the organism carries same genes repeated twice in a single chromosome and the chromosome becomes longer.

(ii) **Changes in the Arrangement of Genes in a Chromosome** : These are :

(aii) **Inversion** : It involves rotation of a block of genes within a chromosome at 180º. For example, if the original chromosome is ABCDEFGHI, it may change to ABCFEDGHI.

(aiv) **Translocation** : It involves exchange of parts between two non-homologous chromosomes to form new chromosomes. For example, the original chromosomes ABCDEF and WXYZ exchange parts to form new chromosomes with gene arrangement ABC and DEFWXYZ.

The structural changes in chromosomes occur due to error during meiotic division and result in changed sequence genes. The genes in new or changed location may produce changed phenotypic expression or may prove to be lethal causing death of the individual.



TYPES OF CHROMOSOMAL ABERRATIONS

(c) **Genomic Mutations (Changes in the Number of Chromosomes)**

Numerical changes in chromosome number are of two types : Euploidy and Aneuploidy.

(i)**Euploidy:** Loss or gain of complete set of chromosomes from an organism.

(a) Loss of complete set of chromosomes from a diploid organism (2n) results in the formation of haploid or monoploid offspring with just one set of chromosomes (n). This is called Monoploidy or Haploid.
(b) Addition of one or more haploid sets of chromosomes to the genome of diploid organism results in the formation of polyploids. Polyploids can be :-

- Triploid : Organisms with 3 sets of chromosomes, i.e., 2n+n = 3n

- Tetraploids : Organisms with 4 sets of chromosomes, i.e., 2n+2n = 4n
- Pentaploids : Organisms with 5n chromosomes
- Hexaploids : Organisms with 6n chromosomes
- Heptaploids : Organisms with 7n chromosomes

(ii) **Aneuploidy** : Aneuploidy is the addition or loss of one or more chromosomes to a haploid genome.

- Loss of one or more chromosomes from the genome represents hypoploidy.
- Addition of one or more chromosomes to the genome represents hyperploidy.

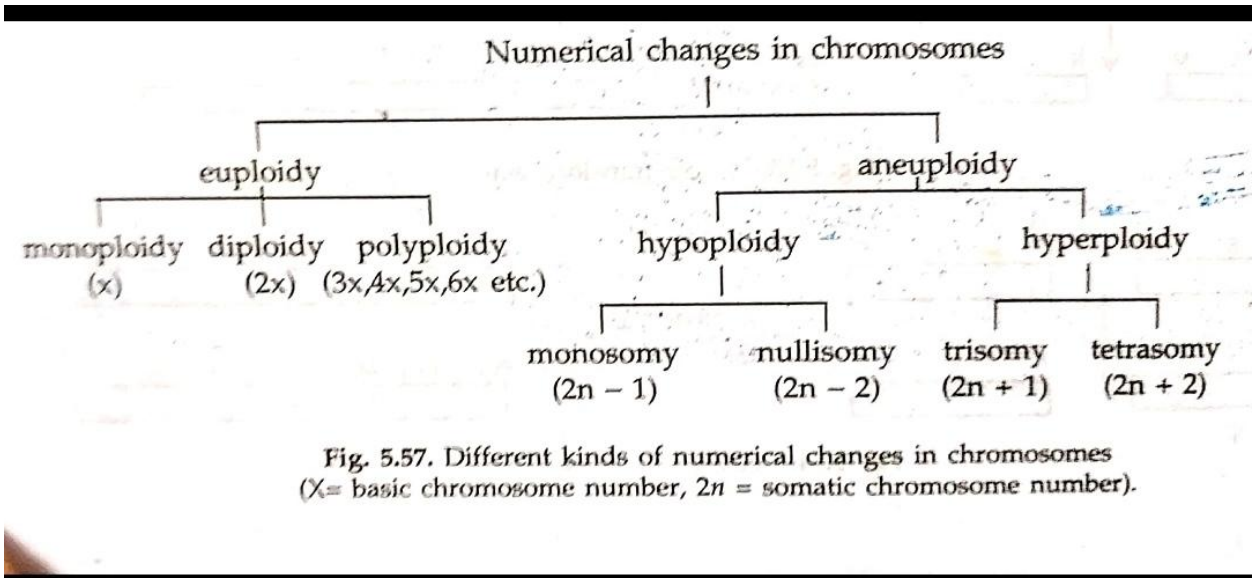


Fig. 5.57. Different kinds of numerical changes in chromosomes (X= basic chromosome number, 2n = somatic chromosome number).

Spontaneous and Induced Mutations and Origin of Mutations

(a) **Spontaneous Mutations** : These mutations arise under natural environment. These are caused either due to intracellular factors or due to background cosmic radiation. Spontaneous mutations have a low frequency estimated to be one per 1,00,000 cells.

(b) **Induced Mutations** : These mutations are introduced artificially or experimentally. Any physical or chemical agent which introduces mutation is called a mutagen or mutagenic agent. The mutagens may be :

- **Physical Mutagens** : These are high or low temperature and the radiations. The radiations are X-rays, α rays, UV-rays etc.
  These cause break in the chromosomes and are responsible for chromosomal mutations.
- **Chemical Mutagens** : These are mustard gas, colchicines, acridine dyes, nitrous acid, aminopurines, 5-bromouracil, hypoxanthine.

IMPORTANCE OF MUTATIONS

- **Variations** : Mutations introduce variations in the individuals of a population or in the members of a species. New variations may provide better adaptability to the new environment.
- **Evolution** : Useful variations introduced by mutations are favoured by nature and lead to evolution of new species. In some cases, even a single mutation produces new variety such Ancon sheep.
- **In Agriculture** : In agriculture, new varieties or cereal plants, fruits, vegetables, etc. have been developed by scientists through incorporation of induced mutations by treatment with mutagens. A few mutant varieties developed by scientists are :
  - Dwarf varieties of Rice and Wheat.
  - Amber-grained wheat developed from maxican variety of wheat.
  - Cabbage, Cauliflower, Knol knol, Tomato, Potato, etc.
  - Seedless varieties of fruits like Banana, Grape and Waterlemon.
  - New varieties of ornamental plants from wild varieties.

(iv) **In Animal Husbandary**: In animal husbandary, mutant forms of pets, domestic animals, cattle, sheep, etc. have been developed for obtaining better yield of milk, butter and meat, better variety of wool, and strong varieties of horses and mules (the beasts of burden).

GENETIC DISORDERS

Genetic disorders are two types :
(a) Mendelian disorders
(b) Chromosomal disorders

(a) **Mendelian Disorders (Genetic disorder due to gene mutation)**: They are mainly due to mutation of single gene. The pattern of inheritance of such disorders can be traced in family by pedigree analysis.

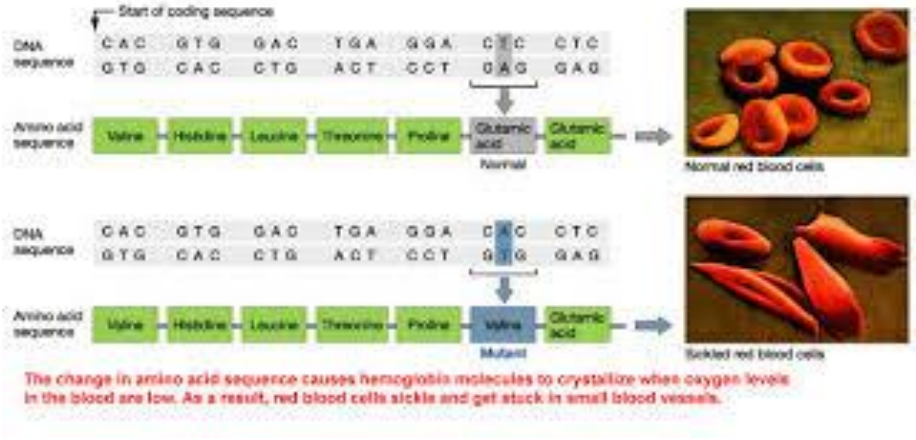
(ai) **Haemophilia** : In haemophilia blood lacks the capacity to coagulate. This disease is also inherited through an X linked recessive gene. Thus a single gene produces the disease in males, while two genes will produce disease in females. Female in heterozygous condition (with one gene) is apparently normal but actually the carrier of the disease. Carrier female transmits the gene to 50 per cent of her sons. This disease is expressed in males, but is transmitted by females. A person suffering from this defect may bleed to death due to small cut and rarely survive to attain marriageable age.

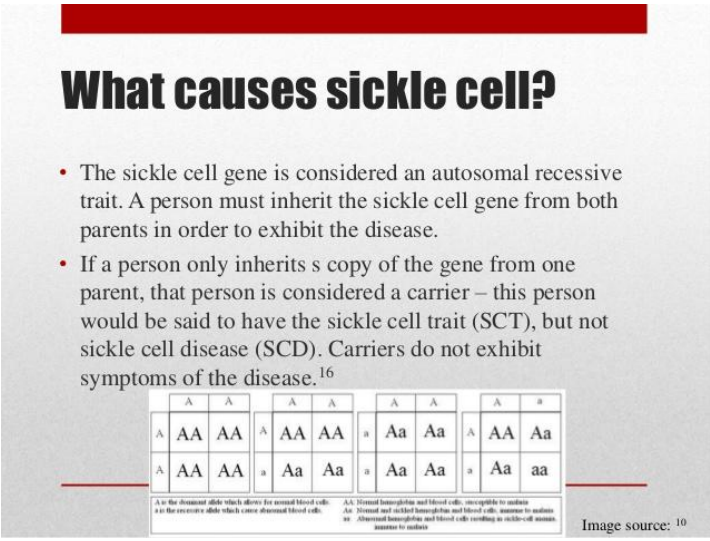
(aii) **Sickle cell anaemia** :. An interesting example of genetic disorder is hereditary disease sickle cell anaemia. The disease is caused be gene (Hb^S) which is lethal in homozygous condition but has a slight detectable effect in the heterozygogus condition. In sickle cell anaemia a change in shape of red blood cells occurs in the venous blood. Being deficient in oxygen tension, these erythrocytes show a marked change in their structrure attaining a sickle shaped structure. As a result rupturing of cell may take place and chronic haemolytic anaemia is

caused. This disease is caused when gene responsible for haemoglobin produced by recessive alleles differs in one amino acid i.e. it incorporates valine in place of glutamic acid.

Sickle cell anaemia is common in persons of African descent and is also found in some other parts of the world where malaria is, or has been major cause death.

In heterozygotes Hb^AHb^s, Some red cells contain haemoglobin A, other haemoglobin S, Because both the types of haemoglobin, rather than a single intermediate form are produced, it is also a case of codominance. Under normal conditions, heterozygotes manifest none of the severe symptoms of HbsHbs persons, though they may suffer some periodic discomfort and even develop anaemia after a time at high altitude.





(aii) **Phenylketonuria (PKU)**

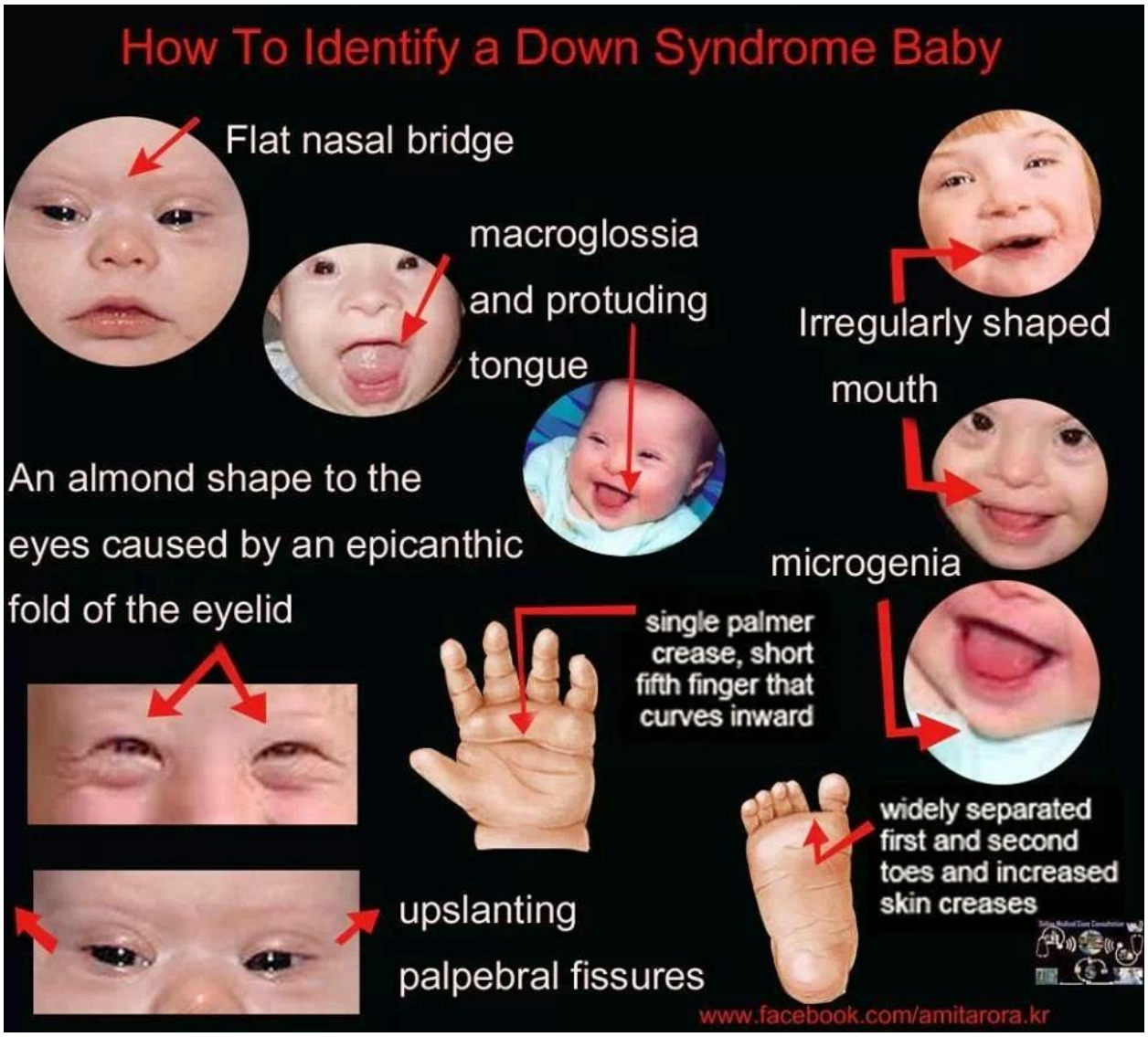
It is characterized by abnormal increase in the level of phenylalanine in the blood due to absence of enzyme phenylalanine hydroxylase. Excess of phenylalanine changes into phenylpyruvic acid which damages brain and causes mental backwardness in the child.

It is inherited as autosomal recessive trait backwardness in the child.

(c) Autosomal genetic disorders

(d)

(aiv) **Down's Syndrome or Mongolism** : Mongolism or Down's syndrome was reported by Langdon Down in 1866. It is due to trisomy of 21nd chromosome that the number is 47. Such condition of thrisomy appears due to formation of n+1 male or female gamete subsequently fertilization by normal n gamete i.e. n+n+1. Formation of n+1 gamete takes place due to the phenomenon of non-disjunction at anaphase stage of meiosis, where paired homologous chromosomes fails to separate. This is found in higher age mothers with age 35 to 40 years. This type of meiotic abnormality is not expected to run in families. But sometimes, 'familial Down's syndrome' appears due to translocation of major part of chromosome 21 to chromosome 14. Such individuals are with 46 chromosomes with partial trisomy of chromosome 21. The affected children have broad forehead, short neck, flat hands with stubby fingers, open mouth, projecting lower lip with a long extending open mouth, projecting lower lip with a long extending tongue and are mentally retarded. The child is mongoloid in appearance with slanting eyes and fold on the eye lid.



(c) **Sex chromosome linked genetic disorders**

(i) **Klinefelter's syndrome**

Such persons have 47 chromosomes with one additional X-chromosome, i.e., 44+XXY. The person is a male with some feminine characters like enlarged breasts (gynecomastia) one long limb, degeneracy of seminiferous tubules, sparse body hairs, mental retardation etc. There may be greater number of X chromosomes 44 + XXXY ascertainable by 2 Barr bodies in interphase or 44+XXXXY etc. resulting in skeletal abnormalities, low mental ability etc. The addition of extra Y leads to 44+XYY (47) chromosomes. The males with such condition show above average height and sub-normal intelligence. They are prone to psychopathic tendencies.

Presence of extra X chromosome in females with 44+XXX : 44+XXXX etc. will show abnormal sexual developments and mental retardation. Females bear formal genitalia. Such individuals are sterile.

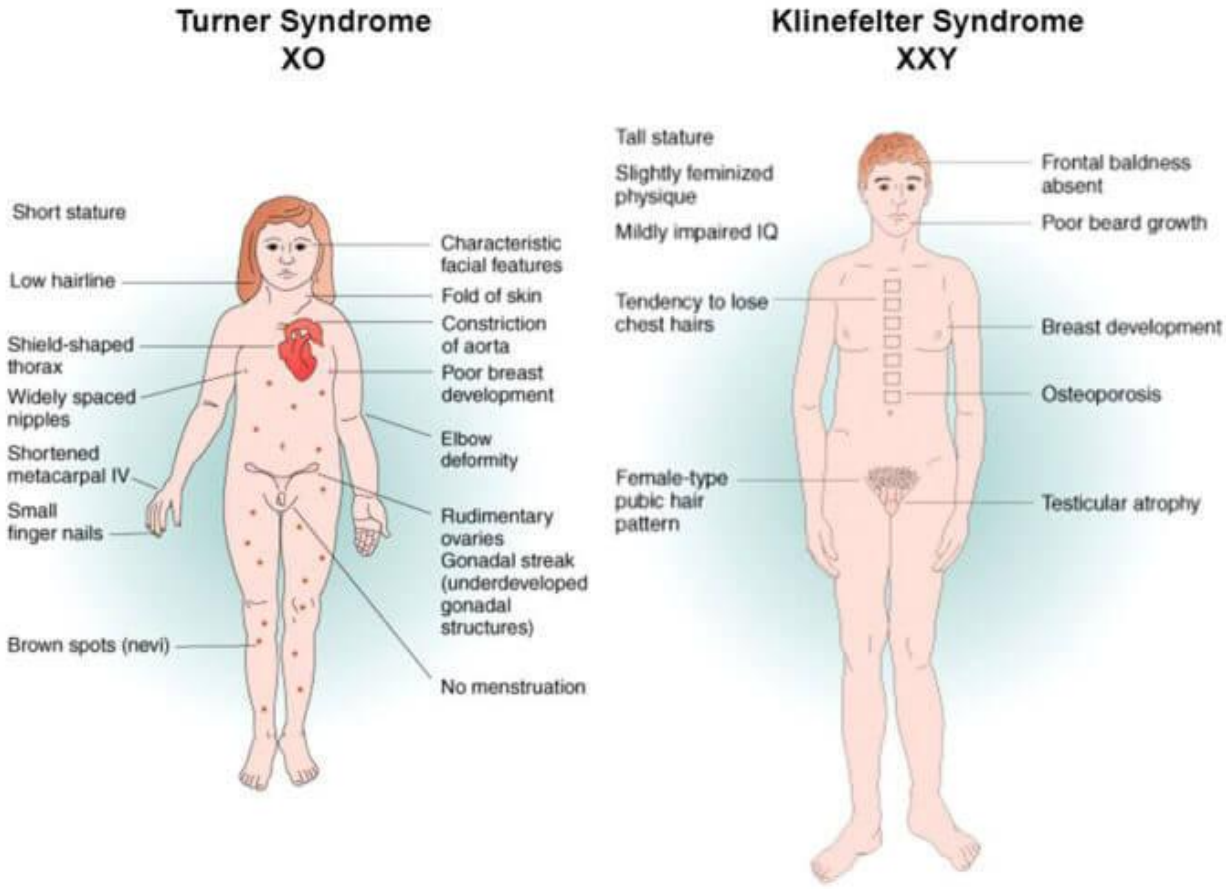
(ii) **Turner's Syndrome**.

It is one of the most common type of female genetic disorder characterized by hypogonadism. Incidence rate is 1 in 2500-3000 live female births. It was first discovered by H.H. Turner (1938).

Cause. : Karyotypic studies showed that genotype of female suffering from Turner's syndrome has 45, (44+XO) chromosomes in which one X-chromosome is less than the normal karyotype. So the female is monosomic for sex chromosome.

Clinical symptoms. Individual is female but is characterized by short stature underdeveloped breasts, reduced ovaries and uterus, no oogenesis and menstrual cycle, little pubic hair mental retardation, infertility, webbing of neck, heavy neck muscles, somatic cells with no sex chromatin etc.

Origin. Turner's syndrome originates from the development of an abnormal zygote formed by the fusion of an abnormal egg (with no X-chromosome) and a normal gymnosperm (sperm with X-chromosome) or due to fusion of a normal egg, (with X-chromosome) and abnormal sperm (with no sex chromosome).



Important Questions for H.S. Examination:

A) Define/Explain the following (1 mark):

1) Allele
2) Pedigree analysis
3) Pleiotropy
4) Multiple allele
5) Linkage
6) X linked gene
7) Mutation
8) Frame shift mutation
9) Sex linkage
10) Muton
11) Mutagen

Linkage map

12) Crossing over
13) Chromosomal mapping
14) Genome

15) Criss cross inheritance

B) Differentiate between the following: (2 mark)

1) Homozygous and heterozygous
2) Transition and transversion
3) Euploidy and aneuploidy
4) Allosome and autosome
5) Chromosomal aberration and genomic mutation
6) Spontaneous and induced mutation
7) Test cross and back cross
8) Genotype and phenotype

C) 3 mark questions:

1) Compare incomplete linkage and complete linkage with suitable examples.
2) Differentiate incomplete dominance and co dominance with suitable examples.
3) Explain the phenomenon of multiple allelism with reference to Human blood groups.
4) Explain criss cross inheritance.
5) What is pedigree analysis? State its significance.
6) Explain the different types of chromosomal aberrations with suitable diagrams.
7) Explain the cause for the following disorders:
   a) Klinefelter syndrome
   b) Turner syndrome
   c) Down syndrome
8) State the cause and symptoms for Sickle cell anaemia.
9) Give the cause for phenylketonuria and haemophilia.
10) State the reasons for Mendel's success in experimentation and also give two reasons why his laws did not receive much importance when they were proposed?

D) Long answer type questions:

1) Explain the different types of mutations.
2) Explain the phenomenon of sex linkage in man
3) Explain sex linked inheritance in *Drosophila.*
4) State the laws of Mendel and their limitations.
5) Explain the mechanism of sex determination in different organisms.

Shaista Ahmed