Genetics

Name .....
Subject .....
College .....
Roll No .....

AGENDA NOTES FOR THE 32nd AUDIT COMMITTEE OF THE DIRECTORS OF MAHARASHTRA NATURAL GAS LIMITED TO BE HELD ON 18/09/2017 AT HOTEL HYATT REGENCY, WEIKFIELD IT PARK, NAGARROAD, PUNE-411014

Mukadam and Director Commercial Mr. S.S. Sontakke. Based on the presentation the aforesaid select committee had selected M/s. Pipalia Singhal & Associates, Chartered Accountants as Internal Auditors for FY 17-18 on the following grounds:

I. The firmM/s. Pipalia Singhal & Associates, Chartered Accountants possesses the domain experience of conducting audit of Oil and Gas Sector companies.
II. They have the Knowledge of PSU processes and methodologies.
III. They have sound knowledge of business processes and internal controls.
IV. The Partners of the firm are instrumental in presentation of papers at different forums and even authors of the books pertaining to Audit domain.

The committee may deliberate on the subject and approve the proposal and forward the following resolutions to the Board for their approval.
Draft Resolution:
"RESOLVED THAT Pursuant to Section 138 of the Companies Act, 2013 read with rule 13 of Companies(Accounts) Rules 2014 and other applicable provisions if any, the Audit Committee hereby approve the appointment of M/s. Pipalia Singhal & Associates, Chartered Accountants as Internal Auditors of the Company for the financial year 2017-18 subject to the approval of the Board of Directors. ``

RESOLVED FURTHER THATM/s. Pipalia Singhal & Associates, Chartered Accountants will conduct the internal audit as per the scope of internal audit assigned to them at a remuneration of Rs.1,50,000 per quarter i.e. 6,00,000/- per annum for FY 2017-18 plus taxes as applicable. ``

Genetics 2 Semester

PRINCIPLES OF GENETICS

-Prof. Shinde S. B
-Prof. Dhoke J.N.
&
-Prof. Pachpor N.S.

DIVISION OF AGRICULTURAL BOTANY

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HISTORY AND SCOPE OF GENETICS

Introduction

Genetics

Genetics is the science of inheritance and variation. Or genetics may be redefined as the science that deals with the structure, organization, transmission and function of genes, and the origin of variation in them. The word 'genetics' is a Greek word 'Gen 'meaning 'to become' or 'to grow into' or 'to generate', was used for first time by **Bateson** 1906.

Inheritance

Transmission of character / traits from generation to generation.

Variation

Differences exhibited in progeny. (As compare to parents / ancesters)

Cytogenetics

Branch of biology devoted to study of various aspects of chromosomes and their effects on the developinent of characters of organisms.

Pre-Mendelian ideas about heredity

Spontaneous Generation

i. Universally accepted but almost never stated facts of genetics that *an organism originated from a pre-existing organism of similar kind.* Thus an organism does not arise or originated directly from nonliving matter. Many biologist then believed that some primitive organism originated from non-living matter, e.g., decaying organic matter. Such a *de novo* origin of living organism from nonliving materials is known as spontaneous generation.

ii. Redi (1662-1697) and Spallanzani (1729- 1799) presented experimental evidence against spontaneous generation, but argument was raised.

iii. Finally Pasture (1822-1895) and Tyndall (1820-1895) conclusively proved that microbes do not originate spontaneously from organic matter.

Preformation

i Aristotle (350 B.C.) believed that the progeny derived their form from semen, while nutrition was provided by the females. Later this concept in 17th and 18th centuries

called as *animalculist.* Harvey (1578-1657) speculated that the progeny developed from eggs and that semen only produce a stimulatory effect (called ***aura seminalis***). This suggestion was further elaborated by scientists called *ovists.*

ii. After the discovery of eggs and sperms, many biologists began a detailed study these gametes. In 1720-1793 **Swammerdam and Bonnet** examining human sperms thought that he had seen a minature human figure called *'homunculus'*, inside the sperm.

iii. A group of biologist (*animal-culists*) claimed that homunculi were present in sperm, while another group (*ovists*) believed that homunculi were located in eggs. i.e. according to these scientists, new human beings were already present in the gametes.

iv. Thus the development of progeny involved only the growth of this preformed human being. This hypothetical phenomenon is termed as preformation. However this theory was rejected because this could not be proved scientifically.

Epigenesis: Wolf (1738-1794)

i. Showed that different adult structures of both plants and animals develop from uniform embryonic tissues.

ii. Embryonic tissues are composed of similar cells progressively differentiate into the adult tissues and organs.

iii. Wolf believed that adult tissues originated completely *de novo* (afresh).

iv. Gametes contain undifferentiated living substances.

v. Gametes form organized body after fertilization.

vi. von Baer (1792-1876) proposed that adult tissues developed through a sequential modification (differentiation) of the embryonic tissues themselves. This concept is universally accepted view of origin of the adult organs and tissues from zygotes.

Inheritance of acquired characters

i. Lamarck (1744-1829) proposed that characters acquired by individuals of one generation are transmitted to the next generation.

ii. It's also popularly known as *'lamarckism'*, was originally suggested by Hippocrates about 400 B.C.

iii. According to the idea of inheritance of acquired character, if person develops strong muscles by exercise, his children will inherit this characteristic. Thus theory was rejected.

Pangenesis: Charles Darwin (1868)

i. Production and development of pangenes (**Pan**- Small, **Genesis**-Originating) i.e. very small invisible bodies produced from every part of bodies.

ii. Each organ of an individual produces very small, almost invisible identical copies of itself called ***gemmules or pangenes***.

iii. Transportation of pangenes to sexual organ i.e. gametes through blood stream

iv. Development and distribution of different organs after fertilization i.e. union of pangenes from both parents. Thus theory of pangenesis is a theory of **'blending inheritance'**.

Germplasm theory: August Weisman (1834-1914)

i. Two types of cells are available in body of an organism.
   a) Somatic cells with somatoplasm
   b) Germ cells / reproductive cells with germplasm.

ii. Genes are situated on chromosomes present in germplasm.

iii. Germplasm is handed over from parents to offspring and gives rise to soma /body.

iv. Germplasm can produce somatoplasm but somatoplasm can not from germplasm.

Knight (1779) conducted experiments on pea much before Mendel but failed to formulate the laws of inheritance because he could not use the mathematics to his result.

J. Kolreuter (1733-1806) a German botanist performed hybridization experiment in tobacco and compared the hybrids with their parents. He showed that both the parents make equal contribution to the hybrids.

Gartner (1772-1850) & Naudin (1815-1899) done experiments similar to Kolreuter and they observed the similar result and could not apply mathematics to their results.

Schleiden and Schwann (1838-39)	-	cell theory
Virchow's (1858)	-	cell lineage theory
Robert hook (1665)	-	first describe 'cell'
Robert Brown (1831)	-	presence of nuclei in cell
Schneider (1873)	-	first account of 'mitosis'

Hertwing & Strausberger (1875) - i) fertilization in plant and animal involves fusion of egg & sperm nuclei.
ii) Describe cell division in plant

W. Flemming (1882) - i) coin term chromatin and demonstrate longitudinal division of chromosomes during meiosis.
ii) Coin the term mitosis.

Van Beneden (1883) - studied 'meiosis' in Ascaries and observed that gametes contain contain 'n' chromosomes and fertilization restores somatic chromosome no. '2n'.

Waldeyer (1888) - coined the term 'chromosome'

Mendelian Period (1822-1886)

1865 - Mendel presented his paper to the Brunn Society for Natural History

1866 - Mendel's paper published in the proceedings of the Brunn Society for Natural History.

1900 - Mendel's work discovered by **Hugo de Vries, Correns and von Tschermak.**

1901 - **Hugo de Vries** coin the term 'mutation'.

1901 - **Landsteiner** gives human ABO blood group

1902 - **Mc Clung** discover X chromosome in insect and suggest that it determine sex.

1906 - **Bateson** introduce the term- Genetics, homozygote, heterozygote, F_1, F_2, allelomorph and epistatic genes.

1908 & 1909 - **Nilsson and Ehle** propose 'multiple factor hypotheses'.

1909 - **Johannsen** propose 'pure line theory'. Coin the term gene, genotype, and phenotype.

1910 - **Morgan** sex linkage in Drosophilla (Nobel Prize 1933)

Recent advances (Post Mendelian Period)

1927 - **Mullar** (Nobel Prize 1946) reported the use of the CIB technique to demonstrate that X rays are mutagenic.

1928 - **Griffth's** discovery of transformation in Diplococcus pneumoniae.

1941 - Beadle and Tatum's work (Nobel Prize 1958) on Neurospora was published, establishing the one gene- one enzyme concept.

1944 -Avery, **Macleod and McCarty** demonstrated that the pneumococcal "transforming principle" is DNA

1946 - **Lederberg** (Nobel Prize 1958) and **Tatum's** discovery of conjugation in bacteria.

1950 - **McClintok's** (Nobel Prize 1983) transposable elements i.e. jumping gene in maize.

1952 - **Harshey** (Nobel Prize 1969) and **Chase** demonstrated that the genetic materials of bacteriophage T_2 is DNA.

1953 - **Watson and Crick** (Nobel Prize 1962) worked out the double helix structure of DNA using the X ray diffraction data of **Wilkins** (Nobel Prize 1962) and the base composition data of **Chargaff.**

1958 - **Meselson and Stahl** demonstrated that DNA replication is semi conservative.

1958 - **Kornberg** (Nobel Prize 1959) isolation of DNA polymerase I from *E. coli.*

1959 - **Ochoa** (Nobel Prize 1959) discovery of the first RNA polymerase.

1961 - **Jacob and Monod** (Nobel Prize 1965) proposed "operon model" for regulating gene expression.

1966 - **Khorana and Nirenberg** (Nobel Prize 1968) the complete genetic code was established

1970 - **Nathans and Smith** (Nobel Prize 1978) isolated the first restriction endonuleases.

1972 - **Berg** (Nobel Prize 1980) first recombinant DNA produced *in vitro.*

1977 - Publication of the DNA sequencing techniques of **Maxam and Gilbert and of Sanger, Nicklen and Coulson (Sanger and Gilbert,** Nobel Prize 1980)

Scope or Significance of Genetics

In the last few decades the science of genetics has provided an insight into all aspects of biology so that it has assumed a central position of great significance in biology as a whole. While on the one hand genetics is used for a study of the mechanism of heredity and variation on the other hand it has provided tools for

Classification of genetics according to significance or scope

I.Transmission Genetics

Study of transmission of genetic material from one generation to other, it includes-

a) Mendelian Genetics

i. Study of qualitative and quantitative traits and influence of environment on their expression.

ii. Includes study of recombination and linkage.

b) Non- Mendelian Genetics

i. Study of role of cytoplasm and it's related organelles

ii. Study of mutation, chromosomal changes, gene mutations.

II.Molecular and Bio-chemical Genetics

i. Involves study of structure and function of gene.

ii. isolation and characterization of gene, DNA sequence regulating gene expression, transgenic plant, recombinant DNA techniques, DNA finger printing

iii. Forensic medicine involving- identification of criminals and doubtful parentage.

iv. Production of insect/ pest/ disease/ herbicide resistance field crops.

v. Production of improved animals producing better milk and meat.

Molecular farming

Phenomenon of transfer of genes (having industrial importance) into plants and animals which can be later on used for production of chemicals.

III.Population and Biometrical Genetics

Study of behaviour and effect of genes in population by using mathematical models e.g. Intelligence (IQ) is governed by

- Genetics (parentage)
- Environment (adopted parentage)
- Development stage (age) of individual

IV.Developmental Genetics

e.g. Cytoplasmic inheritance

- Cytoplasm derived from mother
- Role of nucleus and cytoplasm in differentiation

V.Forward and Reverse Genetics

i. Forward genetics as one begins with the phenotype of an organism and proceeds sequentially to identify the gene or DNA sequence responsible for this phenotype.

ii. Reverse genetics begin with a DNA sequence of unknown function and then proceeds to determine its role in the development of phenotype.

CELL STRUCTURE AND FUNCTION

Introduction

Cell is the structural and functional unit of all living organism, except viruses. Cell shows a large variation, but all the cells belonging to a single tissue are generally similar in size and shape. Cells may be spherical, cylindrical, rod shaped, hexagonal cylindrical or of irregular shape.

In case of Prokaryotic cell the DNA lies free in the cytoplasm and the region is known as nucleoid, where as in case of Eukaryotic cells, the DNA is found inside the nucleus, which is surrounded by nuclear envelope. Some of the differences between prokaryotic and eukaryotic cells are given the table 2.1.

Table 2.1. Difference between Eukaryotic and Prokaryotic cells

S.No	Eukaryotic cells	Prokaryotic cells
1	Nuclear is surrounded by nuclear envelope	Nuclear envelop is absent
2	DNA is associated with histones and non-histone protein to form chromatids fibre.	DNA is naked
3	Cytoplasm contains ER, golgi bodies etc.	Absent
4	Mitochondria present	Absent. Oxidative phosphorilation associated with plasma lemma.
5	All green plants possess chloroplast with typical grana	Chloroplast absent. BGA have lamellar photosynthetic structure but they do not have grana.
6	Cytoplasm have microtubules provides stability to cytoplasm	Do not have microtubules
7	Ribosome 80_s (60_s & 40_s sub units)	Ribosome 70_s (50_s & 30_s sub units)
8	Movement of chromosome associated with spindle fibres	Spindle fibres absent. Movement of chloroplast accompanied by enlargement of plasma lemma.
9	Most of ribosome attached to ER	All ribosome are free in cytoplasm
10	Nucleolus is present in the nucleus.	Nucleolus is absent

Generally the animal cells are similar to plant cells but the former contains centriole which is absent in plant cell. Some of the chief differences which are present in the plant cells are.

Table 2.2. Difference between Animal cells and Plant cells

Cell organelle	Animal Cell	Plant Cell
Lysosomes	Lysosomes occur in cytoplasm	Lysosomes usually not evident
Cilia	Present	It is very rare
Shape	Round (irregular shape)	Rectangular (fixed shape)
Chloroplast	Animal cells don't have chloroplasts	Plant cells have chloroplasts because they make their own food
Vacuole	One or more small vacuoles (much smaller than plant cells).	One, large central vacuole taking up 90% of cell volume.
Centrioles	Present in all animal cells	Only present in lower plant forms.
Plastids	Absent	Present
Cell wall	Absent	Present

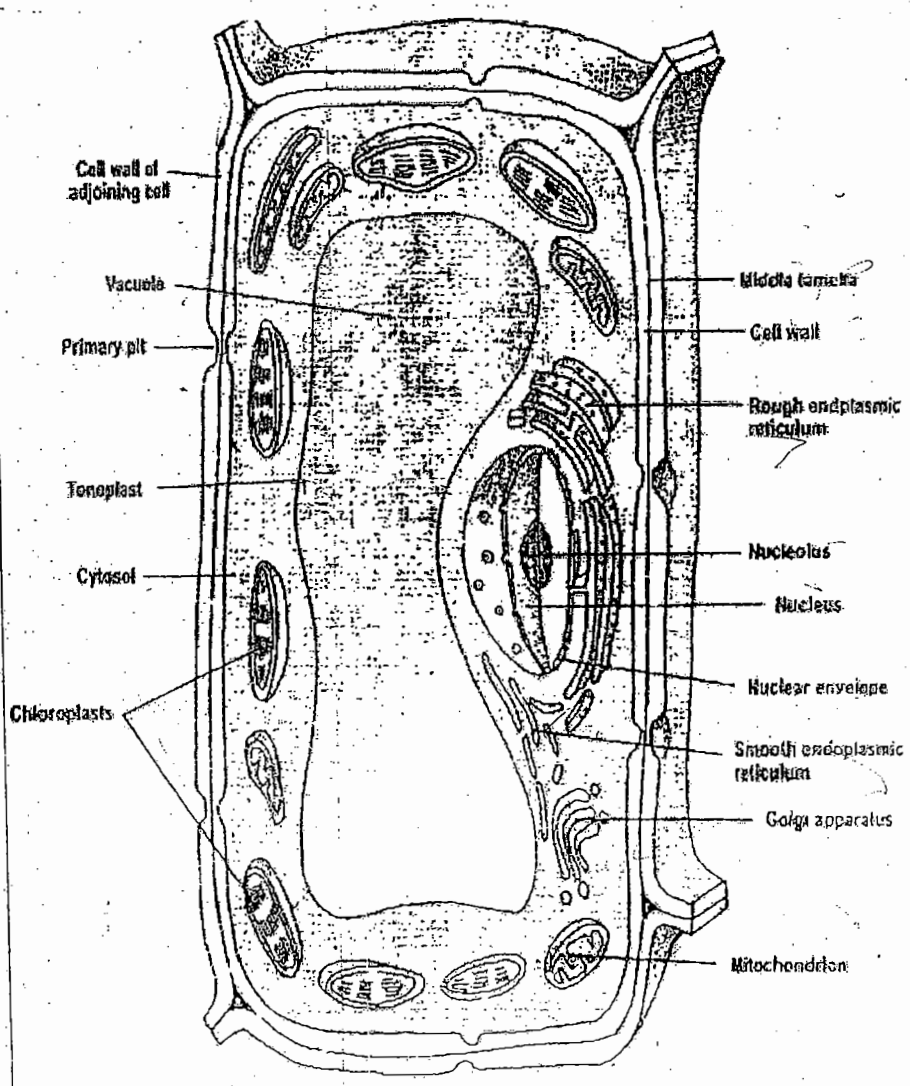


Fig. 3.19. Plant cell structure : A three-dimensional view of a section passing through the nucleus and the large central vacuole. The presence of a cell wall and plastids (chloroplasts in this picture) distinguishes it from an animal cell.

Plant cells have following organelles

A. Cell Organelles

1. Cell wall
2. Plasma lemma
3. Cytoplasm
4. Nucleus

B. Cytoplasmic organelles

1. Endoplasmic reticulum (ER)
2. Ribosome
3. Golgi bodies
4. Lysosomes
5. Spherosomes
6. Vacuoles
7. Micro tubules
8. Mitochondria
9. Chloroplast / Plastids
   i. Leucoplasts
   ii. Chromoplasts

C. Nucleus organelles

1. Nuclear Membrane
2. Nucleolus
3. Chromosome
4. Nuclear sap / Nucleoplasm / Karyolymph

A. Cell organelles

1. Cell wall

Non-living and rigid coat around plant cell is called cell wall

Function-

i.Provides definite shape, mechanical support and strength to tissues and organs.

ii.Cell wall is composed of middle lamella, primary cell wall, and secondary cell wall.

iii.It is primarily made up of cellulose, hemicellulose.

2. Plasma lemma / plasma membrane

Membrane enclosing cytoplasm of a cell is known as plasma lemma. It is composed of lipids and proteins.

3. Cytoplasm

Substance except nucleus surrounded by plasma lemma is known as cytoplasm

4. Nucleus

Dense, oval, spherical body lying embedded in the protoplasm along with chromosome is called **nucleus.** Nucleus is the store-house of almost all the genetic information needed for the function of a cell/ organism.

Function-

i. Controls physiological activities
ii. Carries hereditary characters
iii. Initiates process of cell division

B. Cytoplasmic organelles

1. Endoplasmic reticulum

Membrane closed spaces along with membranes in cytoplasm mostly in differentiated cells are called ER.

Double membrane structure which contains-1) vesicle 2) tubules 3) cisterns these parts are attached to nuclear membrane on one side and cell membrane on other side.

Types of ER

i.Smooth ER

Smooth surface ER in which no protein synthesis takes place. It functions as glycogen synthesis.

ii.Rough ER

Outer surface of ER are rough due to attachment of ribosomes, mostly composed of cisterns and found in cells which are active involved in protein synthesis.

Function-

i. Provide structural base for protein and phospholipids synthesis
ii. Provide channel for transport inside and out side cell and m RNA.

iii. Several enzyme molecules e.g. ATPase, glucose-6-phosphatase etc.

2. Ribosomes

Small cellular particles composed of proteins and RNA attached to outer surface of rough ER and then converts smooth ER to rough ER, Mostly synthesized in nucleolus.

Function-

It is essential for protein synthesis.

3. Golgi bodies

Composed of 2 to 7 flat cisternae (close to each other) and originate from ER elements.

Function-

Food material such as protein, lipids, and phospholipids synthesized with the help of ER and transported to golgi bodies, where they are stored, often modified and then packed into vesicles cut off from them. Thus it functions as packaging unit for plant cells.

4. Lysosomes

Vesicle containing hydrolytic enzymes (mostly acid phosphatase) with the help of these enzyme materials is digested and thereafter lysosomes are called as secondary lysosomes (containing digested food material)

Function-

i. **Lysis**- digestion of food and microbes ingested by cell.

ii. Cause **autolysis** of cell i.e. lysis of concerned cell (RBC are also digested)

5. Spherosome

Vesicles containing lipids (98%)

Function-

Lipid storage

6. Vacuole

Unit membrane (tonoplast) surrounding cell sap (i.e. sugar, salt, proteins, phenols, anthocyanin pigments.) within cytoplasm are called vacuoles.

7. Microtubules

Fibres made up of protein. Found in the form of spindle fibres and constitute spindle apparatus.

8. Mitochondria

Rod like cytoplasmic organelle, main site for cellular respiration and main source of energy, containing DNA so, associated with cytoplasmic inheritance and surrounded by double membranes.

Function-

i. Contains enzyme for oxidation of amino acid, fatty acid, and enzymes for oxidative phosphorylation.

ii. By this oxidation process ATP (energy) form, this is necessary for biochemical reaction of cell.

iii. DNA responsible for cytoplasmic inheritance.

9. Chloroplast / Plastids

Chloroplast containing the most important pigment on earth, i.e. chlorophyll chloroplast is enclosed by two concentric membranes. Membrane-free space enclosed by these two membranes is referred to as **stroma.** Within the stroma are embedded **grana.** Each granum contains flat cisternae stacked on top of each other.

In addition to chloroplasts, plant cells contain plastids i.e. self replicating cytoplasmic organelles in plant. Two more types of plastids called (1) **leucoplasts**- colourless plastids, function in storage of starch, protein, and fats (2) **chromoplasts**- it contain phucoxanthin, phycocyanin etc.

Function- Photosynthesis

C. Nucleus organelles

1. Nuclear Membrane (NM)

Provides selective continuity between nuclear and cytoplasm materials.

2. Nucleolus

Spherical body found within nucleus. synthesizes ribosomal RNA; disappear during cellular replication

3. Chromosome

Chromatin fibers are basic units of chromosome structure. They are also the fundamental basis of inheritance since they contain the genetic materials.

4. Nucleoplasm (nuclear sap)

Contains materials for building DNA and messenger molecules, which act intermediates between nucleus and cytoplasm

CHROMOSOME STRUCTURE AND FUNCTION

Introduction

Chromosomes are the darkly stained bodies seen during the metaphase stage of mitosis. Strasburger discovered chromosomes in 1875 and the term chromosome was coined by Waldeyer in 1888. Chromosomes are composed of thin chromatin threads called chromonemeta. These chromonemata undergo coiling and super coiling during prophase and it become readily observable by the light microscope. The main features of eukaryotic chromosomes are given below:

1. Chromosomes are clearly visible during mitotic metaphase. Hence, they are studied during metaphase.
2. Chromosomes bear genes in a linear fashion and thus are concerned with transmission of characters from generation to generation.
3. Chromosomes of eukaryotes by a nuclear membrane.
4. Chromosomes vary in shape, size, and number in different species of plant and animals.
5. Chromosomes have property of self-duplication, segregation and mutation.
6. Chromosomes are composed of DNA, RNA and histones. DNA is the major genetic constituent of chromosome.

Chromosome shape

Chromosome shape usually observed during anaphase. The shape of chromosomes is determined by the position of centromere, a part of chromosome on which spindle fibres are attached during metaphase. Chromosomes have generally three different shapes, viz., rod shape, J shape and V shape. These shapes are observed when the centromere occupies terminal, sub-terminal and median (middle) position on the chromosomes, respectively.

Chromosome size

Chromosome size is measured at mitotic metaphase. It is measured in two ways viz., in length and diameter. Plants usually have longer chromosomes than animals. The maximum length of chromosome is observed during interphase and minimum during anaphase.

Chromosome size varies from species to species. Maize chromosomes have the length of 8-12 μ. Giant chromosomes have length upto 300 μ.

Chromosome number

Each species has definite and constant somatic and gametic chromosome number. Somatic chromosome is the number chromosomes found in somatic cells and it is represented by diploid number (2n). The somatic cells contain two copies of each chromosome (except sex chromosome) one of which is inherited from father while other is inherited from mother. These two chromosomes are identical in morphology, gene content and gene order they known as homologous chromosomes. Gametic cells or gametes contain one half of the somatic chromosome number which is represented by haploid number (n).the gametic chromosome number of a true diploid species is called basic number. It is the minimum haploid chromosome number of any species, which is denoted by x. for example, in wheat, the basic number is 7, whereas the haploid number is 7, 14 and 21 for diploid, tetraploid and hexaploid species, respectively. Thus, haploid chromosome number differs from basic number. Both are same in case of true diploid species but differ in case of polyploidy species.

Number of chromosomes varies from 2n = 4 (n = 2) in *Haplopappus gracilis* (compositae) to 2n = >12000 in some pteridophytes. In plant kingdom, chromosome number is higher in dicots than in monocots.

Chromosome structure

The structure of chromosome becomes easily visible during metaphase due to coiling of interphase chromosomes. Each chromosome consists of seven parts, viz., 1) Centromere 2) chromatid 3) secondary constriction and satellite 4) Telomere 5) chromomere 6) chromonema 7) matrix. A brief description of these parts is given as below,

1. **Centromere (Primary constriction)**

It is a localized region of the chromosome with which spindle fibres are attached during metaphase is known as centromere or primary constriction or kinetochore. Centromere has four important functions, viz., (i) orientation of chromosomes at metaphase, (ii) movment of chromosomes during anaphase. (iii) formation of chromatids, (iv) chromosome shape. Centromere may occupy various position on the chromosomes, viz., terminal, sub-terminal, median etc. generally each chromosome has one centromere, but in some cases, the number of

centromere may vary from nil to many. The centromere divides the chromosome into two arms of varying length.

2. Chromatid

One of the two distinct longitudinal subunits of a chromosome is called as chromatid. These subunits of a chromosome get separated during anaphase. Chromatids are of two type's *viz.*, sister chromatids and non-sister chromatids. Sister chromatids originate from homologous chromosomes. Chromatids are formed due to chromosome and DNA replication during interphase. Two chromatids of a chromosome are held together by anaphase each chromatid becomes a chromosome.

3. Secondary Constriction

Some chromosome exhibits secondary constriction in addition to primary constriction. It may be present either in short or long arm of the chromosome. The chromosomal region between secondary constriction and nearest telomere (end of the chromosome) is called as satellite or trapant. The chromosome having satellite is known as satellite chromosomes. The position of secondary constriction in the chromosome is constant. The number of satellite chromosomes in a genome varies from species to species.

4. Telomere

The two ends of the chromosomes are known as telomeres. Telomeres are highly stable and they do not fuse or unit with telomeres of other chromosomes. The structural integrity and individuality of the chromosome is maintained by telomeres.

5. Neucleolar Organizer Region (NOR)

During interphase, nucleolus of the cell is always associated with secondary construction of satellite chromosome. So the secondary constriction is also called as NOR. The NOR contain several copies of gene coding for ribosomal RNA.

6. Chromomeres

The chromosome of some of the species show small bead like structures called as chromomeres. The distribution of chromomeres in the chromosome is constant. Available evidence indicates that chromomere represents a unit of DNA replication, chromosome coiling, RNA synthesis and RNA processing.

7. Chromonema

Under light microscope, thread like coiled structures are found in the chromosomes and chromatids which are called chromonema (plural chromonemata). Chromonema is considered to be associated with three main functions. It controls size of chromosomes, results in duplication of chromosomes and is the gene bearing portion of chromosomes.

8. Matrix

A mass of acromatic material in which chromonemata are embedded is called matrix. Matrix is enclosed in a sheath which is known as pellicle. Both matrix and pellicle are non genetic materials.

Karyotype

Karyotype is a phenotypic appearance of chromosomes of a particular species. In the study of karyotype, various features of chromosomes are taken into account *viz.*, (i) number (ii) position of centromere (iii) size (iv) possibility of satellite (v) degree and distribution of heterochromatin etc. it is represented by arranging the somatic chromosome complements according to their length keeping their centromeres in straight line. Thus the longest chromosome is placed in the extreme left and smallest in the extreme right.

Idiogram

Diagrammatic representation of morphological features of haploid chromosome complements of a species is known as ideogram.

Heterochromatin

The region of the chromosome, which takes up deep stain during interphase and prophase, is called heterochromatin. It is classified into two type, constitutive heterochromatin and facultative heterochromatin.

i. **Constitutive Heterochromatin:** These regions of the chromosome remains permanently in the heterochromatin stage. i.e. it does not revert to euchromatic stage.

ii. **Facultative Heterochromatin:** It is the region of the chromosome which undergoes euchromatin stage.

Euchromatin:

The region of the chromosome, which takes up little stain during interphase, is called Euchromatin. It is the active region of the chromosome, involved in transcription.

Classification of chromosomes

Chromosomes can be classified in different ways. The various criteria which are usually used for the classification of chromosomes include, (a) position of centromere, (b) number of centromere, (c) shape at anaphase, (d) structure and appearance, (e) role in sex determination, and (f) structure and function. A brief classification on the bases of these criteria is presented below:

A. Position of centromere

1. Metacentric chromosome

A chromosome in which centromere is located in the middle portion, such chromosomes assume V shape at anaphase

2. Sub-metacentric chromosome

A chromosome in which centromere is located slightly away from the centre point or has sub median position, such chromosomes assume J shape at anaphase.

3. Acrocentric chromosome

A chromosome in which centromere is located very near to one end or has sub- terminal position, it is called as sub-terminal chromosome. Such chromosome assumes J shape or rod shape during anaphase.

4. Telocentric chromosome

A chromosome in which centromere is located at one end is called as telocentric. Such chromosome assumes rod shape during anaphase.

5. Holokinetic chromosome

A chromosome with diffused centromere. Centromere does not occupy a specific part o chromosome. Whole body of such chromosome exhibits centromeric activity.

B. Number of centromere

1. Acentric chromosome

A chromosome without centromere. Such chromosome remains as laggard during cel division and is eventually lost.

2. Monocentric chromosome

A chromosome with one centromere. It represents normal type of chromosomes.

3. Dicentric chromosome

A chromosome having two centromeres. Such chromosome makes dicentric bridge at anaphase and are produced due to inversion and translocations.

C. Shape at anaphase

1. V shape chromosome

A chromosome which assumes V shape at anaphase. It includes metacentric chromosome.

2. J shape chromosome

A chromosome which assumes J shape at anaphase. It includes sub-metacentric and sub-terminal chromosomes

3. Rod shape chromosome

A chromosome which assumes rod like shape during anaphase

D. Structure and appearance

1. Linear chromosome

A chromosome with linear structure or having both the ends free. Such chromosomes are found in eukaryotes.

2. Circular chromosome

A chromosome with circular shape and structure. They are found in bacteria and viruses.

E. Essentiality

1. A-chromosome

Normal members of chromosome complements of a species which are essential for normal growth and development.

2. B-chromosome

Chromosome which is found in addition to normal chromosome complements of a species. They are also called as accessory, supernumerary or extra chromosomes and are not essential for normal growth and development.

F. Role in sex determination

1. Allosomes

Chromosomes which differ in morphology and number in male and female sex and contain sex determination gene. They are generally of two types, *viz.*, X and Y or Z and W type.

2. Autosomes

Chromosome which do not differ in morphology and number in male and female sex and rarely contain sex determining genes.

G. Structure and function

1. Normal chromosome

Chromosome with normal structure (shape and size) and function.

2. Special chromosome

Chromosomes which significantly differ in structure and function from normal chromosomes. Such chromosomes include lamp brush chromosomes and B- chromosomes.

Composition of chromosome

Chemical composition: It contains DNA, RNA, histones, and non-histone proteins

i. **DNA / RNA**- heredity material

ii. **Histone**- five fraction e.g. H_1, H_2A, H_2B, H_3, H_4. It hold together chromosome fibre

Functions of chromosomes

1. Contain genes- provide genetic information
2. Protect genetic material from chemical and mechanical damages
3. Regulation of gene function
4. Precise chromosome distribution due to centromeric function
5. chromosome stability due to telomeres

Chromosome Models

Chromatin fibres are the basic units of chromosome structure. Chromosome model refers to organization of chromatin fibres in a chromosome. Two models namely folded fibre model and nucleosome structure and organization. If chromatin fibre in a chromosome. These models are briefly described below:

1. Folded fibre model

This model was proposed by DuPraw in 1965. According to this model, chromatin fibres are about 230 Å in diameter. Each chromatid consists of single DNA double helix. The folding and super coiling of very long chromatin fibre causes reduction in length and increase in thickness of the chromosome.

2. Nucleosome-solenoid model

This model was proposed by Kornberg and Thomas in 1974. Chromatin is composed of DNA, RNA, histones and other proteins. Chromatin fibres are 300 Å in diameter. The nucleosomes are sub units of chromatin and have bead like appearance. Each nucleosome is composed of a histone octamer and 146 base pairs (bp) of DNA. Each nucleosome consists of a core particle and linker or spacer DNA. The core particle has two copies each of H_2B, H_3 and H_4 histone molecules. Thus it has a histone octamer. The core particle is about 100 Å in a diameter and 60 Å in height. A duplex DNA stand is tightly wound around this core particle making two circles. Spacer or linker DNA has four base pairs. One molecule of histone H_1 is connected with linker DNA. The super coiled nucleosome fibre is known as solenoid.

According to this theory, a very long molecule of DNA (146 bp) is packed into a single unit of nucleosome and several unit of nucleosome constitute chromatin fibre. The chromatin fibre of 300 Å which is visible under electron microscope at metaphase develops from the nucleosome fibres as a consequence of super coiling of latter. This model is universally accepted as a model of chromatin fibre organization.

SPECIAL TYPES OF CHROMOSOME

Introduction

Some tissues of certain organism contain chromosomes, which differ significantly from normal chromosomes in terms of either morphology or functions. Such chromosomes are referred to as special chromosomes following types of chromosomes may be included under this category. The giant chromosomes are (1) polytene (2) lampbrush chromosomes (3) accessory or B-chromosomes (4) sex chromosomes (5) artificial chromosomes or YAC (6) isochromosome (7) ring chromosome (8) super chromosome. These different forms of chromosomes their functions & significance given one by one by following.

Lamp brush chromosomes

Lamp brush chromosomes described in detail in shark oocytes by Ruckert in 1892. It was first observed in salamander (amphibian) oocytes in 1882 by flemming. Lamp brush chromosomes occur at the diplotene stage of meiotic prophase in primary of all animal species both vertebrates & invertebrates. During diplotene, homologous chromosomes begin to separate from each other, remaining in contact only at several points along their length. Each chromosome of pair has several chromomere distributed over its length. Chromomeres generally a pair of lateral loops extends in the opposite directions perpendicular to the main axis of chromosome. These lateral loops give these chromosomes the appearance of lamp brush which is reason for their name. These chromosomes are extremely long in some cases being 800-1000μ in length. These chromosomes 30 times less highly packed than meiotic chromosomes. The size of loops may range from an average of 9.5 μ in frog to upto about 200 μ in newt. Each loop is made up of fully extended single DNA double helix RNAase & protease digestion of loops generates DNAase susceptible 20 A thick fibers. One end of each loop is markedly thinner than is them other end. There is extensive RNA synthesis at the thin ends of loops, while there is little or no RNA synthesis at the thin ends. The chromatin fiber of chromomere is progressively uncoiled toward the thin end of loop, the DNA in this region supports active RNA synthesis, but later RNA becomes associated with protein to yield a matrix of ribonucleo proteins, this makes the loop markedly thicker. In some cases loops corresponding to particular genes have been isolated. The DNA at the thick end of loop is progressively withdrawn & resemble into chromomere. As oocytes progresses from diplotene to metaphase I the loop are slowly withdraw into chromomeres as normal chromosome.

Among the plant they reported in tomato, wild onion, fungus neurospora. Loops represent the site of gene action (transcription).

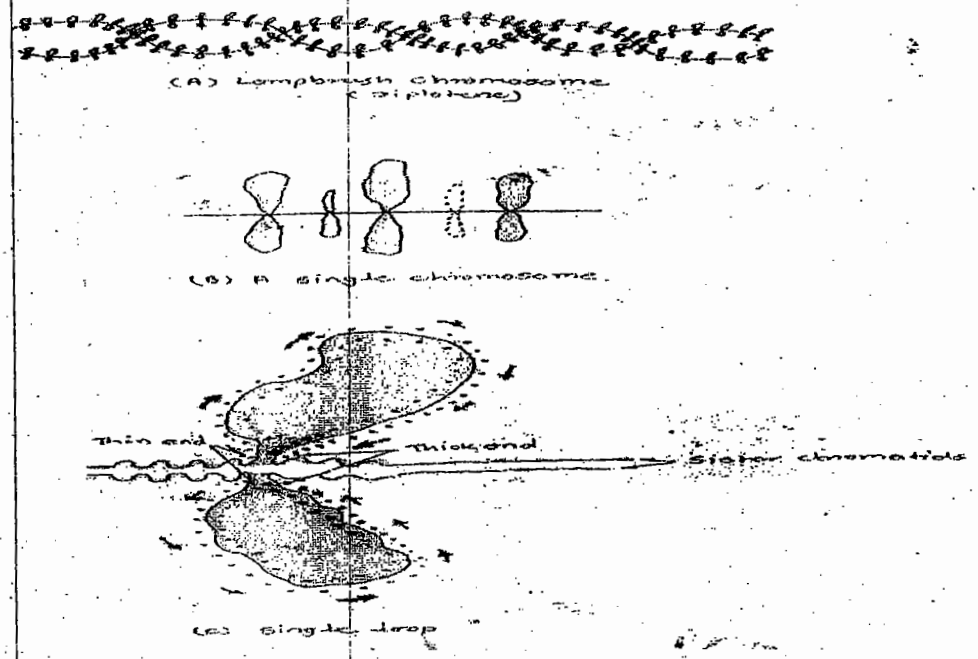


Fig : lampbrush chromosome

Function and significance of lamp brush chromosomes:

To produce large numbers & quantities of proteins & RNA_S stored in eggs. Gene transcription is those whose gene product is required during early stages of embryogenesis.

Giant chromosomes or polytene chromosomes:

An Italian cytologist E.G. Balbiani (1881) had observed peculiar structures in the nuclei of certain secondary cell. Unfortunately be did not recognize them as chromosomes. Painter, Heitz & Bauer rediscovered them in drosophila & recognized them as chromosomes. Since these chromosomes were discovered in the salivary gland cell they were called salivary gland chromosomes. A present name polytene chromosome was suggested by Kollar due to occurrence of many chromonemeta (DNA) in them. It is found in certain tissues e.g. salivary gland of larvae, gut epithelium, malphigian tubules & some fat bodies of some files e.g. Drosophila. Mosquito, Midges. It is also found in antipodal cell and suspensors of young embryos of many plants. These are formed by somatic pairing of homologues chromosomes, followed by 8-10 rounds of chromosome replication without any nuclear or cell division i.e. endoreduplication. In Drosophila salivary glands, the giant chromosomes radiate as five long & one short arm from amorphous structure called chromocentre. Chromocentre is formed by

fusion of centromeric regions of all the chromosomes. Short arm radiating from the chromocentre represents chromosomes IV, one of the long arms is due to the x-chromosomes while the remaining four long arms represents the arms of chromosome II & III. Total length of Drosophila giant chromosomes is about 2,000 μ while it's metaphase chromosome measure only 7.9 μ. Giant chromosomes contain several dark staining regions called bands & relatively light or non-staining inter-band regions. Total number of bands in four giant chromosomes of Drosophila is estimated as around 5,000. Some of these bands are as thick as 0.5 μ while some may be only 0.05 μ thick. In Drosophila the location of many genes is correlated with specific band. During certain stages of development specific bands & inter band regions associated with them greatly increase in diameter as compared to the rest of the chromosome, the structure are known as puffs or **Balbiani** rings. Puffs are believed to be produced due to uncoiling of chromatin fibers present in the concerned chromomers. These uncoiling chromatin fibers extend out side the chromosome in form of loop. The puffs are site of active RNA synthesis. Some hormones e.g. ecdysone, induce puffs in specific band directly others indirectly by the products of earlier puffs. Certain proteins, including RNA polymerase II accumulate at puffs. Transcription also occure in band & single active gene can gire rise to puff.

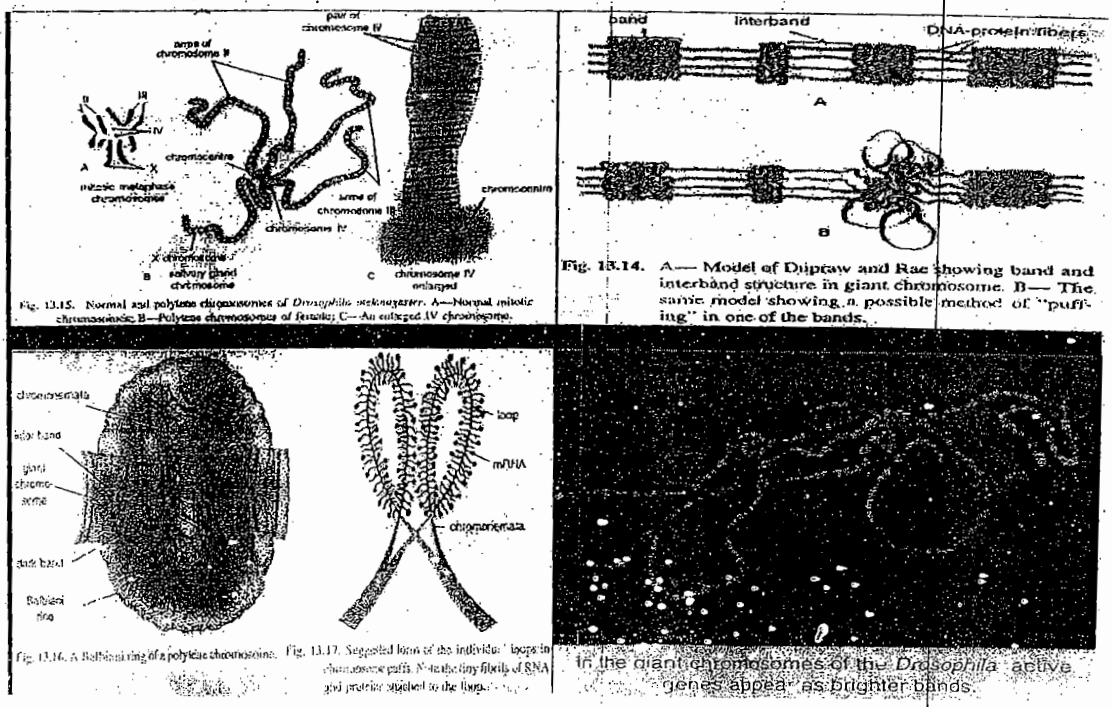


Fig. 13.15. Normal and polytene chromosomes of *Drosophila melanogaster*. A—Normal mitotic chromosomes. B—Polytene chromosomes of female. C—An enlarged IV chromosome.

Fig. 13.14. A— Model of Diptaw and Rae showing band and interband structure in giant chromosome. B— The same model showing a possible method of "puffing" in one of the bands.

Fig. 13.16. A Balbiani ring of a polytene chromosome. Fig. 13.17. Suggested form of the individual loops in chromosome puffs. N-marked tiny fibrils of RNA and protein applied to the loop.

In the giant chromosomes of the *Drosophila* active genes appear as brighter bands.

Fig: Giant chromosome

Function and significance of giant chromosome

- Puffs are site of RNA synthesis.
- It use for cytological map of chromosome.
- Transcription is Ist step in gene action.

Accessory chromosomes

In many species one to many chromosomes in addition to the normal somatic complement are found the extra chromosomes are called accessory chromosomes, B-chromosomes or supernumerary chromosomes. About 600 plant species & more than 100 animal species reported to possess supernumerary chromosomes. It is broadly similar to normal somatic chromosome in their morphology. But they have some peculiar function. It is generally smaller in size than the chromosomes of the normal somatic complement but some species they may be larger. It is generally gained by & lost from the individuals of species without any apparent adverse or beneficial effect. The presence of several B-chromosomes often leads to some reduction in vigour & fertility in maize. Origin of B-chromosomes in most species is unknown. In some animal species they may arise due to fragmentation of heterochromatic 'Y' chromosome. It is relatively unstable, show irregular distribution during meiosis. In plant species supernumeraries show non-disjunction during Ist or IInd microspore mitosis. In the former case it is passed on to the generative nucleus while in later case the sperm containing the accessory chromosomes preferentially fertilizers the egg cell. In some animal species it is to be transmitted through female only. It first studied by Randolph (1928) % give term B-chromosome. B-chromosome segregates normally. It delays the flowering time. Extensive study in plant maize, scale cereals. They are genetically inactive because these chromosome largely Heterochromatic. In many species origin & function unknown.

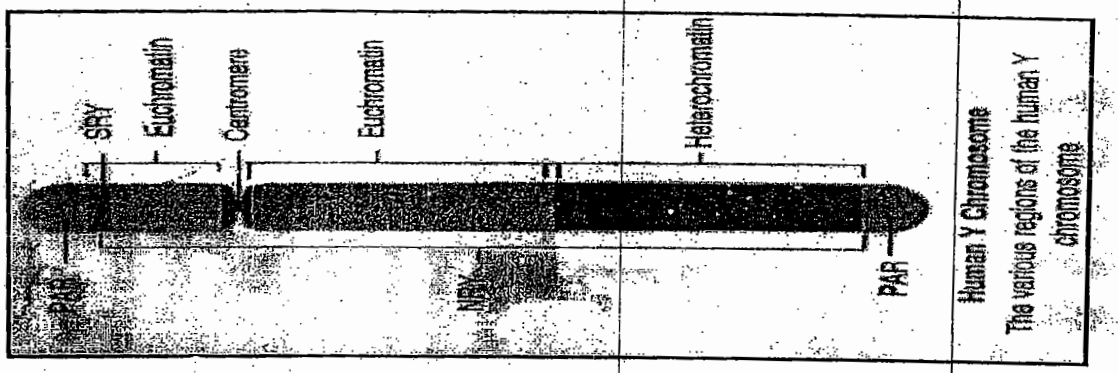
Function and significance of B-chromosome:

It is utilized in distant hybridization of wheat. The most significant effect of pollen fertility & seed set.

Sex chromosomes

German biologist, Henking in 1891 noted that half of certain insect contain in extranuclear region, Henking called this 'X-body'. Mc clung in 1902 explain its significance by observations in grass hoppers. Wilson & Miss Stevens realized that x-body was chromosome. So x body becomes known as x-chromosomes. Miss Stevens in 1905 found that

male & females of beetle have the same number of chromosome, but one of the pairs males is heteromorphic one member of pair in the heteromorphic appears identical to the member of pair in the female. She called this the x-chromosome; other member of the heteromorphic pair is never found in females she called this in 'y-chromosome'. The X & Y sex chromosomes exhibit structural difference. Cytological studies have shown that the x-chromosome of most organisms is straight rod like & comparatively larger than Y-chromosomes. The Y-chromosome is smaller in size with one end slightly curved or bent to one side in Drosophila. X-chromosome has large amount of euchromatin & small amount of heterochromatin. The euchromatin has large amount of DNA material hence much genetic information. Y-chromosome contains small amount of euchromatin & large amount of heterochromatin. The Y-chromosome has little genetic information. There fore sometimes it is referred to as genetically inert or inactive.



Function and significance of sex chromosomes

Y & X chromosome determine the sex of the organism. Yeast is microorganism that is very easy to maintain in large scale in lab. Yeast is important eukaryotic host for recombinant DNA. It's haploid DNA contain only 1.4×10^6 bp. Different type of vectors used in yeast may be grouped as follows.

Yeast artificial chromosomes

Yeast is microorganism that is very easy to maintain in large scale in lab. Yeast is important eukaryotic host for recombinant DNA. It's haploid DNA contain only 1.4×10^6 bp. Different types of vectors used in yeast may be grouped as follows.

- Yeast Episomal plasmid (YEP)
- Yeast Replicating plasmid (YRP)
- Yeast centromere plasmis (YCP)

- Yeast artificial chromosome (VAC)

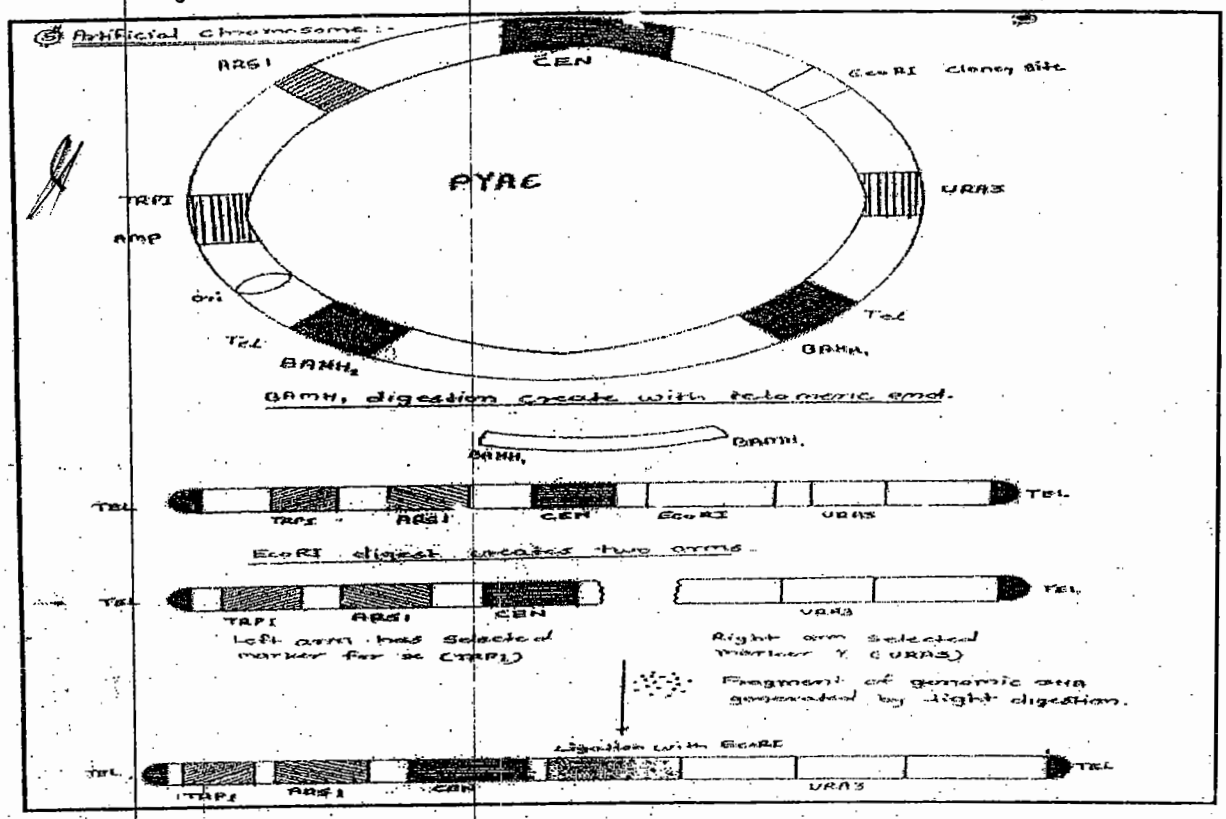


Fig: Yeast artificial chromosome

YAC resemble the linear structure. Szosatak & black burn in 1982 develop 1st vector which could be maintain as lycan molecule there by mimicking a chromoscme by cloning yeast telomere into a YRP such vector are known as YAC. Method of cloning large DNA sequences in YAC where developed by Burke *et al.* (1987). Yeast genes present in different yeast vector can become integrated into the host genome this is called permanent transformation. It generally occurs through homologues recombination between the gene present in a vector & that present in the yeast chromosome. Rarely the gene may become inserted at random chromosome site. The homologues recombination may occur by regular crossing over gene conversion. Very large DNA sequence clone in YAC. YAC contain all the essential feature of chromosome required for it's propagation in yeast cell including.

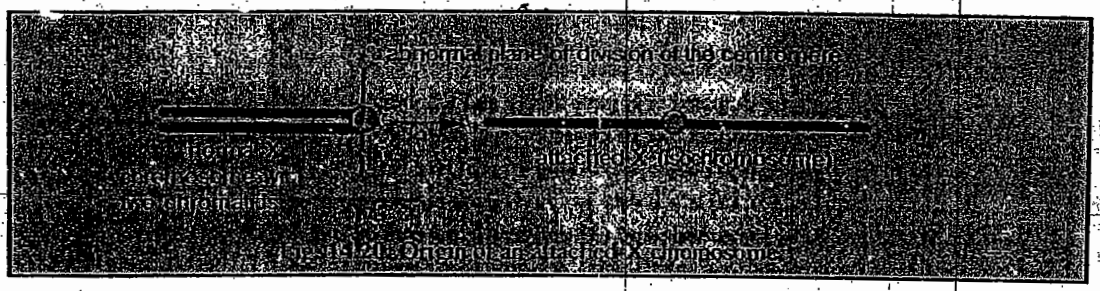
- An ARS sequence for replication
- A CEN sequence for centromeric function
- Telomeric sequence at the two ends for protection

- One or two selected marker e.g. TRP 1 & URA 3
- The necessary sequences from an E. coli plasmid
- Telomeric sequence in yeast chromosomes is a 20-70 tandem repeat of the 6 base sequences 5 'CCCC AA 3' it's complementary 5 'TTGGGG 3' in other strand.

Function and significance: It is a vector used to clone large DNA fragment

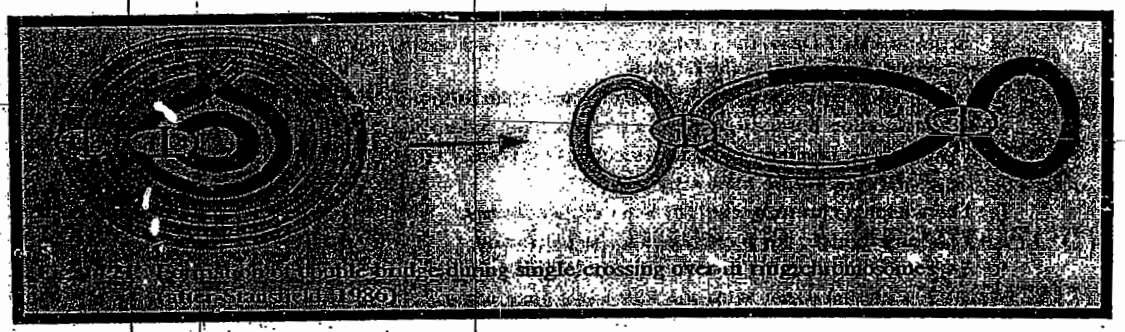
Iso-chromosomes

An iso-chromosome is chromosome in which both arms are identical. It is thought to arise when centromere divides in the wrong plane yielding two daughter chromosomes each at which carries the information of one arm only but present twice e.g. Telocentric x chromosome of Drosophila may be changed into an "attached-x" which is formed due to mis division of the centromere.



Ring chromosomes

Chromosomes are not always rod-shaped occasionally ring chromosomes are uncounted in higher organism. Some times breaks occur at each end of the chromosome & broken ends are joined to form a ring chromosome crossing over between rings chromosomes can lead to bizarre anaphate figures.



Super chromosomes

Polytene is increased by infection with microsporidium parasite protozoan parasite (Thalophemia) such parasite cause the salivary gland to grow very large to produce super chromosomes.

CELL DIVISION

Introduction

Cell is a basic unit of structure and function in all living systems. The process of reproduction or formation of new cells from the pre-existing cells is referred to as cell division. [Cell division= Karyokinesis (nuclear division) + Cytokinesis (cell division)]

Types of cell division

I. **A mitosis**- Direct cell division
II. **Mitosis** - Indirect cell division
III. **Meiosis** - Reduction division

I. A mitosis

Asexual reproduction in unicellular organism (e.g. Bacteria, Protozoa) in short splitting of nucleus (after it's elongation it acquires dumbbell shape) results into two nuclei. Immediately followed by division of cytoplasm due to cytoplasmic constriction

II. Mitosis

The term mitosis was coined by Flemming in 1882. Mitosis is the process by which a cell nucleus divides to produce two daughter nuclei containing identical set of chromosomes to the parent cell. It is usually followed immediately by division of whole cell to form two daughter cells. This process is known as mitotic cell division.

Cell cycle

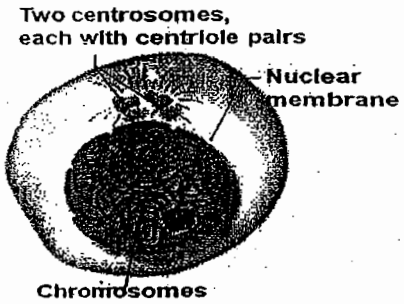
The sequence of an event which occurs between one cell division and the next is called cell cycle. It has two main stages.

1. Interphase
2. Mitosis or M phase

Interphase

It is the period between successive cell divisions consisting of process associated with growth and preparation for mitosis. The period of DNA synthesis during interphase is called the 'S' phase or synthesis phase and it is separated in time from the previous cell division by a gap called 'G_1' phase. After DNA synthesis a further gap called 'G_2' phase occurs before the

Interphase



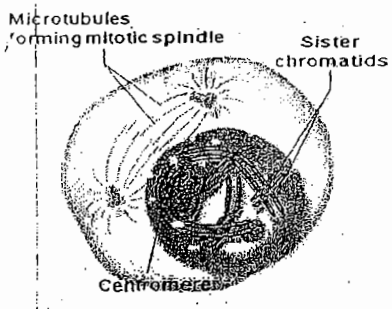
next cell division begins. The G_1 phase shows considerable variation where as G_2 show more constancy for a given type of cell. During interphase, each DNA molecule replicates an exact copy of itself. This copying process produces a chromosome with two identical function stands called chromatids, both attached to a common centromere.

Mitoticphase

The mitotic phase leads to separation of replicated DNA into two daughter nuclei without recombination. The M phase consists of two major events *viz.*, division of nucleus (Karyokinesis) followed by division of cytoplasm (Cytokinesis). The karyokinesis has got four distinct stages as follows:

1. **Prophase**
   - i. Coiling and condensation of chromosomes takes place which make them visible as thread like structures
   - ii. Each chromosome has two identical longitudinal splits called identical or sister chromatids, which are attached by common centromere.
   - iii. Migration of centrioles to opposite ends of the cell.
   - iv. Disappearance of nucleolus / and beginning of the breakdown of the nuclear membrane.
   - v. Formation of spindle fibre.

Prophase



2. Metaphase

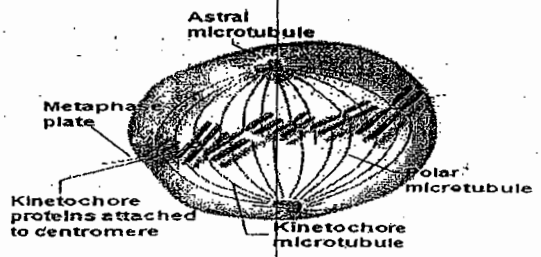
i. Formation of spindle fibres is completed and chromosomes are attached to the spindle fibres at the point of centromere.

ii. Movement and arrangement of all chromosomes on metaphase plate or equatorial plate.

iii. Sister chromatids of each chromosome are joined together at the point of centromere, but their arms are free.

iv. Chromosomes are clearly visible.

Metaphase



3. Anaphase

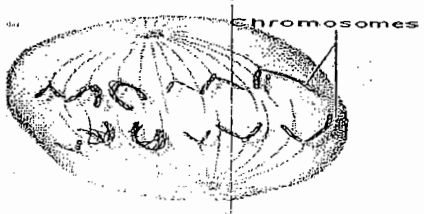
i. This is the shortest phase of the mitotic division.

ii. This stage begins with splitting of centromere into two, which allow the sister chromatids to separate and move to opposite poles.

iii. The separated sister chromatids are called as new chromosomes.

iv. The arms of each chromosome drag behind their centromeres giving them characteristics shapes depending upon the location of centromere.

Anaphase



4. Telophase

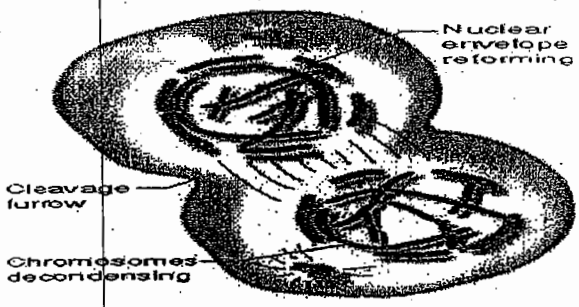
i. Chromosomes reach the opposite poles and spindle fibres begin to disintegrate.

ii. Nuclear membrane is reestablished.

iii. Nucleoli is reformed.

iv. Chromosomes again become thinner and longer by uncoiling and unfolding.

Telophase



Cytokinesis

It is the division of cytoplasm. This normally follows telophase and leads in to G_1 phase of interphase. In animals, cytokinesis is accomplished by formation of cleavage furrow which deepens and pinches the cell into two daughter cells. In plants, cytokinesis involves the construction of cell plate at the center of the cell and spreading laterally to the cell. Later cellulose and strengthening materials are added to the cekk plate converting it into a new cell wall.

Significance of mitosis

1. **Genetic stability:** Mitosis produced two daughter cells which have the same number of chromosomes as that of parent cell. These daughter cells are genetically identically to the parent cell and no genetic variation can be introduced during mitosis.
2. **Growth:** the number of cells within the organism is increased by mitosis and this is the basis for growth of organisms.
3. **Cell replacement:** Replacement of old cells and dead cells in an organism is achieved by mitosis.
4. **Asexual reproduction:** Production of new individuals through asexual reproduction is achieved by mitosis.

III. Meiosis

Meiosis is the process by which a cell nucleus divides to produce four daughter nuclei each containing half the number of chromosomes of the original nucleus or cell. It is also celled as reduction division since it reduces the number of chromosome in the cell from the diploid number (2n) to the haploid number (n). Like mitosis, it involves DNA replication during interphase in the parent cell, but this is followed by two cycles of nuclear and cytoplasmic division known as Meiosis I (reduction division) and Meiosis II (multiplication division). Thus a single diploid cell gives rise to four haploid cells.

First meiotic division or reduction division

It has four stages namely Prophase I, Metaphase I, Anaphase I, and Telophase I.

Prophase I

i. It is the longest phase of meiotic division. It has five sub stages namely, Leptotene, Zygotene, Pachytene, Dipotene and Dikinesis.

ii. Chromosomes are scattered throughout the nucleus in a random manner.

Leptotene

i. Progressive condensation and coiling of chromosome fibres.

ii. Chromosomes are scattered throughout the nucleus in a random manner.

Zygotene

i. Chromosomes become shorter and thicker.

ii. Homologous chromosomes lie side and this pairing process is called synapsis.

iii. Each synapsed homologue is called bivalent. It consists of four chromatid strands called tetrad.

iv. Synaptonemal complex also develops during this stage.

Pachytene

i. Exchange of chromosomal segments between non-sister chromatids of the homologous chromosome. This process is called crossing over.

ii. The point of exchange of chromatid during crossing over by *chiasma*.

iii. The homologous chromosomes are attached to each other by chiasmata.

iv. Synaptonemal complex can be seen between synapsed chromosomes.

Diplotene

i. Separation of homologous chromosomes takes place from one another which begin from the centromere to end of the chromosomes. This process is called terminalisation.

ii. Nucleolus decreases in size.

iii. Nuclear membrane disappears.

Diakinesis

i. This stage begins after the complete terminalization of chiasmata.

ii. Chromosomes are in more contracted stage.

iii. Due to further conuraction and terminalisation, these appear as round bodies evenly scattered throughout the cell.

iv. Nucleolus disappears.

v. The spindle fibres begin to be formed at the end of this stage.

Metaphase I

i. Homologous chromosomes lie on each side of the equatorial plate and attached with spindle fibres.

ii. Due to the contraction of spindle fibres, centromeres of each chromosomes are directed towards the equator.

Anaphase I

i. At first anaphase, the centromeres do not divide, but continue to hold sister chromatids together.

ii. The homologous separate and individual chromosome moves to opposite poles.

iii. This leads to reduction of number of chromosomes from diploid (2n) to haploid (n) state.

Telophase I

i. Chromosomes uncoil and relax and regrouping of chromosomes occurs.

ii. Nucleolus and nuclear membrane reappear.

iii. Two haploid daughter nuclei are formed.

iv. Cytokinesis in telophase I divides diploid mother cell into two haploid (n) daughter cells. This ends the first meiotic division.

The brief period between the first and second meiotic division is called **interkinesis.**

Second meiotic division or multiplication division

The second meiotic division is equal to mitosis division. However meiosis II differs from mitosis in the following ways.

i. Interphase (interkinesis) prior to meiosis II is very short. It does not have 'S' period because each chromosome already contains two chromatids.

ii. The two chromatids in each chromosome are not sisters throughout. In othrerwords some chromatids have alternate segments of non-sister chromatids due to recombination.

iii. The meiosis II deals with haploid chromosome number, where as normal mitosis deals with diploid chromosome number.

Meiosis II has four stages. They are Prophase II, Metaphase II. Anaphase II, and Telophase II.

Prophase II - the apparatus reappears.

Metaphase II - the Centromeres have lined up on the equatorial plane.

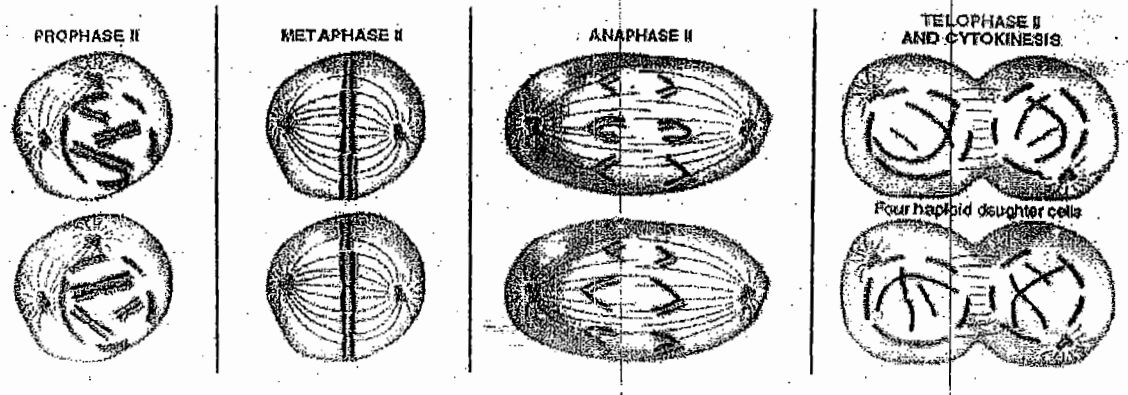
Anaphase II - the centromere of each chromosome divide and allowing sister chromatids to separate, cytokinesis followed by

Telophase II - divide two cells into four meiotic products.

Synaptonemal complex

It is a person framework, which is found between paired chromosomes. It consists of one central and two lateral elements. There are transverse filaments on both sides of the central element. The lateral elements are attached to homologous chromosomes. Synaptonemal complex is considered to be associated with pairing of homologous chromosomes and recombination. However its origin and exact role in synapsis is still not properly known.

Stages of Meiosis II



1 of each type of chromosome (n) in each daughter cell (gamete)

Significance of meiosis

1. Meiosis enables the chromosome number of a sexually reproducing species to be constant from generation to generation.

2. Meiosis introduces the genetic variation in the offspring of sexually reproducing individuals by means of independent assortment and crossing over (recombination).

CHROMOSOMAL ABERRATION

Introduction

Chromosomes may undergo changes. This is called chromosomal aberration or chromosomal mutation. The change may occur either in structure of the chromosomes or in the number of chromosomes. Based on these, the chromosomal aberrations are grouped into two major kinds- structural and numerical

Chromosomal aberration: Structural

There are four kinds of variation in the structure of chromosomes.

A. Intra chromosomal aberrations

1. Deletion (deficiencies)
2. Duplication (addition)
3. Inversion (paracentric and pericentric)

B. Inter chromosomal aberrations

1. Translocation (homozygotic and heterozygotic)

A. Intra chromosomal aberrations

1. Deletion

It is an "intra-chromosomal aberration" in which an interstitial or terminal chromosomal segment is lost. That is, some genes are deleted. Based on which it is called inter calary or terminal deficiency.

Cytological effect: In deletion heterozygotes, "deletion loop" occurs pairing of homologous chromosomes. The portion of the normal chromosome, homologous to the deficient segment bulges out.

Genetic effect: When a segment of a chromosome is absent, some genes are also absent. If these lost genes are physiologically important, deletion leads to death of the organism.

Deficiencies produce unique phenotypic effects in *Drosophila*. The character such as beaded, delta, gull, minute and notch are associated with some deletions in chromosomes.

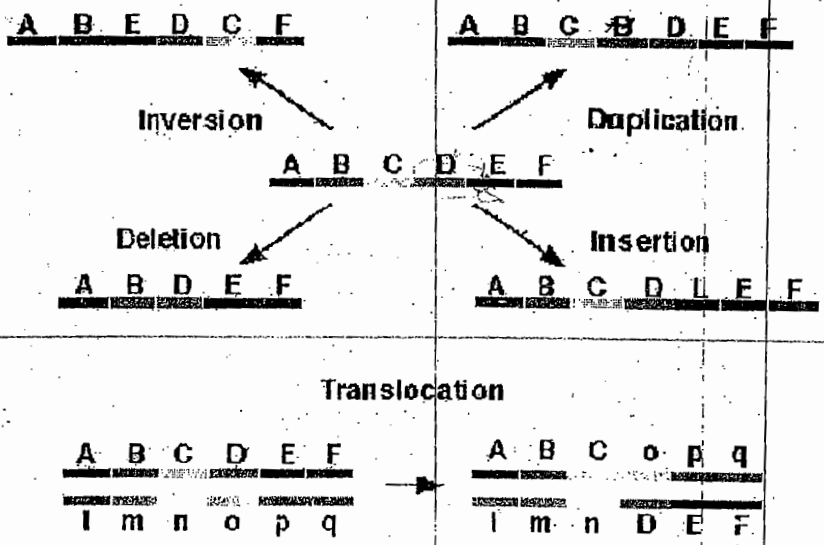
In human beings, 'Cri-du-chat' syndrome is characterized by a mewing cat during infancy. This 'Cri-du-chat' syndrome is caused by deletion in the short arm of 5th chromosome

2. Duplication

It is an 'intrachromosomal aberration' in which a segment is represented two or more times in a chromosome.

Cytological effect: During meiotic pairing of heterozygotes, the chromosome with duplicated segment forms a loop to maximize the juxtaposition of similar segments of homologous chromosomes.

Genetic effect: The duplications are not lethal. The unusual dosage of genes can be investigated. Duplications are useful in evolution of new character without loss of original traits. Relocation of chromosomal materials, due to duplication results in an altered phenotype. This is called position effect.



3. Inversion

It is an intrachromosomal aberration. Inversion occur when a part of chromosome become detached, turn through 180^0 and reinserted in such a way that the genes are in reverse order.

Inversions are of two kinds

i. **Pericentric inversion**

The inversion segment includes centromere.

ii. **Paracentric inversion**

The inverted segment does not include centromere. Centromere lies outside the inverted portion.

Origin of inversion: A chromosome may from a loop. Breakages occur at the point of intersection. When the sticky ends unite with new parents, inversion results.

Cytological effect: in inversion heterozygote the part of the uninverted chromosome corresponding to the inversion forms a loop. A similar loop is formed by the inverted section of the homologous chromosome but in reverse direction.

Genetic effect: Paracentric inversion produce dicentric and acentric chromosomes. Pericentric inversion produces duplication and deficies. Inversion acts as cross over suppressor and inversion maintains heterozygosity from generation to generation.

B. Inter chromosomal aberrations

1. Translocation

It is an inter chromosomal aberration where in exchange of chromosomal segments occurs between non-homologous chromosomes.

Cytological effect: In the translocation heterozygote, pairing of homologous chromosomal segments is effected by a cross-shaped configuration. This cross opens out into a ring as chiasma terminalizes. The meiotic products are of three kinds (i) normal (ii) balanced (iii) unbalanced.

Genetic effect: Translocation gives three kinds of genetic effects.

i. Translocation alters the linkage relationship of genes.
ii. Heterozygotic translocation causes semi sterility because most of the gamets fail to receive full, balanced complement of genes required for viable development.
iii. The phenotypic expression of a gene may be modified when it is translocated to a new position.

Chromosomal aberration: Numerical

Variation in number of chromosomes is called ploidy. A set of chromosomes present in an organism is called genome. In a genome each type of chromosome is represented only once. Most of the sexually reproducing plant species are diploid i.e. have two sets of chromosomes. Any change in the chromosome number from the diploid condition is referred to as heteroploidy. The heteroploidy is of two types namely,aneuploidy and euploidy. The variation in number may involve any particular chromosome or in entire sets.

Aneuploid

Loss or gain of one or more particular chromosomes occur within a set is called aneuploidy. The aneuploidy organism bears irregular number of chromosomes. Aneuploidy arises due to non-disjunction. Aneuploids are of three types.

Types of aneuploids

Types	Genomic constitution
A. Hypoploidy	
Monosomic	2n-1
Double monosomic	2n-1-1
Nullisomic	2n-2
B. Hyperploidy	
Trisomic	2n + 1
Double trisomics	2n + 1+ 1
Tetrasomic	2n + 2
Pentasomic	2n + 3

1. Monosomics

A monosomic is an individual that lacks one chromosome of the normal complement of somatic cells (2n-1). If the lost chromosome is one that is not absolutely essential for the organism, it may survive but if the lost chromosome is very important, the organism may not survive.

2. Nullisomics

A nullisomic is an individual that lacks both members of one specific pair of chromosomes (2n-2). A nullisomic diploid does not survive. However a nullisomic polyploidy (hexaploid wheat 6x-2) may survive but exhibit reduced vigour and fertility, nullisomic analysis helps to identify genes with specific chromosomes in a polyloid species.

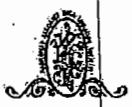
3. Polysomics

An individual having either single or one pair of extra chromosome in the diploid complement is known as polysomic. Polysomic analysis is also called as hyperploids. Polysomics are of two types (i) trisomics and (ii) tetrasomics.

i. Trisomics

A trisomic is an individual with one chromosome more than the normal complement of the somatic cell (2n+1). In general the extra chromosome does not produce so striking effect as a missing one. In wheat trisomic (2n = 43) are nearly indistinguishable from normal plants. Trisomic give rise to two kinds of gametes i.e. one kind with 'n' chromosome and other with 'n + 1' chromosomes. Trisomics are more stable genetically than monosomics.

ii. Tetrasomics



Addition of two chromosomes of one pair or two different pairs is known as tetrasomy and such individuals are called as tetrasomics.

Use of aneuploids

i. Aneuploids are extremely useful in several genetic studies.

ii. They are useful to determine the phenotypic effects of loss or gain of different chromosomes.

iii. Aneuploids have been used to produce chromosome substitution lines which give information on the effects of different chromosomes of a variety.

iv. They are used to produce alien addition and alien substitution lines which are useful in gene transfers from one species into another.

v. Aneuploid analysis permits the location of gene as well as of linkage group of a specific chromosome.

Aneuploids in Human beings

i) Down's syndrome

It is due to trisomic condition of 21st chromosome. It is also called Mongolian idiocy. Individuals are mentally deficit and physically retarded, broad face and flat stubby nose.

ii) Kline felter's syndrome (44+XXY)

It is due to trisomic condition of sex chromosome. The individual is male with XXY chromosome. The individuals with this syndrome have defective development of testis, famine character like Enlarged breast, under-developed body hair, presence of one barr body in body in the cells.

iii) *Turners syndrome (44+X)*

It is due to monosomic condition of sex chromosome. The individual is female with 44 autosomes and one 'X' chromosome. The female individual is without menstrual cycle. No barr body is present in body cells.

The origin of Aneuploids

i. Spontanebus

ii. Meiotic irregularities

iii. Triploid individuals

iv. Translocation heterozygote

Use of Aneuploids in crop improvement

i. Aneuploids are useful tools for locating the genes on a specific chromosome. Monosomics and nullisomics are used for this purpose.
ii. Monosomics are also used in interspecific gene transfer i.e. Monosomics are used in transferring chromosomes with desirable genes from one species to another.
iii. Aneuploids are used for developing alien addition and alien substitution lines in various crops.
iv. Primary trisomics are useful in identification of chromosomes involved in translocation.

Euploiy

These are variations that involve entire set of chromosomes. In Euploids the chromosome number is an exact multiple of the basic or genomic number. Euploids are differing in multiples of 'n' or 'x'.

Monoploid

The Monoploid organisms have one set of chromosomes or one genome (n) in the nuclei of their body cells. The monoploids are often weak and sterile. Monoploids differ from haploid which carry half or gametic chromosome number (n). In true diploid species, both monoploid and haploid chromosome number is the same (n = x). Thus a monoploid can be a haploid but all haploids cannot be monoploids.

Types	Genomic formula
Monoploid	n
Diploid	2n
Triploid	3n
Tetraploid	4n
Pentaploid	5n
Hexaploid	6n

Diploids

Normal diploids are known as disomics. They have regular bivalent pairing during meiosis. Diploids also have disomics genetic with two alleles at each locus.

Polyploids

Polyploids refer to any organism in which the number of chromosome sets exceeds two i.e. an organism with more than two sets of chromosomes or genome. They have larger cells than diploids. These larger cell sizes contribute to larger plant size and higher yield. Polyploids have generally larger, thicker and darker green leaves, bigger flowers, and fruits than the

diploids. In each genus, there is an optimum level of polyploidy beyond which growth may be depressed with increasing number of chromosomes. e.g triploid (3n).

There are two types of Polyploids.

i. Autopolyploid

In autopolyploids the multiple sets of chromosomes are identical e.g. genomes are identical with each other.

Autopolyploids arise by abnormal mitosis and meiosis and induced artificially by colchicines.

Auto triploid	3x	Banana 2n = 3x = 33
Auto tetraploid	4x	Groundnut 2n = 4x = 40
Auto hexaploid	6x	Sweet potato 2n = 6x = 90

Autotriploid

The triploid organisms have three sets of chromosomes. a triploid may originate by the union of monoploid gamets (n) with a diploid gamete (2n). Since an autotriploid remains sterile and cannot produce seeds, it has great commercial value in producing seedless varieties of economic plants. e.g. seedless watermelon.

Autotetraploid

The organism with four genomes (4n) in the nuclei of their somatic cells is called tetraploids. They arise due to somatic doubling of chromosome number. Doubling is accomplished by either spontaneously or it can be induced by chemicals such as colchicines.

ii. Allopolyploid

In allopolyploids the multiple sets of chromosomes are not identical. The chromosomes are of different origin. Generally each genome has two copies. Allopolyploid arise by crossing species of different genomic group.

e.g. Allotetraploid- two distinct genome ($2x_1 + 2x_2$)

Allohexaploid- three distinct genome ($2x_1 + 2x_2 + 2x_3$)

Amphidiploid

A species or types of plant derived from doubling the chromosomes in the F_1 hybrid of two species, is called an amphidiploid. In amphidiploid the two species are known.

E.g. *Gossypium hirsutum* - 2n = 4x = 52

G. barbadense - 2n = 4x = 52

Nicotiana tabacum - 2n = 4x = 48

Triticum aestivum - 2n = 6x = 42

Saccharum officinarum - 2n = 8x 80

Morphological and Cytological features of Polyploids are

i. Larger in size than diploid
ii. Generally more vigorous than diploids
iii. Slower in growth and late in flowering
iv. Polyploids may have reduced fertility than diploids

Role of Polyploids and their evolution

i. About 1/3 of angiosperms are Polyploids. These suggest that Polyploids have significant role in the evolution of crop species.
ii. Allopolyploids have contributed great extent in the evolution of plants than auto Polyploids.
iii. The identification of diploid parental species is primarily based on pairing between the chromosome of diploid and the Allotetraploid species.
iv. Allopolyploids combine the genome of different species; hence the resulting individuals differ from progenitor.
v. Evolution is a slow process; but due to allopolyploids new species originate very quickly.
vi. Polyploids have wider adaptation to different environmental condition than diploids.

Role of Polyploids in evolution of crops

1. Wheat

Triticum monococcum x *Aegilops speltoides*
AA **BB**
n = 7 n = 7

↓

AB (2n = 14)

↓ (spontaneous chromosome doubling)

Aegilops squarrosa x *Tetraploid emmer wheat*
DD ***AABB***
n = 7 n = 14

↓

ABD (2n = 21)

↓ (spontaneous chromosome doubling)

AA BB DD (Hexaploid wheat)
2n = 42

Amphidiploid

Fig: Evolution of hexaploid wheat

2. Nicotiana / Tobacco

Nicotiana sylvestris
n =12

x

N. tomentosa
n = 12

↓

F_1

↓ (chromosome doubling)

N. tobaccum (cultivated tobacco)

Fig: Evolution of cultivated tobacco

3. Cotton

G. arboreum
AA
n = 13

x

G. raimondi
DD
n = 13

↓

AD

↓ (chromosome doubling)

AA DD (*G. hirsutum)* tetraploid cotton
2n = 26

Fig: Evolution of tetraploid cotton

MENDELIAN PRINCIPLES

Introduction

Gregor Johann Mendel, father of genetics was borne in 1822 to a family of poor farmer near Brunn in Austria, now it is part of Czechoslovakia. During young age, his education was seriously hampered by poverty and in order to continue his education he entered the Augustinian monastery at Brunn and was ordained a priest in 1847. A few years later, he was sent to the University of Vienna for training in physics, mathematics and the natural sciences. Although his performance at the University was not outstanding, his training provided him with many technical and mathematical skills that were of value in performing his later experiments. After completing his studies, he returned to Brunn in 1854 and he became a teacher and in 1857 began his famous experiments on peas in the monastery garden. After seven years of experimentation he presented his finding before the Natural History Society of Brunn during 1865. This paper was published in the annual proceedings of the society in 1866 entitled "Experiments in Plant Hybridization" and he died in 1884.

Rediscovery of Mendel's findings

Mendel's work was not recognized until 1900. In 1900, his finding was rediscovered independently by three scientists Hugo De Vries of Holland, Carl Correns of Germany and Eric von Tschermak of Austria. Then only the significance of Mendel's work was realized.

Mendel's choice of materials

i. Pea plants have constant clear-cut alternatives of characters.
ii. These are annual plants. They could be grown and crossed easily.
iii. They are normally self-fertilizing. But cross-fertilization can be done easily.
iv. Hybrids are fully fertile.

Merits of Mendel's method

Mendel was able to discover the law of inheritance because of his intelligent methods of study and by the application of mathematics.

i. Mendel selected garden pea which is a self-fertilized short duration crop.
ii. He always used pure breeding parents and did crossing experiments with in the same species.
iii. He confined his study only on one character at a time.

iv. After finding the inheritance of characters separately, he studied two characters together.

v. He counted the number of each type of the progeny and he analyzed his numerical results in the form of ratios i.e., he applied mathematics to his findings.

Characters studied by Mendel in pea plants

Mendel studied seven pairs of contrasting characters. They are

Characters	Dominant form(wild type)	Recessive form
Seed characters		
i.) Seed shape	Round	Wrinkled
ii.) Seed coat colour	Grey	White
iii.) Cotyledon colour	Yellow	Green
Pod characters		
iv.) Pod colour	Green	Yellow
v.) Pod shape	Inflated	Constricted
Flower characters		
vi) Flower position	Axial	Terminal
Stem character		
vii.) Length/ Height of stem	Tall	Dwarf

Law of segregation

Mendel's experiment

Mendel tested the seven characters individually, by crossing a variety carrying a particular trait of a character (e.g. Tall) with another variety carrying a different trait of the same character (e.g. Dwarf). When he crossed tall variety with dwarf, he obtained only tall plant in F_1 generation. When the F_1 plants were selfed the tall and dwarf plants were segregated in 3:1 ratio in F_2 generation.

He made crosses for all seven characters and results appeared to fit the following pattern.

1. When crosses were made between the parents having contrasting characters, the F_1 always showed one of the parental traits and not the other.

2. The traits that had disappeared (or) hidden in the F_1 were reappeared in the F_2 generation, but only in the frequency of one quarter of the total number.

3. Irrespective of which parent used as male or female, both direct and reciprocal crosses gave the same result.

Parent	Tall	x		Dwarf
Génotype	TT	↓		tt
Gamets	(T)			(t)
F_1		Tall (Tt)		
		↓ (selfing)		
		Tall x	Tall	
Gamets		(T) (t)	(T) (t)	

♂ \ ♀	T	t
T	TT (pure tall)	Tt (hybrid tall)
t	Tt (hybri tall)	tt (pure dwarf)

F_2	3/4 (Tall)		:	1/4 (Dwarf)
Phenotypic ratio	1 (TT): 2(Tt)		:	1 (tt)
Genotypic ratio	3		:	1
	1/3 of Tall	2/3 of Tall	:	1/1 of Dwarf
	↓	↓		↓
F_3	Tall Only	Tall: Dwarf 3: 1	:	Dwarf only

4. Mendel called the determining agent responsible for each trait a 'factor' from the evidence of F_1 and F_2 generation, the 'factor' that determine each trait could be hidden but not destroyed.

5. Though the F_1 hybrids contain factor for both Tall and Dwarf traits, all the F_1 were tall. The phenomenon of the suppression of expression of one trait by another is called as dominance. The trait which expression is suppressed in F_1 is called recessive. In Mendel's experiment tall is a dominant character designated by capital letter 'T' and dwarf is a recessive character designated by small letter's'.

6. According to the symbols used the Tall hybrids 'F_1' contains factor T (since it is tall) and't' (since it produces some dwarf plants in F_2). Since T is dominant over t, the Mendel's dwarf plant contains.

7. To find out the number of factors controlling each trait Mendel selfed the plants in F_2 generation. The 'dwarf' plants upon self fertilization always gave rise (breed true) to dwarf in all the generations. Whereas tall plants breed true and 2/3 of the tall F_2 plants upon self fertilization produce tall and dwarf plants in 3:1 ratio.

8. From the results, it is clear that the hybrid plant for tallness contains two factors 'T' and't'. The true breeding tall and dwarf plants also contains TT and tt respectively. So the true breeding line produce only one kind of gamete, whereas, the hybrid tall plants contain two

kind of factors which can separate or segregate from each other during gametogenesis and produce two kinds of gametes in equal proportion. The random combination of these gametes leads to the production of tall and dwarf progeny in the (phenotypic) ratio of 3:1 and with the genotypic ratio of 1:2:1.

Law of segregation (law of purity of gametes)

"Allelic genes in a hybrid do not blend or contaminate each other, but segregate and pass in to different gametes during gametogenesis".

For example, the F_1 hybrids (Tt) of a monohybrid cross between tall (TT) and dwarf (tt) pea plant have one dominant allele (T) for tallness and one recessive allele (t) for dwarfness.

Though the tall and dwarf alleles remain together but does not contaminate or mix with any one. Both the alleles segregate to produce gametes either having dominant allele 'T' or recessive allele't'. The law of segregation is universal in its application and it has been found to occur in both plants and animals.

Chromosomal basis of segregation

In the anaphase stage of meiosis, the members of homologous chromosome pair segregate or separate from each other and move to opposite poles. As a result the allelic gene/present in the homologous chromosome are also separated and carried to opposite poles.

Terminology

Gene

An inherited factor that determines biological characteristics of an organism is called gene. In modern term, gene may be a segment of DNA which code for single polypeptide chain.

Allele

Alternative form of a gene occupying the same locus of the homologous chromosome

Locus

The position of gene on the chromosome

Dominance

The suppression of expression of one allele by another allele of the same gene is called dominance.

Recessive

The characters which lack the ability to express in F_1 generation are called recessive.

Genome - Set of chromosome

Genotype

Genetic constitution (make up) of an individual is called genotype. The genotype of tall plant is TT/Tt and dwarf plant is 'tt'.

Phenotype

External appearance of an individual produced by the genotype in co-operation with the environment is called phenotype.

Character

It is any morphological, anatomical, biochemical or behavioral feature of an organism.

Hybrid / F_1 generation (F_1= *first filial or progeny generation;* 'filial' is derived from a Latin word meaning 'son' or daughter)

Progeny obtained by crossing two parents (genetically dis-similar) is called hybrid.

Parents

Varieties / strain differing for one or more characters and which are involved in crossing are called as parents.

Female parent / ♀

Parental plant whose stigma is pollinated by pollens of desired male parent is called as female parent.

Male parent / ♂

Parental plant whose pollens are used for pollination of desired female parent is called male parent.

Pollination

Transfer of pollen of desired male parent to receptive stigma of female parent is known as pollination.

Pollinating agent- hand pollination, wind, water and insect

Emasculation

Process of removal of all immature anthers or even androecium of bisexual flower is called as emasculation.

Xenia

Effect of pollens on embryonic and maternal tissues is called as xenia.

Homozygous (TT/tt)

Individual having identical alleles for a character is known as homozygous genotype.

Metaxenia - effect of pollens on maternal tissues,

Heterozygous (Tt)

Individual having different alleles for a character is known as heterozygous genotype.

Monohybrid

The F_1 offspring produced by crossing two true breeding parents, which are differ in one character only. Monohybrid individuals are heterozygous at one locus.

Dihybrid

The F_1 offspring produced by crossing two true breeding parents, which are differ in two characters. Dihybrid individuals are heterozygous at two loci.

Polyhybrid

The F_1 offspring produced by crossing two true breeding parents, which differ for more than three characters, are known as polyhybrid.

Reciprocal cross

A cross in which both the parents are involved vice-versa.

Back cross

Cross of F_1 hybrid with either of parent

Test cross

It is a cross between the F_1 hybrid and its recessive parent. Note- Test cross is always back cross but back cross may not be test cross. ratio should be always 1:1

Used of test cross

1. **Test cross verifies the Mendel's factorial hypothesis**

According to Mendel, a monohybrid tall (Tt) produces two kinds of gametes in equal proportion and recessive parent produce only one kind of gamete't'. Hence this back cross should give tall and dwarf plants in 1:1 ratio. In actual experiment also we get tall and dwarf in 2:1 ratio. Thus Mendel's factorial hypothesis is verified.

2. **Test cross is used for identifying the genotype of an unknown parent**

A tall pea plant may be either homozygous (TT) or heterozygous (Tt). Its genotype may be determined by test cross. If the test cross progeny were tall, then the unknown tall genotype is 'homozygous'. If that test cross progeny have tall and dwarf plants in equal proportion, then the unknown genotype is heterozygous.

Incomplete dominance

When F_1 hybrid do not resemble to any of the parent but more or less intermediate between these parents; then it is termed as 'incomplete dominance'.

Parent	Red	x	White
Génotype	RR	↓	rr
Gamets	(R)		(r)
F_1		Pink (Rr)	
		↓ (selfing)	
F_2	Pink	x	Pink
Gamets	(R) (r)		(R) (r)

♂ \ ♀	R	r
R	RR (pure red)	Rr (hybrid pink)
r	Rr (hybrid pink)	rr (pure white)

Phenotypic and Genotypic ratio **1 : 2 : 1**
Red : Pink : White

Co-dominance

When both the alleles of a pair are fully expressed in heterozygous, they called co-dominance.

Law of independent assortment

Mendel's Experiment

After studying the characters independently, Mendel dealt two characters, such as colour and shape of the seeds together. He crossed a pure line of yellow round seeds with a pure line of green wrinkled seeds. The F_1 dihybrids received genes for yellow and round characters from one parent and genes for green and wrinkled characters from another parent. Since yellow is dominant over green and round is dominant over wrinkled, all F_1 offspring were uniformly yellow round. When F_1 offspring were self fertilized, they produced four kinds of offspring in F_2 in 9:3:3:1 ratio.

Parent	Yellow Round	x	Green Wrinkled
Génotype	YYRR	↓	yyrr
Gamets	(YR)		(yr)
F_1		Yellow Round (YyRr)	
		↓ (selfing)	
F_2	Yellow Round	x	Yellow Round
Gamets	(YR) (Yr) (yR) (yr)		(YR) (Yr) (yR) (yr)

♀ \ ♂	YR	Yr	yR	yr
YR	YYRR Yellow Round	YYRr Yellow Round	YyRR Yellow Round	YyRr Yellow Round
Yr	YYRr Yellow Round	YYrr Yellow Wrinkled	YyRr Yellow Round	Yyrr Yellow Wrinkled
yR	YyRR Yellow Round	YyRr Yellow Round	yyRR Green Round	yyRr Green Round
yr	yYRr Yellow Round	Yyrr Yellow Wrinkled	yyRr Green Round	yyrr Green Wrinkled

Phenotypic ratio — 9 Yellow Round : 3 Yellow Wrinkled : 3 Green Round : 1 Yellow Wrinkled

Genotypic ratio — 1:2:2:4 : 1:2 : 1:2 : 1

Mendel's interpretation

The factor for yellow colour many be represented by 'Y' and the green by 'y'; the round seed by 'R' and wrinkled by 'r'. Thus, a pure line yellow round is YYRR and a pure line green wrinkled is 'yyrr'. The gametes produced by these parents are YR and yr respectively. The F_1 offspring formed by the union of these gametes have the genotype of Yy Rr. The phenotype is yellow round. Since the parents differ in two characters, this offspring is called dihybrid.

During gametogenesis, the F_1 dihybrid produces four kinds of gametes YR, Yr, yR, yr, because, the segregation of the seed colour alleles occurs independently of the segregation of the seed shape alleles. This is called law of the independent assortment.

Law of independent assortment

"The segregation of one pair of allele is independent of the segregation in any other pair of allele".

In other words, when two or more independent characters are considered together, the factors responsible for them assort themselves freely and at random when gametes are formed.

Chromosomal basis of independent assortment

Among two pairs of homologous chromosomes, the members of one pair move to opposite poles independently of the members of the other pair. As a result of this independent assortment of chromosomes, the genes present in the non-homologous chromosomes undergo independent assortment.

The four kinds of gametes, YR, Yr, yR, and yr produced by F_1 dihybrid unite at random and produce 16 types of offspring in F_2 generation in the ratio of 9 Yellow Round, 3 Yellow Wrinkled, 3 Green Round and 1 Green Wrinkled.

This ratio shows that each pair of alleles behaves independently and bears no permanent association with other pair of alleles. The allele 'Y' is found along with the alleles 'R' in the parent. But it does not always remain associated with it. The allele 'Y' becomes assortment of alleles form the basis for the 9:3:3:1 ratio.

Dihybrid Test Cross

A Dihybrid YyRr is crossed with the double recessive parent yyrr. The Dihybrid produces four kinds of gametes *viz.* YR, Yr, yR and yr, in equal proportions. The green wrinkled produces only one kind of gamete. The expected result is Yellow round, yellow wrinkled, green wrinkled and green round in 1:1:1:1 ratio in actual experiment, the same ratio was obtained.

Parent	Yellow Round	x				Green Wrinkled	
Genotype	YyRr	↓				yyrr	
Gamets	(YR) (Yr) (yR) (yr)					(yr)	
BC_1F_1	YyRr	:	Yyrr	:	yyRr	:	yyrr
	1		1		1		1
	Yellow Round		Yellow Wrinkled		Green Round		Yellow Wrinkled

Table: Number of different kinds of gametes produced by F_1 and number of individuals in perfect F_2 population

Number of genes segregating	Number of different kinds of gametes produce by F_2	Number of individuals in the perfect F_2	Number of different homozygous genotype in F_2	Number of different phenotype in F_2
1	2	4	2	2
2	4	16	4	4
3	8	64	8	8
n	2^n	4^n	2^n	2^n

Reasons for over looking of mendels result

The important reasons for the neglect of Mendel's findings related to mechanism of inheritance for a long time are given below.

i. Mendel generalized his result based on his studies on garden pea. Later on he worked on hawkweed (*Hieraceum*) on the advice of C.V. Nagali. Mendel could not prove his result on this plant as the embryo is formed from the ovule without fertilization.

ii. Mendel explained his result with help of Mathematics. The scientist at that time did not appreciate this approach.

iii. Mendel could not support his findings through cytological studies

iv. After his failure to demonstrate the result on hawkweed, he did not give proper publicity to his work and kept quite.

DOMINANCE

The suppression of the expression of one trait of a Character by another trait of the same character is called Dominance.

Types of Dominance

I. Complete dominance

If the phenotypes of the heterozygote as well as homozygote dominant individuals are identical then the concerned dominant allele is said to have complete dominance.

eg. In garden pea, the homozygote (YY) and heterozygote (Yy) individuals produce only yellow colour seed. (Yy = YY)

II Incomplete dominance

Some alleles are neither dominant nor recessive. In this condition hybrids are intermediate in phenotype. This is called incomplete dominance. (Rr ≠ RR)

e.g. Flower colour in 4 0 clock.

a. Homozygous dominant genctype (RR) produce red colored flower.

b. Homozygous recessive genotype (rr) produce white colored flower.

c. Heterozygous genotype (Rr) produce pink colored flower.

III Co-dominance

Expression of phenotypic trait of both homozygotes in the heterozygote condition is called co-dominance. In co-dominance, both alleles of a gene have the full expression in heterozygous individuals.

(eg.) Coat colour in shorthorn cattle or blood group in human beings

In shorthorn cattle, a pair of gene controls red and white coat colour. Crosses between red (C^R C^R) and white (C^W C^W) cattle produce F_1 offspring of reddish gray or roan. Superficially this would seem to be a case of incomplete dominance, but close examination of roan animal reveals that the coat colour is composed of a mixture of red hairs and white hairs. The coat colour of the roan is not intermediate between red and white but due to the phenotypic expression of both homozygotes.

Genotypic and phenotypic ratios are identical in incomplete dominance and co-dominance. The difference lies in the operating ways of the gene.

IV Over dominance or Hetero dominance or Super dominance

The phenomenon of expression of phenotype in heterozygote in greater intensity than in the two concerned homozygotes is called over dominance.

The over dominance is not the property of an alleles and it is due to the heterozygous state (inter allelic interaction) of the concerned gene.

(eg.) Eye colour in fruit fly

Dominant allele WW,Ww- Red eye

Recessive allele ww- white eye

Eye pigments sepiapteridine and himmelblaus are present in row concentrations in 'ww' types. 'WW' have relatively higher concentrations of these pigments. However, flies heterozygous for this gene 'Ww' have appreciably higher concentrations of these two pigments than the two homozugotes.

Penetrance

The percentage of expression of a gene is called penetrance. When a gene is expressed in 100 percentages of cases, the gene is said to have **complete penetrance**. Sometimes environmental factors may suppress completely the expression of a gene is called in **complete penetrance**. This gene has a reduced penetrance.

For example, a dominant gene causes blue sclera in human eye. This dominant gene is not expressed in about 10 per cent if the peoples have normal white sclera, even though they carry the dominant gene. These people can transmit the gene to their children. The children may express this dominant gene. Thus this dominant gene for blue sclera has reduced penetrance of 90 per cent.

Expressivity

The degree of variation in the expression of a penetrant gene is called expressivity.

Types of expressivity

1. Uniform expressivity

When phenotypic expression of penetrant gene is similar/ identical in all individuals who carry those genes, then it is known as uniform expressivity. (eg.) most of qualitative characters

2. Variable expressivity

When phenotypic expression of penetrant gene is differing in all individuals who carry those genes, then it is known as variable expressivity.

(eg.) Dominance gene controls chlorophyll deficiency in Lima bean seedlings leaves. It shows variable expressivity i.e. chlorophyll deficiency is either

i. On tips of leaves OR
ii. On leaf margins of leaves OR
iii. On tips and leaf margins.

The reduced penetrance and variable expressivity may be due to modifying genes or due to external environmental factors.

Atavism

The reappearance of offspring which resemble with their remote ancestors is called as throwbacks or atavism or reversion.

It has been observed in certain cases that characters may often remain hidden generation after generation through the effect of inhibiting or epistatic factor or some other gene interaction. Sometime it happens that some wild character which was present in the ancestor reappear in the offspring.

Atavism result as account of chance combination of gene that allows a long suppressed character to reappear. (e.g.) homozygous gene 'cc' in albino rats does not permit the appearance of agouti pattern. But in an out cross, when gene 'c' is replaced by it allele 'C' agouti colour appears in the offspring.

Black × Albino

Ccaa ↓ ccAA

CcAa (agouti)

Phenocopy

An environmentally induced which resembles the effect of a gene mutation is called phenocopy. The term "phenocopy" was first proposed by Richard Goldschmidth. He subjected pupae of *Drosophilla* to high temperature (35^0c) for a short time at different periods in their development. Several phenotypes appeared which were similar to the phenotypes produced by certain mutant genes. Goldschmidth found that the genes had not been changed by the heat treatment, and the descendants of the phenocopies were normal in their phenotypes, when they were grown at normal temperature.

Rappoport (1939) found that when the larvae of the normal brown bodied fruit flies were reared on food with silver salts, the emerging adults had yellow body. They were genotypically brown but phenotypically yellow because of the changed environment. These phenocopies when their larvae are fed with food without silver salts produce only brown-bodied adults, as their genotype is that of brown body. Thus a phenocopy can last only for that generation in which the environment that induced the change is present.

Pleiotropism

The phenomenon of multiple phenotypic expression of a single gene is called pleiotropism. For example, the tomato mutant gene 'ls' suppresses the growth of

a) The axillary
b) The development of petals in flower.
c) It produces apocarpous pistil and dilatory anthers. According to Williams this gene suppresses the growth of meristematic tissue at the apex regardless of its position. For this reason, a single gene produces many fold effects.

In human the gene for disease *phenylketonuria* has pleiotropic effect and produces various abnormal phenotypic traits, collectively called syndrome. For example, the affected individuals have excess quantity of amino acid phenylalanine in their urine, cerebrospinal fluid and blood. They have short stature, mental retardation, widely spaced incisor, pigmented patches on skin, excessive sweating and non-pigmented hair and eye.

Modifying genes

A modifying gene is one that alters the expression of a major gene but has no effect on the allele of the major gene. The modifiers have similar but individually small effects and are usually present in large members that they cannot be individually identified.

In, Guemsey breed of cattle the solid colour (light yellowish brown) of the coat is due to dominant gene 'S' and the spotted coat (white spotting) is due to its recessive allele 's'. A number of modifying genes influence the intensity of spotting. If a large no. of modifying genes are present in animals with 'ss', the animals are highly spotted. If only a small number of modifying genes are present, they are medium spotted. If the modifying genes are absent, animals will have only few spot. These modifying genes have no effect in the presence of the gene for solid colour and animals with 'SS' or 'Ss' have solid coats irrespective of the number of modifying genes present.

Lethal genes

A lethal gene causes the death of all the individuals carrying it before these individuals reach the adulthood.

The lethal genes are classified into following types

i. Recessive lethal

The lethal effect is expressed in the individuals only when the alleles are in homozygous state. This condition is known as recessive lethal.

ii. Dominant lethal

The lethal gene whose lethal effects occur in heterozygous individuals is known as dominant lethal.

iii. Semi-lethal

Semi lethal genes do not lead to death of all the individuals that carry them in appropriate genotype. They cause death of more than 90% of the individuals.

iv. Sub-vital

Mutant genes reduce the viability of the individuals which carry them in appropriate genotype. Such gene kills less than 90% of the individuals in which it is present.

v. Vital genes

Those mutant genes which do not affect the survival of the individual in which they are present are known as vital genes.

vi. Super vital genes

Some mutant alleles enhance the survival of those individuals which carry them in appropriate genotype.

MULTIPLE ALLELES

Introduction

When more than two allelic forms of a gene occupy the same locus of the homologous alleles. In other words all the mutant form of a single gene constitutes a series of multiple alleles.

Characteristic features of multiple alleles

1. The members of a multiple allelic series occupy the locus of homologous chromosome.
2. Only two members of such alleles are present at a time in a diploid organism.
3. There is no crossing over in the multiple allelic series. If two alleles are involved in the cross the same two alleles are recovered in F2 or test cross progeny.
4. In a series of multiple alleles, wild type ins always dominant. Rest of the alleles in the series may exhibit dominance or intermediate phenotypic expressions.
5. The cross between two mutant alleles will always produce mutant phenotype (intermediate). Such cross will never produce wild phenotype. That is multiple alleles do not show complementation.
6. Multiple alleles always control the same character of an individual. However, the expression of the character will differ depending on the allele present.

Examples for multiple alleles

Several cases of multiple alleles are known both in animals and animals and plants. Some of the examples are given below:

Coat color in rabbits

A classical example of multiple allele is found in coat color of rabbits. The coat color of the rabbits. The coat color of the rabbit is controlled by four allelic form of a gene. The allele C produces full color or wild type ; c^{ch} produces white coat color rabbit with black tips on the ear, nose feet and tail called himalayan ; c produce no pigment resulting albino rabbits. inheritance studies revealed that alleles for coat color show a gradation of dominance in the order of $C > c^{ch} > c^{ch} > c$ (table 6.1.).

Table 6.1. Inheritance for colour in different crosses of rabbits.

S. no.	Parents	F_1	F_2
1.	Coloured X albino	Coloured	3 Coloured : 1 Albino
2.	Coloured X Chinchilla	Coloured	3 Coloured : 1 Chinchilla
3.	Coloured X Himalayan	Coloured	3 Coloured : 1 Himalayan
4.	Chinchilla X Himalayan	Chinchilla	3 Chinchilla : 1 Himalayan
5.	Chinchilla X albino	Chinchilla	3 Chinchilla : 1 Albino
6.	Himalayan X Albino	Himalayan	3 Himalayan : 1 albino

All these experiments clearly indicate that

1. That coat colour of the rabbit is controlled by a series of multiple alleles via., C, c^{ch}, c^{ch} and c.
2. The allele C is dominant over all other alleles.
3. The allele c is recessive to all other alleles.
4. The allele c^h is receissive to C but dominant over c^h and c
5. The allele c^h is recessive to C and c^{ch} but dominant over c.
6. Thus the alleles C, c^{ch} , c^{ch} , c forms a series of multiple alleles.

Phenotypes	Genotypes
Coloured	CC, Cc^{ch}, Cc^h , Cc
Chinchilla	c^{ch} c^{ch}, $C^{ch}C^{ch}$
Himalayan	c^h c^h, c^h c
Albino	(cc)

Blood groups in buman beings

One of the most firmly established series of multiple alleles in human involve the genetic iocus controlling the blood types A, B, AB and O

In 1900, Landsteiner discovered blood groups A,B, ABAnd 0 in human beings. He found that agglutionation may occur during transfusion of blood from one person to another this agglutionation occurs due to 'antigen- antibody reaction'.

Antigen is a specific protein found on the surface of the RBCs. Antibody is another kind of specific protein found in the plasma. There are two kinds of antigens viz., antigen A

and antigen B and two kinds of antibodies viz., A antibody' and 'B antibody'. The agglutination which is antigen-antibody reaction' is a highly specific one. 'A antigen' can react with 'A antibody' alone and 'B antigen' can react with 'B antibody alone. When there is 'A antigen' and 'B antibody', there will be no reaction and agglutination will not occur. Among human beings, the blood group is classified on the basis of the antigen present.

Persons with A antigen belong to A group. They have 'A antigen' on their RBCs and 'B antibody' in their plasma. Persons with B antigen belong to B group. They have 'B antigen' on their RBCs and 'A antibody' in their plasma.

Persons with both a and b antigens belong to AB group. They have A and B antigens on their RBCs are without A and B antigens. But they both A and B antibodies in their plasma (Table 6.2)

Table 6.2 Blood groups in human beings

Antigen	Antibody	Blood group
A antigen	B antibody	A group
B antigen	A antibody	B group
A and B antigen	No antibodies	AB group
No antigen	A and B antibodies	O group

Inheritance of blood group

Specific alleles control the production of specific antigens. Antibody is produced by immunological inechanism. The blood group is determined by a series of alleles viz., I^A, I^D and i

Blood groups and their possible genotypes

Blood group	Possible genotypes
A	$I^A I^A$, $I^A i$
B	$I^B I^B$, $I^B i$
AB	$I^A I^B$
O	ii

The I^A controls the production of 'A antigen'. The allele I^B controls the production of 'B antigen'. Both I^A and I^B are dominant over the recessive allele I I^A and I^B lack dominance over each other. The heterozygote $I^{A'}$ and I^B is not intermediate between the homozygotes I^A I^A and I^B I^B . It shows characteristics of both homozygotes. That is, both the alleles are expressed. This is called codominance (Table 6.3.).

Medical applications of blood group inheritance

It is necessary to match the donor and recipient before a blood transfusion is made. If A group blood is transfused into a B group man, the 'A antibody' of the recipient and agglutination fatal. In blood transfusion, the antigen of the donor and antibody of the recipient must be considered and matched.

Since O grup contains no antigen, it can be given to any blood group. Hence, it is called universal donor. AB group contains no antibody. So, it can receive any type of blood group. Hence, AB group is called universal recipient

Self – incompatability system in plants

Self – incompatibility

The inability of the pollen grains to fertilize the same flower or other flower of the same plant is known as self-incompatibility. Self-incompatibility system is controlled by a series of multiple alleles. In sporophytic self incompatibility system, the self incompatibility reactiojn of the pollen grain is determined by the genotype of the plant in which it is produced

$S_1 > S_2 > S_3 > S_4$

Genotypes of the plants	S_1 S_2	S_1 S_3	S_1 S_4	S_2 S_3	S_2 S_4	S_3 S_4
Genotype of the gametes	(S_1) (S_2)	(S_1) (S_3)	(S_1) (S_4)	(S_2) (S_3)	(S2) (S_4)	(S3) (S_4)
Incompatibility reaction of the pollen grain	All S_1	All S_1	All S_1	All S_1	All S_1	All S_1
Incompatibility reaction of the style	S_1	S_1	S_1	S_2	S_2	S_3
	Complete incompatibility (S_1 S_2 selfed)	Partial incompatibility (S_1 S_2 x S_1 S_4)		Complete compatibility (S_1 S_2x S_3 S_4)		

Pseudo alleles

Pseudo alleles refer to closely linked and functionally related genes. A cluster of pseudo alleles is known as pseudo allelic series or complex locus or a complex region.

Characteristics of pseudo alleles

1. Pseudo alleles govern different expressions of the same character.
2. Pseudo alleles occupy a complex locus, which is divided into sub loci.
3. They exhibit low frequency of genetic recombination by crossing over.
4. They exhibit cis-trans position effect

Iso alleles

An allele that is similar in its phehotypic expression to that of other independently occurring allale is known as isoallele. Isoalleles are two types

a. Mutant isoalleles : such alleles act within the phenotypic range of a mutant character.
b. Normal isoalleles : such alleles act within the phenotypic range of a wild character

GENE INTERACTION

Introduction

In Mendel's dihybrid cross each pair of allelic gene influence other character two or more pairs of genes may influences some times a single character. Depending upon the form of interaction the 9: 3: 3: 1 ratio is modified in various ways. The phenomenon of two or more genes affection the expression of each other in various ways in the development of a single character of an organism is shown as gene interaction.

Inheritance of comb pattern in fowls (without modification of 9: 3: 3: 1 ratio)

This was reported by W Bateson and R. C. Punnett. Domestic breeds of chickens have different comb shapes. Rose comb is found in Wyandotte breed, pea comb in Brahmas and single comb in leghorns. Each of these types breeds true. When Rose comb fowl is crossed with Pea comb, the F_1 Chicken shows a new comb type known as Walnut When the F_1 walnut combed birds are crossed together, four kinds of combs appear in the F_2 generation in the ratio of 9 comb nor walnut was expressed in the original parent lines. These two phenotypes were explained as the result of gene product interaction.

The F_2 ratio of 9: 3: 3: 1 is expected only in dihybrid cross. The number of Walnut in F_2 generation (9) indicates that they are double dominants. The number of single comb in F_2 generation (1) indicates that they are double recessives. The Walnut comb depends on the presence of two dominant genes R and P. R alone produces rose comb and P alone produces pea cob. The absence of both R and P produces single comb.

Parents	Rose	x	pea
	RRpp		rrPP
Gametes	(Rp)	↓	(rP)
F_1	RrPp (Walnut)		
F_2	RrPp	x	RrPp
	Walnut		Walnut
Gametes	(RP) (Rp) (rP) (rp)		

♀ \ ♂	RP	Rp	rP	rp
RP	RR PP Walnut	RR PP Walnut	Rr PP Walnut	Rr PP Walnut
Rp	RR Pp Walnut	RR pp Rose	Rr Pp Walnut	Rr pp Rose
rP	Rr PP Walnut	Rr Pp Walnut	rr pp pea	rr Pp pea
rp	Rr Pp Walnut	Rr pp Rose	rr pp pea	rr pp Single

Although the usual 9: 3: 3: 1 ratio was obtained, the result from this cross was unusual in three important respects.

i. The F_1 resembles neither parent new characters appear in the F_1 - Walnut.

ii. Two phenotypes (Walnut and single) not expressed in the original parents appeared in F_2

iii. The genes 'R' and 'P' were non-allelic and the comb pattern is influenced by two different genes.

Epistasis

Epistasis is a phenomenon in which the expression of one gene is masked or prevented by another non-allelic gene.The gene which prevent the expression of another gene is called epistatic gene,. the gene whose expression in masked is called hypostatic gene. Epistassis should not be confused with dominance. Epistasis is the interaction between different genes (non-alleles) where as dominance is the interaction between different alleles of the same gene.

Espistatic interactions (Modification of 9 : 3 : 3 : 1 ratio)

Where epistasis is operative between two gene loci, the number of phenotypes appearing in the offspring from dihybrid parents will be less than four. There are six types of epistatic ratios commonly recognized, three of which have 3 phenotypes and the other three having only 2 phenotypes.

Dominant epistasis (12 : 3 : 1) or Epistatic gene interaction

In dominant epistasis, a dominant allele at one locus can mask the expression of both alleles (dominant and recessive) at another locus, it is known as dominant epistasis. When the

dominant allele at one locus, for example A allele, produces a certain phenotype regardless of the allelic condition of the other locus, the 'A' locus is said to be epistatic to the B – locus. Further more, the dominant allele A is able to express itself in the presence of either B or b, then this epistasis is said to be dominant epistasis. Only when the genotype of the individual is homozygous recessive at the epistatic locus (aa) can the alleles of the hypostatic locus (B or b) be expressed. Thus the genotypes A-B- and A-bb produce two additional phenotypes. The classical 9: 3: 3: 1 ratio becomes modified into a 12: 3: 1 ratio.

In sorghum, the nature of the grain is either pearly or chalky. When a plant with pearly grains and another with chalky grains are crossed the F_1 is pearly, In the F_2 there is segregation of 3 pearly; 1 chalky, similarly, the colour of the grain either red or white, When a plant with red grains in segregation of 3 red: 1white. Red colour of the grain masks another character ie., the pearliness or chalkiness of grain When the colour of the grain is white, it is possible to say whether it is pearly or chalky but when the colour is red it is not possible to find out whether it is pearly or chalky

Recessive epistasis (9:3:4) or supplementary gene interaction

In recessive epistasis the recessive allele of one locus mask the expression of both dominant and recessive alleles at another locus, it is known as recessive epistasis.

Supplementary gene

Gene which by itself has no effect but qualitatively alter the effect of another gene. If the recessive genotype at one locus (eg aa) suppress the expression of alleles at the B-locus, the A – locus is said to exhibit recessive epistasis over the B locus. Only if the dominant allele is present at the 'A; locus can the alleles of the hypostatic B- locus be expressed. The genotypes A-B- and A-bb produce two additional phenotypes. The 9 : 3 : 3 : 1 ratio becomes 9 : 3 : 4 ratio.

(e.g.) Coat colour in mice is either agouti, black or a albino.

Agouti colour is commonly occurring one (wild type) and is characterized by colour banded hairs. The hair near the body is gray followed by yellow band and finally the distal part is either black or brown

The agouti and black coat colour in mice is controlled by 'A' and 'a' alleles respectively. Another non allelic dominant gene 'C' controls the production of an enzyme which converts a colorless precursor into melanin pigment and is required for the production

of any pigment. The homozygous recessive 'cc' lacks the enzyme, no melanin is produced and the animal is white coated.

3. Duplicate genes with cumulative effects (9 : 6 : 1) or Additive gene interaction

Two non-allelic genes have similar effect when they are separate, but produced enhanced effect when they come together. Such gene interaction is known as duplicate genes with cumulative effect.

If the dominant condition (either homozygous or heterozygous) at either locus (but not both) produces the same phenotype, the F_2 ratio becomes 9 : 6 : 1 For example, where the epistatic genes are involved in producing various amounts of substance such as pigment, the dominant genotypes of each locus may be considered to produce one unit of pigment independently. Thus genotypes A-bb and aa B produce one unit of pigment each and therefore have the same phenotype. The genotype aabb produces no pigment, but in the genotype A-B- the effect is cumulative and two units of pigments are produced. The 9: 3: 3: 1 ratio is modified into 9: 6: 1 ratio

In a cross between two light purple grains ie., P_1 and P_2 the F_1 was with dark purple grains. The F_2 segregated for 9 dark purple: 6 light purple: 1 white. Light purple of the grains is evidently due to the presence of a dominant gene P_1 or another dominant gene P_2. The two non-allelic dominant genes P_1 and P_2 possess an additive effect and the colour of the grain is dark purple when the genes P_1 and P_2 are present together, when both the dominant genes are absent, the colour of the grain is white.

4. Duplicate Dominant genes (15:1) or Duplicate gene interaction

Duplicate gene s are two pairs of alleles either alone or together produce the same effect. They are identical genes but are situated on two different pairs of chromosomes. Each gene is dominant to its allele but does not add to the effect of the other. Eg. Floating habit in rice.

When a non floating rice strain is crossed with a floating strain, the F_1 is non floating. The F_2 segregates for 15 non floating and 1 floating habit. The presence of a single dominant allele of any one of the two genes governing thetrait produces the dominant phenotype ie., non floating habit while recessive phenotype ie., floating habit is produced only when both the genes are in the homozygous recessive state. The 9: 3: 3: 1 ratio is modified in to a 15: 1 ratio

if the dominant alleles of both loci each produce the same phenotype without cumulative effect.

5. Duplicate Recessive genes (9 : 7) or Complementary gene interaction

Complementary genes

Non allelic genes that act together to produce a phenotype different from that produced by either alone.

In the case where identical phenotypes are produced by both homozygous recessive genotypes, the F_2 ratio becomes 9: 7 the genotypes aaB–, A-bb and aabb produce one phenotype. Both dominant alleles, when present together, complement each other and produce a different phenotype.

In sweet pea, the development of purple flowers require the presence of two dominant genes P_1 and P_2 or both the genes in homozygous recessive condition produce white flowers. Since both the dominant alleles P_1 and P_2 when present together, they complement each other and produce a new phenotype and hence called complementary genes.

6. Dominant and Recessive interaction (13 : 3) or Inhibitory gene interaction

In this type, a dominant allele at one locus can mask the expression of both alleles at second locus. Only two F_2 phenotypes result when a dominant genotype at one locus (eg. A-) and the recessive genotype at the other (bb) locus produce the same phenotypic effect. Thus A-B-, A bb and aabb produce one phenotype and aaB-produces another in the ratio of 13:3. (e.g.) node colour in sorghum.

In sorghum, when crosses were made between plants with purple node and green node, the F_1 was with purple node. The F_2 segregated for 3 purple: 1 green. In certain other crosses between plants with green node. Since purple is dominant over green, the F_1 is expected to be purple, but it is observed to be green. The gene for purple node is unable to express itself probably because of the presence of another gene. This gene is called inhibitory gene. It is capable of inhibiting the production of purple colour. Plants are purple, only if they possess the gene for purple colour, in the absence of the inhibitory gene. In the presence of the inhibitory gene, plants with the gene for purple. Are unable to exhibit the purple colour and are only green. Plants, which do not have the gene for purple colour, are also green whether they

have the inhibitory gene or not. The summary of all six epistatic ratios are given in the Table 7.1.

7.1. Summary of epistatic ratios

	A-B-	A-bb	aaB-	aabb
Classical ratio	9	3	3	1
Dominant epistasis / Epistatic gene interaction /mass	12		3	1
Recessive epistasis / Supplementary gene interaction	9	3	4	
Duplicate genes with cumulative effect / Additive gene interaction	9	6		1
Duplicate dominant genes duplicate gene interaction	15			1
Duplicate recessive genes / Complementary gene action.	9	7		
Dominant and recessive interaction / Inhibitory gene action	13		3	

MULTIPLE FACTOR INHERITANCE

Introduction

The inheritance of many different genes influencing the same phenotype in a cumulative fashion is called multiple factor inheritance.

Features of polygenic traits

The term polygene was introduced by Mather in 1941. This term has found wide usage in quantitative genetics replacing the older term multiple gene. Main features of polygenic character are briefly presented below:

1. Each polygenic character is controlled by several genes and has cumulative effect.
2. Polygenic characters exhibit continuous variation rather than a discontinuous variation hence; they cannot be classified into clear cut groups.
3. Effect of individual gene is not easily detectable in case of polygenic character and, therefore, such trails are also known as minor gene characters.
4. The statistical analysis of polygenic variation is based on means, variances and co-variances, whereas the discontinuous variation is analysed with the help of frequencies and ratios. Thus, polygenic characters are studied in quantitative genetics and oligogenic characters in Mendelian genetics.
5. Polygenic traits are highly sensitive to environmental changes, whereas oligogenic characters are little influenced by environmental variation.
6. Classification of polygenic characters into different clear cut groups is not possible because of continuous variation from one extreme to the grouping is possible because of discrete or discontinuous variation.
7. Generally, the expression of polygenic characters is governed by additive gene action, but now cases are known where polygenic characters are governed by dominance and epistatic gene action is primarily of non-additive type (dominance and epistasis).
8. In case of polygenic charactes, bio-metric measurements like size, weight, duration, strength, etc. are possible, whereas in case of oligogenic characters only the counting of plants with regards to various kinds like colour and shape is possible. Thus, metric measurement is not possible in case of oligogenic characters.

9. Transgressive segregants are possible from the crosses between two parents for a polygenic character. Such segregants are not possible in case of qualitative or oligogenic traits.
10. The transmission of polygenic characters is generally low because of high amount of environmental variation. On the other hand, oligogenic characters exhibit high transmission because there is little difference between the genotype and phenotype of such character. Thus, polygenic characters differ from oligogenic ones in several aspects.

Yule (1906) gave the theoretical explanation for the multiple factor hypothesis. According to him quantitative characters are controlled by many gene with cumulative effect without dominance and would produce continuous variation.

The experimental evidence for multiple factory hypothesis was provided by Nilsson & Ehle (1908) in studies on the inheritance of seed colour in wheat and oats. They obtained 3:1, 15:1 and 63:1 ratios between coloured and white seeds from different crosses and revealed that seed colour in wheat and oats is produced by one, two or three genes. The seed colour genes interact in duplicate manner, so that white colour seed is produced only when all the genes are present in the recessive state. Further the coloured seeds showed a varied intensity for colouring pattern and they obtained in the ratio of 1 dark red : 4 medium dark red : 6 medium red : 4 light red : 1 white. This suggested that the seed odour in wheat is controlled by genes which show lack of dominance and have small and cumulative effects (Table 8.1).

In order to explain the 1:4:6:4:1 ratio in kernel colour in wheat, Nilson – Ehle made the following assumptions.

i. In crosses showing 15:1 ratio in the F_2 seed colour is governed by two genes.
ii. One of the alleles of each colour gene produces seed colour and is called positive allele denoted by capital letter (eg) R_1, R_2 etc.
iii. These genes do not snow dominance and each of the genes (positive allele) has a small, equal effect on seed colour.
iv. The positive alleles of different coloured genes are additive in phenotypic effect

Inheritance of kernel colour in wheat

Parents	Dark red	x	White
	$R_1 R_1 R_2 R_2$	↓	$r_1r_1r_2r_2$
Gametes	$(R_1 R_2)$		(r_1r_2)

$R_1 r_1R_2 r_2$ (Selfing)

Medium Red

F_2 generation

♂ / ♀	$R_1 R_2$	$R_1 r_2$	r_1R_2	$r_1 r_2$
$R_1 R_2$	$R_1R_1R_2R_2$ DR	$R_1R_1R_2r_2$ MDR	$R_1r_1 R_2R_2$ MDR	$R_1r_1R_2r_1r_2$ MR
$R_1 r_2$	$R_1R_1R_2r_2$ MDR	$R_1R_1r_2r_2$ MR	$R_1r_1R_2r_2$ MR	$R_1r_1r_2r_2$ LR
$r_1 R_2$	$R_1r_1R_2R_2$ MDR	R_1r_1 MR	$r_1r_1R_2R_2$ MR	$r_1r_1R_2r_2$ LR
$r_1 r_2$	$R_1r_1R_2r_2$ MR	R_1r_1 LR	$r_1r_1R_2r_2$ LR	$r_1r_1r_2r_2$ White

Table 8.1 Genotype and phenotype frequencies produced by two genes with cumulative effect on seed colour in wheat

Genotype	Frequency	No. of positive allele	Phenotype	Frequency
$R_1R_1R_2R_2$	1	4	Dark Red	1
$R_1r_1R_2R_2$	2	3	Medium DarkRed	- - -
$R_1R_1R_2r_2$	2	3	Medium DarkRed	- - -
$R_1r_1 R_2r_2$	4	2	Medium Red	- - -
$R_1R_1r_2r_2$	1	2	Medium Red	- - -
$r_1r_1R_2R_2$	1	2	Medium Red	- - -
$R_1r_1 r_2r_2$	2	1	Light Red	- - -
$r_1r_1R_2r_2$	2	1	Light Red	- - -
$r_1r_1 r_2r_2$	1	0	White	1

Oligogenes

Genes having larger effect on the characters they govern and shows discontinuous variation are called oligogenes or major genes.

Polygenes

Genes individually having small cumulative effect but jointly responsible for continuous variation, specific to a metric trait are called polygenes or minor genes.

S. No	*Qualitative characters*	*Qualitative characters*
1.	Controlled by oligogenes (one or two genes)	Controlled by polygene (each with small cumulative effect)
2.	Shows discontinuous variation	Shows continuous variation
3.	Less influenced by envirinment	Much influenced by environment
4.	Concerned with individuals mating and their progeny	Concerned with population of organisms consisting of all possible kinds of mating.
5.	Exhibit high heritability	Exhibit low heritability.
6.	Analysis by making counts and ratios	Analysis by mean, variance and covariance

Transgressive segregation

The appearance in F_2 individuals falling out side the parental range in respect to some character is called transgressive segregation. Transgressive segregation results due to fixation of dominant and recessive genes in separate individuals. Such segregation occurs when the parents are intermediate to the extreme values of the segregating populations. The superior plants are produced by an accumulation of plus or favourable genes from both the parents as a consequence of recombination. The example for transgressive segregation is given below.

Parents AA BB cc dd ee x aa bb CC DD EE

↓

F_1 Aa Bb Cc Dd Ee

F_2 aa bb cc dd ee and AA BB CC DD EE are the transgressive segregants.

LINKAGE

Introduction

When two or more genes present on the same chromosome and they do not exhibit independent assortment, they are said to be linked and the phenomenon of transmission of linked genes is called linkage. Large deviations from a 1: 1: 1: 1 in the testcross progeny of dihybrid exprement is used as a first evidence for linkage. This effect of linkage was first reported in1906 by Bateson and punnet is sweet peas. Among sweet peas they crossed a variety having blue flower (B) and long pollen grains (L) with another variety having red flower (b) and round pollen grains (l). The F_1 blue long (Bb Ll) was crossed with the double recessive parent, red-round (bb ll).

According to Mendel four kinds are expected in the ratio of 1 blue long: 1 blue round : 1 Red long : 1 Red round. But in Bateson and Punnet's experiment the four kinds are produced in different ratio i.e.7 Blue long: 1 blue round: 1

Parents	Blue long	x	Red round
	BBLL	↓	bbll
F_1		Bb Ll (Blue long)	
	Bb Ll	x	bbll
	Blue long		Red round
Gamets	(BL) (Bl) (bL) (bl)		(bl)

Test cross progenies

Offspring	Expected ratio	Actual ratio
Blue long	1	7
Blue round	1	1
Red long	1	1
Red round	1	7

Bateson and Punnet suggested that the F_1 hybrid blue long (Bb Ll) produced the gametes (BL) and (bl) about seven times than the gametes (Bl) and (bl)

They carried out another experiment. They crossed a variety of blue round with another variety of red long. The F_1 hybrid was blue long. It was test crossed.

Parents	Blue round	x	Red long
	BB ll	↓	bb LL
F_1		Bb Ll (Blue long)	
	Bb Ll	x	bb ll
	Blue long		Red round
Gametes	(BL)(Bl)(bL) (bl)		(bl)

Test cross progeny	Expected ratio	Actual ratio
BbLl Blue long	1	1
Bbll Blue round	1	7
bbLl Red long	1	7
Bbll Red round	1	1

Bateson and Punnet suggested that the F_1 hybrid blue long (Bb Ll) Produced gametes (Bl) and (bL) about seven time than the gametes (BL) and (bl)

Bateson and Punnet put forward the following points

1. The alleles, which come form the same parents, tend to enter the same gametes. The alleles which come from different parents tend to enter different gametes.
2. When the genes are linked, greater than 50% of progeny with parental phenotypes and less than 50% of progeny with recombinant phenotypes will occur.

Though Bateson and Punnet (1906) explained the effect of linkage in sweet pea they did not interpret their results in terms of the behavior of genes located on the same chromosome and the occurrence of crossing over between homologous chromosomes during meiosis.

T.H. Morgan's Experiments in Drosophila

T.H. Morgan (1911) was first to relate linkage to the segregation of homologous chromosomes and the occurrence of crossing over between homologous chromosomes during meiosis in the fruit fly *Drosophila melanogaster*.

T.H. Morgan explained the effect of linkage by considering of result of two crosses involving pairs of alleles of two genes located on the second chromosome of *D. melanogaster*. Once gene affect the body colour [gray body (b$^+$) which is dominant over black body (b)].

The second gene affects phenotype of the wing [long wing (s_+) which is dominant over vestigial wing or short wing (s)]

In the first cross (cross I) he made crosses between true breeding long wing and grey bodied flies eith true breeding short wing and black bodied flies. This cross produced heterozygous F_1 flies with long wings and gray bodies. Among the progeny of this test cross, 82 per cent exhibited one or ther other (41 per cent each) of the parental combination of traits. The other 18 per cent of the progeny had new or recombinant combinations.

Next, consider a different cross (cross II) one between homozygous flies with long wing and black bodies and homozygous flies with short wing and gray bodies. Again in cross II, 82 per cent of the test cross progeny have parental phenotypes (phenotypes identical to one or the other of the original parents) and 18 per cent have new or recombinant phenotypes.

Although the F_1 flies have the same phenotype (long wing, gray bodies) in b oth crosses, the test cross progeny of the F_1 female flies contain very different frequencies of the four phenotypic classes in the two cases. For example, 41 per cent of the test cross progenies in cross I are wild type (have long wing and gray bodies); in cross II only 9 per cent are wild type. Clearly, this shows that the allelic forms of the two genes that are present together on the homologous chromosomes of the parent tend to remain together on the chromosome of the progeny.

In cross I, the F_1 flies carried the wild type froms (s^+ b^+) of the two genes on one homologue and the mutant forms (s^+ b^+) on the other homologue. The genotype of a heterozygote of this type is frequently written as s^+ b^+ / s b. This arrangement of mutant and wild type form of two genes in a heterozygote is called the **coupling state or cis – configurations**. The alternative arrangement, illustrated in cross II where each homologue contains one mutant gene and one wild type gene (s^+ b / s b^+) is called the repulsion **state or trans configuration.**

Morgan formulated his theory of linkage from this experiment. According to this theory

1. Genes located on the same chromosome are inherited together. They are said to be linked.
2. Genes are present in the chromosomes in a linear order.
3. The distance between th linked genes determines the strength of linkage. Closely located genes show strong linkage and widely located genes show weeak linkage.

4. Only those genes located in different chromosome show independent assortment But now we came to know that genes that are located far apart on the same chrosome would also assort independently.

Type's of linkage

Linkage is classified on the basis of following three criteria.

I. Based on crossing over

(i) Complete linkage

Linkage in which crossing over does not occur is known as Complete linkage or absolute linkage. In complete linkage, test crtoss progenies posses only parental types.

(ii) Incomplete linkage

If some frequency of crossing over occurs between linked genes, it is known as incomplete linkage. In incomplete linkage, the test cross yields some recombinants besides parental combinations.

II. Based on genes involved

(ii) Coupling linkage

It refers to linkage either between dominant genes or between recessive genes.

III. Based on chromosomes involved

(i)Autosomal linkage

It refers to linkage of such genes, which are located in other than sex chromosomes.

(ii)X-chromosomal linkage

It refers to the linkage of genes. Which are located in sex chromosomes.

Crossing over

Crossing over may be defined as "interchange of chromosomal segments between non-sister chromatids of a homologous chromosome pair". The tern crossing over was first used by Morgan and Cattell in 1912 (Fig. 9.1.).

Fig. 9.1. Chiasma and crossing over

The main features of crossing over are given below.

i. Crossing over takes place at four strand stage during pachytene stage Pachytene stage of Meiosis I.

ii. Crossing over occurs between non-sister chromatids of the homologous chromosomes.

iii. Crossing over produces new combination of genes between linked genes.

iv. The value of crossing over or recombination may vary from 0 – 50 %

v. The frequency of recombinants can be worked out from the test cross progeny. It is expected as the percentage ratio of recombinants to the total population (parental types + recombinants). Thus,

$$\text{Crossing over frequency (\%)} = \frac{\text{No. of recombinants}}{\text{Total progeny}} \times 100$$

Cytological basis of crossing over

Morgan first proposed crossing over to explain. The formation of recombinant combination of genes that were linked by gametic data. F. Janssens first correlated the chiasma frequencies with recombination frequencies, and showed that a direct relationship between crossing over and chaismata.

Direct cytological evidence for crossing over was first given by Curt Stern (1931) in *Drosophila* and in maize by H.B. Creighton and B. Mc Clintock.

Curt Stern Experiment

Curt stern found aberrant X chromosomes in a variety of *Drosophila* and on was an X chromosome to which a portion of Y chromosome was attached. This is inverted 'L' shaped. The other was a broken 'X' chromosome. Under microscope these two kinds of chromosomes can be easily distinguished from each other as well as from the normal 'X' chromosome.

He produced female flies heterozygous for these two morphologically distinguishable X-Chromosomes. These that is located on the X-chromosome. One gene affect the eye shape, the partially dominant mutant allele B results in bar - shaped eye and its wild type allele b^+ produces round eye in homozygous condition. The second gene affect the eye colour, the mutant allele care results in carnation coloured eye and its dominant wild-type allele care

produce red eyes. The females used in stern's study carried the allelic pairs in the cis - configuration as shown below.

Stern crossed such heterozygous females (car B / car^{+} B $^{+}$) with males having carnation coloured, normal shaped eye (car b^{+} / Y males) and studied the offspring of the next generation. The cross and results obtained by him are diagramed in Fig 9.2

In the absence of crossing over only two kinds of eggs were expected, one with broken X chromosome carrying the genes car and B and other with attached 'X' chromosome carrying the genes car^{+} and B^{+} If crossing over occurred between two aberrant X chromosomes, two more types of eggs would be expected. That is one recombinant type with normal X chromosome carrying the genes car and B andother recombinant type with broken as well as attached 'X' chromosome carrying the genes car and b^{+}. Thus the heterozygous females (car B / car B) have produced all four kinds of eggs in which, the X chromosomes were expected to be different not only genetically but also structurally.

After fertization, these four kinds of eggs produced the four types of phenotypes in F1 offspring in both sexes.

i. Caration barred eye
ii. Red colour and round eye (wild type)
iii. Carnation round eye
iv. Red bar shaped eye

These four kinds of offsprings were expected to have different X chromosomes. The shape of 'X' chromosome observed in the progeny agreed with their observation. That is, if crossing over involved the breakage and exchange of parts of homologous chromosomes then the recombinant male progeny with bar shaped red eyes (Car $^{+}$ B / Y) were found to carry the short x chromosome but now translocated piece of the Y chromosome attached X chromosomes at one end. Thus curt Stern's experiment was unique demonstration of the hypothesis that the genetic crossing over is accompanied by physical exchange between the homologous chromosomes.

Factors controlling crossing over

i. High and low temperatures increase the frequency of crossing over.
ii. X-rays and other irradiations increase the crossing over frequency.

iii. The age of the individual also affects the crossing over frequency. It was found that crossing over frequency is higher in older ages.

iv. Gene mutations affected the frequency Some mutations are known to decrease the frequency.

v. Crossing over at one point of the chromosome tends to prevent other crossing overs in nearby places. This phenomenon is called *interference.*

vi. Crossing over does not take place in Drosophila male; and silk worm females. Thus sex also affects the crossing over.

vii. Crossing over is less frequent near centromeres and the tips of chromosomes.

viii. Inversions of chromosome segments suppress the crossin over.

Significance of Crossing Over

i. Crossing over provides a direct evidence for the linear arrangement of genes in the chromosome.

ii. Since crossing over results in recombination of genes variations are produced. Variations are the *raw material for evolution.*

iii. Crossing over helps in the construction of chromosome maps.

Crossing over forms the basis for chromosome mapping Crossing over between particular linked genes occurs at *constant frequencies.* The percentage of crossing over is directly proportional to the distance between the two genes. Thus, the percentage of crossing over between any two genes indicates the relative distance between them. Percentages of crossing overs between various genes of a chromosome can be calculated experimentally. From this data the relative distances between the various genes can be worked out. From the relative distances of various genes, their exact locations in the chromosome can be determined. *Sturtevant*, a student of T.H. Morgan formulated this idea of chromosome mapping. Construction of a linkage or genetic map or chromosome mapping

Construction of genetic map

The method of construction of genetic maps of different chromosomes is called genetic mapping. The genetic mapping includes the following:

Determination of linkage groups

Before starting the genetic mapping of the chromosomes of a species, one has to know the exact number of chromosomes of that species and then, he has to determine the total number of genes of that species by undergoing hybridization experiments in between wild and mutant strains.

Determination of map distance

After knowing total number of genes in each linkage group of a species, the relative distance between each linked gene have to be determined. The distance between two given genes is calculated according to the percentage of crossing over, because, cross over frequency is directly proportional to distance between the genes. For example, if the percentage of crossing over between two linked genes is 1 per cent means, the map distance between two linked genes is one unit of map distance, which is known as centimorgan. If the mean number of chiasmata is known for a chromosome pair, the total length of the map for that linkage group may be predicted:

Total length = Mean number of chiasmata X 50

Two point test cross

The percentage of crossing over between two linked genes is calculated by test crosses in which a F_1 dihybrid is crossed with a double recessive parent. In such crosses, crossing overs occur at two points, hence it is called two point test cross. For example, a dihybrid having the genotype AC/ac is test crossed with a double recessive parent ac/ac and test cross produces 74% parental combinations and 26% cross over types. Then, the distance between the loci A and C is estimated to be 26 **centimorgans** If the distance between the two loci is large in two point test cross, double cross over types are undetected and it would appear as parental types. Hence, we underestimate the true map distance (crossover percentage).

Three point test cross

Double crossovers usually do not occur between genes less than 5 map units apart. For genes further apart, it is advisable to use a third marker between the other two in order to detect any double crossover. A thee point test cross or trihybrid test cross gives us information regarding relative distance between these genes, and also shows us the linear order in which these genes should be present on chromosome.

Determination of gene order

After determining the relative distances between the genes of a linkage group, it becomes easy to place genes in their proper linear order. For example, if the liner order of three genes ABC is to be determined, then these three genes may be in any one of three different orders depending upon that which gene in the middle. If we suppose that the distance between the gene A-B=, B-C=7, A-C=5, we can determine the order of genes correctly in the following manner. Let us assume that gene A is in the middle, the distances between B-C are not equitable, gene A cannot be in the middle.

In case II, let us assume that gene B is in the middle (A-B-C), the distances between A-C is not equitable; therefore, gene B cannot be in the middle. In case III, let us assume that gene C is in the middle (A-C-B), the distance between A-B are equitable, therefore, gene C must be in the middle.

Combining map segments

The different segments of map of a complete chromosome are combined to form a complete genetic map of a long chromosome.

Interference and coincidence

In higher organisms it has been found that one chiasma formation reduces the probability of another chiasma formation in an immediately adjacent region of the chuomosome, probably because of physical inability of the chromatids to bend back upon themselves within certain minimum distances. The tendency of the crossover to interfere with the other crossover is called inferference. The net result of this interference. is the observation of fewer double crossover types than would be expected according to map distances. The strength of interference varies in different segments of the chromosome and is usually

expressed in terms of a coefficient of coincidence, the ratio between the observed and the expected double crossovers.

$$\text{Coefficient of coincidence} = \frac{\text{\% of observed double crossovers}}{\text{\% of expected double crossovers}}$$

The coincidence is the complement of interference, so Coincidence + Interference = 1.0

When the interference is complete (1.0), no double crossovers will be observed and coincidence becomes zero. When we observe all the double crossovers expected, coincidence is unity and interference becomes zero.

Determination of map distance using three point test cross

In a three point test cross, eight different phenotypic classes are obtained. These eight classes are identified in two different ways, *viz.*, (1) by phenotypic frequencies, and (2) by alteration of gene sequence in the genotype as a result of single crossing over or double crossing over between three linked genes. Parental types have the maximum phenotypic frequencies, and the single crossovers have phenotypic frequencies between these two classes. Suppose, ABC / abc are three linked genes located on two different chromosomes in F_1 of a cross between AABBCC and aabbcc parents.

1. Single crossover between A and B will alter the position of two genes. *Viz.*, B and C
2. Single crossover between B and C will alter the position of only one gene, i.e. C
3. Double crossover between A and C will alter the position of only middle gene, i.e. B

Thus eight types of gametes are produced by F_1 and only one type of gamete is produced by homozygous recessive parent. Union of male and female gametes will produce eight different phenotypic classes (Table 9.1).

Calculation

The recombination percentage or unit distance between genes is worked out by calculating the crossing over percentage between different genes. Suppose number of crossover progeny between genes A and B is P, between genes B and C is Q, between A and C is R, and total progeny is T, Then,

Table 9.1. Summary of the results obtained from a three point test cross between ABC/ abc x abc /abc

Genotypic Classes	Phenotypic classes	Assumed frequencies	Remarks
ABC/abc	ABC	349	Parental types
abc/abc	abc	360	
Abc/abc	Abc /	114	Single crossover between A and B
aBC/abc	aBC	116	
ABc/abc	ABc	128	Single crossover between B and C
abC/abc	abC	124	
AbC/abc	AbC /	5	Double crossover between A and C
aBc/abc	aBc	4	
Total		1200	

Recombination (%)

1. Between genes A and B $= \dfrac{P + R}{T} \times 100$

$$= \frac{230 + 9}{1200} \times 100 = 19.92$$

P = 114 + 116 = 230

R = 5+4 = 9

2. Between genes B and C $= \dfrac{Q + R}{T} \times 100$

$$= \frac{252+9}{1200} \times 100 = 21.75$$

Q = 128 + 124 = 252

R = 5 + 4 = 9

$$3.\ \text{Between genes A and C} = \frac{P+Q}{T} \times 100 = 40.30$$

Gene Sequence

The gene sequence is determined with the help of crossing over percentage between two genes. Greater the recombination percentage between tow genes, more is the distance between them and vice versa. In this case, the maximum crossing over % is between gene A and C (40.3 %). This indicates that B is located between A and C as given below:

A ..... B C

19.72 21.75

41.67

Coefficient of Coincidence

It is calculated with the help of following formula:

$$\text{Coefficient of coincidence} = \frac{\text{Observed double crossovers}}{\text{Expected double crossovers}} \times 100$$

$$\text{Observed double cross overs} = \frac{9}{100} \times 100$$

Expected double cross overs = Product of two single recombination values

= 19.92 x 21.75 / 100 = 4.33 %

$$\text{Coefficient of coincidence} = \frac{0.75}{4.33} \times 100 = 17.32\ \%$$

Coefficient of interference = 1-0.1732 = 0.1732 = 0.8268 or 82.68 %

SEX DETERMINATION

Introduction

Nature contain a vast array of diverse mechanisms of sex determination. In lower organisms the two sexes are phenotypically indistinguishable exept for the reproductive organs and in some lower eukaryotes the two genetically distinct type of gametes are some time morphologically indistinguishable and called as isogamy (iso – means 'same') gametes eg. Green algae *Chlamydomonas reinhardtii.*

In higher form, there are many distinct morphological differences between male and female sexes. This phenomenon is called sexual *dimorphism.* Basically the phenomenon is called sexual dimorphism Basically the reproductive organs and sex cells are different between males and females. This forms primary sexual character. The male and female sexes differ from each other in many somatic characters. For example mammary glads in females and beard in males are secondary sexual characters.

Two kinds of chromosomes

In dioecious organisms, chromosomes are tow kinds. They are autosomes and allosomes.

Autosomes

Chromosomes containing gene, which determine the various somatic characters.

Allosomes

H. Henking (1891) first identified the chromosomes involved in sex-determination. Allosomes are otherwise called as sex-chromosomes. These are the chromosomes responsible for the determination of sex. The allosomes are of two types *viz.*, X and Y.

Modern geneticists have reported many different mechanisms of determination of sex in living organisms

Some important and common mechanisms of sex determination are following:

Homogametic and Heterogametic sexses

The individuals carrying the same type of sex chromosomes namely XX are called homogametic. They give only one kind of gametes (X). The individuals having dissimilar sex chromosomes namely XY are called heterogametic. They give two kinds of gametes (X) and

(Y). Among human being and *Drosophila* the female is homogametic. In birds, moths and butter flies, the females are heterogametic and males are homogametic.

XX-XY type of sex determination

In insect like Drosophila and in human beings, the male have dissimilar sex chromosomes – XY chromosomes. In female, two similar sex chromosomes are present – XX chromosomes The female produces only on kind of egg(22 autosomes + one X chromosome) and hence homogametic The male produces two kind of sperms one with 22 autosome = one X chrosome and other with 22 autosome = one Y chromosome and hence heterogametic. The egg (X) fertilized by (X) sperm produces female offspring XX. The egg (X) fertilized by (Y) sperm produces male offspring XY.

In many species including most birds, moths and some fish the sex determination is identical to that of XX – XY mechanism but female is heterogametic (usually designed as ZW) and males being homogametic (usually designated as ZZ) This mechanism of sex determination is sometimes called as ZZ – ZW/ However, mechanically this system is identical to the XX – XY system but with the relationship between sex – chromosomes and sex phenotypes reversed. Stated differently in birds, the chromosome composition of the egg determines the sex of the offspring where as in humans and fruit flies, the chromosome composition of the sperm determines the sex of the offspring.

The 'Y' chromosome and sex determination in mammals

In both *Drosophila* and humans, a normal female has XX sex chromosome composition and normal males have XY sex chromosome composition. Thus it might be tempting to the X chromosome and genes for maleness on Y chromosome. In mammals the presence of 'Y' chromosome is required for the development of male sex phenotype.

In contrast recent evidence shows that Y chromosome plays no significant role is sex determination I Drosophila. In mammals surprisingly X chromosomes present in any number (eg. XXX) in the absence of Y chromosome give rise maleness. The gene on the Y chromosome in human that is responsible for the development of testis is called TDF (for Testis Determining Factor) the TDF gene exhibits very dominant effect on the development of the sex phenotype.

XX-XO type of sex determination

In some of the insects like grasshoppers, all the eggs carry an X chromosome, but it was included in only half of the cells forming sperm. All the sperm however had the usual complement of other chromosomes (autosomes). Eggs fertilized by sperm containing the X chromosome produced zygote with two X chromosomes which become female. Eggs receiving sperm without an X chromosome produced zygotes with one'X' which become males (Fig. 10.1). Males are referred as hemizygous for the X chromosomes or for genes located on the X chromosome.

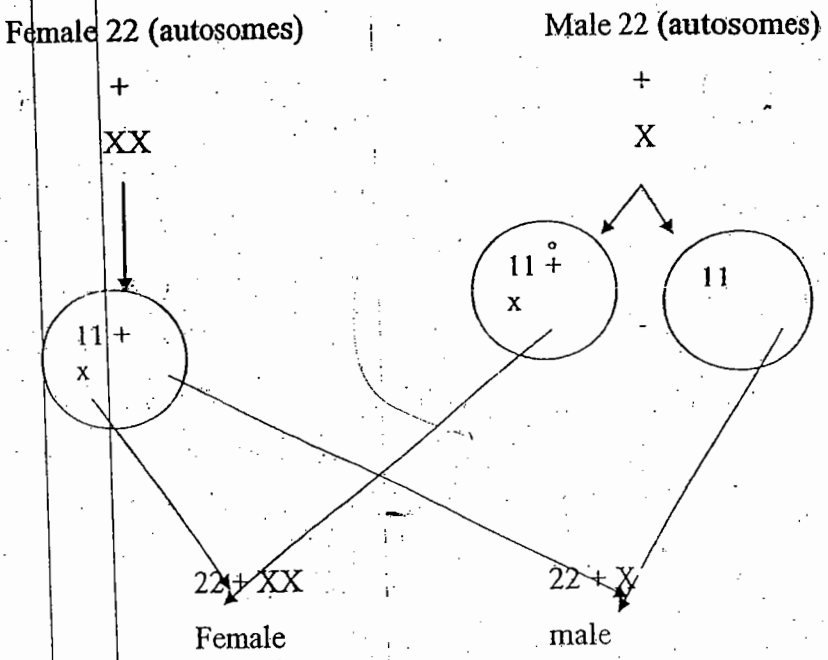


Fig 10.1 XX-X0 type of sex determination

Bridges genetic balance theory in *Drosophila*

Soon after the identification of X chromosome, the sex determination id *Drosophila* was more complicated than the preliminary observation. C B. Bridges shows that female determining genes ware located on the X chromosome and male determining genes were on the autosomes of *Drosophila.* The genetic balance theory of sex determination was devised to explain the mechanics of sex determination in *D. melanogaster.*

Bridges experimentally produced various combinations of X chrosomes and autosomes in *Drosophila.* A triploid female was crossed with a diptoid male. The triploid female produces four types of eggs.

(i) X 2n (A) (ii) XX 2n (A) (iii) X n (A) (iv) XX n (A)

The diploid male produces two types of sperms

(i) Xn (A) (ii) Yn (A)

When the four types of eggs are fertilized by the two types of sperm at random eight kinds of offsprings are produced (table 10.1)

Table 10.1. Sex expression in Drosophila in relation to X/A rules

	Xn (A)	Yn (A)
X2n (A)	XX3n (A) = 2X/3 = 0.66 Inter sex	XY3n (A) = 1X3/3 = 0.33 Super male
Xn (A)	XX2n (A) = 2X/2 = 1.00 Female	XY2n (A) = 1X /2 + 0.50 Male
XX2n (A)	XXX3n (A) = 3X/3 = 1.00 Triploid female	XXY3n (A) = 2X/3 = 0.66 Inter sex
XXn (A)	XXX2n (A) = 3X / 21.50 Super female	XXY2n (A) = 2X/2 = 1.00

Eight kinds of offspring are produced as follows

1. Triploid female with three X chromosomes and three sets of autosomes.
2. Normal diploid female with two chromosomes and two sets of autosomes.
3. Diploid XXY female with two X chromosomes and on Y chromosome and two sets of autosomes.
4. Interesexes with tow X chromosomes and three sets of autosomes.
5. Intersexes with two X chromosomes and one Y chromosome and three sets of autosomes.
6. Normal males with one X and one Y chromosome and two sets of autosomes.
7. Super females with three X chromosomes and to sets of autosomes.
8. Super males with one X and one Y chromosome and three sets of autosomes.

First, it was supposed that the XX individual is female and XY male. After the finding of non-disjunction, this early formulation was altered slightly that the XX is female and *X* male. The importance of Y chromosome in the sex determination was removed.

In Bridge's experiment, there is an individual with two X chromosomes. Yet it is not female. It is shifted out of the female class by the addition of one set of autosomes and it

becomes an intersex. So autosomes also play a positive role in the determination of sex. The intersexes lead to the conclusion that in Drosophila, sex is determined by the X chromosomes as well as by the autosomes.

The intersex differs from female by the assumption of certain male characters. This occur due to "the internal prepondence of male tendency genes" present in the autosomes which are added as an additional set.

Every individual has both male and female potentialities, because X chromosomes have female tendency genes and the autosomes have male tendency genes. The sex is decided by the balance that is, by preponderance of either male tendency genes or the female tendency genes. The deciding factor is the ratio between the number of X chromosomes and number of the sets of autosomes in the zygote. This is called **'sex index'** by Bridges.

$$\text{Sex index} = \frac{\text{Number of X chromosomes}}{\text{Number of sets of autosomes}}$$

If the ratio is 1.0, the individual will be female and if it is 0.5 male will result. The ratio between 0.5 and 1.0 result in intersex. The ratio 1.5 leads to super female and 0.33 leads to super males.

Haplo – diploid sex determination

In several species o Hymenoptera such as honey bees, ants, wasp and saw flies' males develop parthenogenetically (from unfertilized eggs) and have a haploid chromosome number (16 in the drone / male honey bees). The queen hone bee and the workers which arise from fertilized eggs carry the diploid chromosome number (32) So, in the honey bees the sex in determined by the haploid and diploid chromosome numbers. It is some times said that a drone honey bee has no father but has grand father. This is possible by the haploid diploidy mechanism of sex determinalion

Similarly in the parasitoid wasp Bracon hebetor (formerly Habrobracon) the females are diploid with 20 chromosomes and males are haploids with 10 chromosomes. Female originate from fertilized eggs and male from unfertilized eggs. This mechanism of sex determination is ofter referred to as hapio-diploidy.

Results of the experiments by whiting showed that the sex determination depends upon the genetic compositon of the certain region of the chromosome ie, homozygous, heterozygous

or hemizygous status of certain chromosome segments and not on diploidy versus haploidy per se. If Xa, Xb, Xc are different chromosomal segments, then female sex is produced by heterozygous of the certain chromosomal segments (Xa xb / xa Xc) and male phenotype is due to hemizygous or homozygous condition of chromosomal segments (Xa, Xb, Xc / Xaxa, Xuxb, Xcxc)

Role of environment and sex determination

In some lower animals, sex determination is non-genetic and depends on factor in the external environments. Males and females have similar genotypes, but stimuli from environments. Males and females have similar genotypes, but stimuli from environmental sources initiate development towards on sex or the other.

In the case of Bonellia, for example, females are free living form with an ovoid body and long proboscis. The males are small, parasitic and live in the reproductive tracts of the larger female. Larvae of Bonellia are potentially. Capable of developing either into males or females. If the larvae are isolated, the will become females.

If they are grown near the females, they will become males. Sex determination is non-genetic and depends on the external environments. The hormone like substances secreted by the female has an effect to turn the larvae into males. So the presence or absence of this hormone like substance in the environment determines the sex in Bonellia.

Gynandromorphism

Gynandromorph is an individual in which one half is male and other half is female. The mosaic condition of sex chromosomes leads to phenotypic sex mosaic. The Gynandromorphism is best studied in *Drosophila*, where there is no dilutions of the characteristics i.e the male side is fully male and the female side is fully female. There are three kinds of gynandromorphis.

(i) **Bilateral gynandromorphs**

It is found in *Drosophila* One lateral side of the fly is male and other lateral side is female. This is due to abnormal mitosis during early cleavage of the zygote.

(ii) **Auterior – posterior gynandromorphs**

Some gynanders possess male characters on the anterior side and female character on the posterior side of the body or vice-versa. These are called anterior – posterior gynanders.

(iii) Sex – piebalds

In some gynanders, the individual is predominantly a male or female with patches of opposite sex scattered on it. They are known as sex-pie balds.

Sex – mosaics

Mosaicism refers to a condition in which a person's cell consists of two or more populations, each with different chromosome complements. Murry Barr found a girl in whom both buccal and vaginal smears showed two barr bodies, thus indicating the XXX chromosome complements. Blood cells showed / no barr body indicating a XO chromosome complements. These mosaics arise as results of errors in mitosis in early stages of embryonic development

Sex determination in plants

Sex-determination in *Melandrium album*

In M. *album* which follows the XY mode of sex determination (Fig. 10.3). The Y chromosome of M. *album* has three district regions influencing sex-determination and male fertility has been localized on the differential part of the Y chromosome (Which does not have a homologous part on the X) The region I suppress the femaleness. In the absence of this region, plants are bisexual. Region II promotes male development. When this region (With or without Region I) is missing a female plant is produced Region III carries male fertility genes, Loss of this region result in male sterility.

Sex determination in papaya

A single gene controls sex determination in papaya. In this plant single gene with three alleles (m, m_1, m2) control the sex-differentiation (Table 10.2). Female plants are homozygous with a genotype of mm, while male plants are heterozygous with a genotype of m1 m; the heterozygote condition of m_2 m produce hermaphrodite plants. The alleles M_1 M2 are recessive lethal. The mating between female m(m) and male (m_1 m) produce 50% female (mm) plants.

Table 10.2 postulated sex determination in papaya

Genotype	Survival	Sex – expression
Mm	Vital	Female
M_1m	Vital	Male
M_2m	Vital	Hermaphrodite
$M_1M_1M_2M_2$	Lethal	

Sex-determination in maize

Maize plants are generally monoecious, i.e. male and female flowers are produced on the same plant. A single recessive gene, ba in homozygous condition (*bata*) interferes in the cob development and making these plants functionally male (Table 10.3.). Another recessive gene, *ts* converts the male flowers in tassels of ts ts plants into female flowers and plants to not produce pollen grains. In plants homozygous for both *ba* and *ts* are functionally female. So the two recessive genes (*ba* and *ts*) have converted a naturally monoecious plant into a dioecious one.

Table 10.3 Sex expression in maize

Genotype	Femaleflower	Male flower	Sex expression
BaBa TsTs	Normal	Normal	Monoecisous
Baba TsTs	Rudimentary	Normal	Male
BaBa tsts	Normal	Develop into Female flower	Female
Babatsts	Rudimentary	Develo into Female flower	Female

SEX LINKED INHERITANCE

Introduction

Inheritance through X chromosome is called sex linked inheritance. It was discovered by T.H. Morgan in 1910. Eye color and bar eye in Drosophila, color blindness and hemophilia in human and barred plumage in fowls are inherited through X chromosome.

Characteristic features of sex linked genes

1. Sex linked genes are located on 'x' chromosome only.
2. In diploid, homogametic sex contains two copies of sex linked alleles where as heterogametic sex contains only one sex linked allele.
3. A recessive gene, in a homogametic sex can express only when it is homozygous state, whereas in heterogametic sex a recessive allele express in hemizygous condition.
4. Sex linked genes follow the criss cross inheritance
5. Sex linked gene exhibit several deviation from the normal segregation pattern.

Criss-cross inheritance of X-chromosome in *Drosophila*

Female is produced when an X egg is fertilized by X sperm. Male is produced when Y sperm fertilizes an X egg.

	Female	Male
P	XX	XY
Gametes	(X)	(X) (Y)
Progeny	XX	XY

A male receives an X-chromosome only from the mother and never from father. The male receives Y chromosome only from this father, never from his mother. Thus the inheritance of X chromosome in *Drosophila* follows specific pattern. The male transmits his X chromosome to grandson only through his daughter. This is called criss-cross inheritance. The female transmits the X-chromosome both to her son and daughter.

The criss-cross pattern of inheritance is characteristics of sex-linked genes. This distinctive criss-cross pattern, from father through daughter to grandson replacing the usual pattern for the f1 and 12 segregation is now interpreted as evidence of sex-linkage.

Cross-cross inheritance of eye color in Drosophila.

A cross was made between white-eye male Drosophila and red-eye female *Drosophila* by T.H. Morgan in 1910 (Fig. 11.1). The F_1 flies were red eyed. When F_1 flies were intercrossed, three fourth of the F_2 flies possessed red eyes and one fourth white eyes. From this familiar 3:1 ratio, it is clear that this is a monohybrid inheritance where red is dominant over white. But, when the F_2 flies were classified for both eye color and sex.

It was found that

(i) All the F_2 females were red eyed.

(ii) Half of the F_2 males were red eyed.

(iii) Half of the F_2 males were white eyed.

When reciprocal cross was made between white eyed female and red eyed male, the F_1 was composed of two different phenotypes ie red eyed females and white eyed males. When the F_1 flies were intercrossed, the F_2 consisted

Of flies in the ratio-2 red eyed: 2 white eyed. When these F_2 flies were classified for both eye color and sex, it was found that

(i) Of two red-eycd flies, one is male and another is female.

(ii) Of two white eyed flies, one is male and another is female.

Direct crosses

P	$X^W X^W$	X	$X^w Y$	
	Red eyed female	↓	White eyed male	
G	(X_w)	↓	(X^w) (y)	
F_1	$X_w X_w$		$X^w y$	
	Red eyed female		Red eyed male	
G	(X^W) $X(^w)$		X^w Y	
F_2	$X^W X^W$	$X^W X^w$	$X^W y$	$X^w y$
	Red eyed female	Red eyed female	Red eyed male	White eyed male

Reciprocal cross

P	$X^w X^w$	X		XW^Y
	White eyed female	↓		Red eyed male
G	(X^w)			(X^w) (y)
F_1	$X^W X^w$	↓		$X_w y$
	Red eyed female			White eyed male
G	(X^W) X $(^w)$			$X^w Y$
F_2	$X^W X^w$	$X^w X^w$	$X^W y$	$X^w y$
	Red eyed female	White eyed female	Red eyed male	White eyed male

Fig: Inheritance of eye colour in *Drosophila*

In the normal Mendelian inheritance, the F2 ratio does not differ from that of reciprocal cross. But in the inheritance of eye color in Drosophila, the F2 ratio depends on the sex of the parent by which eye color is introduced.

In *Drosophila*, the white eye color follows a criss cross inheritance. The kind of inheritance from father to grand son only through daughter is called cris – cross inheritance. The male transmits his red eye color to his grand son through his daughters, never to or through his sons. The male transmits his X chromosome to his grand sons only through his daughter, never to or through his sons. Thus, the transmissions of eye color and X chromosome are similar. Hence, it is assumed that the gene for eye color is located in the X chromosome and Y chromosome carries no allele for eye color.

Holandric genes

Most sex-linked genes in male heterogametic animals are on the X chromosome. However Y chromosome also contains few genes that produce visible effects on the phenotype of the organism. Such genes are called Y linked or holandric genes. Holandric genes would be transmitted directly fonn father to son and never appear in females.

Sex-influential dominance / Sex influenced character

The condition in which the same gene acts as dominant in one sex and recessive in other sex is called as sex-influenced dominance. That is, the sex influences the gene either to be dominance. That is, the sex influences the gene either to be dominant or recessiye. The sex

influenced genes are present in autosomes. This differential behavior of the gene is due to female and male sex hormones.

For example in human being baldness in due to sex-influenced gene. This trait is dominant in men and recessive in wome. A man is bald in homozygous recessive as well on heterozygous condition for baldness. Whereas women exhibit baldness only in homozygous recessive condition for baldness and heterozygous condition for baldness in female sex produce normal phenotype.

$H^N H^N$ - Normal female and normal male

$H^N H^B$ - Normal female and bald male

$H^B H^B$ - bald female and male

Sex-limited gene expression / sex limited characters

Sex limited genes are those which produce characteristics that are expressed in only one of the sexed. Sex limited genes may be located on any of the chromosomes. The sex hormones are found to be a limiting factor in the expression of sex limited gene. Sex limited genes are responsible for secondary sexual characteristics.

For example beard in man and breast in women are produced by sex-limited genes. A woman does not have a beard, thought she carries all the genes necessary for beard. Similarly man does not have breasts though he carries all the genes necessary for breast. The expression of sex-limited characteristics depends upon the presence or absence of sex hormones.

CYTOPLASMIC INHERITANCE

Introduction

Besides chromosomes, various organelles of cytoplasm also contain DNA. The mitochondria genetic characters themselves. Themechanism in whichcytoplasmic inclusions (e.g., alpha, beta sigma and kappa particles) and organelles (plastids, mitochondria, centriole, etc) take part in transmission of characters from generation to generation is called cytoplasmic inheritance. Since cytoplasmic inheritance is based on cytoplasmically located, independent, self-replicating, extra chromosomal DNA molecules, it is also called extra chromosomal inheritance.

The smallest inheritable extra chromosomal unit is called as plasma gene and all the plasmagenes of a cell constitute the Plasmon (like the genome).

Cytoplasmic inheritance is due to the plasmagenes, located in cell organelles that are integral constituents of normal cells. The characteristic features of this inheritance are summarized below.

1. Differences in reciprocal crosses

In Mendelian inheritance, the results of reciprocal crosses are identical (one exceptional – sex linked inheritance). If the character is transmitted through cytoplasm, the reciprocal cross results will be different.

2. Somatic segregation

Plasma genes generally show somatic segregation during mitosis, a feature of rare occurrence in the case of nuclear genes.

3. Non – mappability

Gene controlled characters show linkages and hence they are mappable. But the characters transmitted through cytoplasm show no linkage. Hence, they are non mappable.

4. Non-Segregation

Segregation is typical of Mendelian heredity. The cytoplasmic heredity fails to show segregation. Sometimes, segregation may occur in cytoplasmic heredity also. But it will not be consistent with the segregation of chromosomes.

5. Indifference to nuclear substitution

When the nucleus is transplanted, no change is found in the cytoplasmic inheritance.

6. Infection like transmission

Cytoplasmic inheritance seems like intection through some agents.

Maternal inheritance

Maternal effects are produced due to the influence of mother's nuclear genotype on the phenotype of its progeny and last for one generation. Characters showing the maternal effect exhibit clear cut differences in F_1 for reciprocal crosses. One of the examples for maternal effect is coiling pattern of shell in snail *Lininea.* In this snail the direction of coiling of its shell is controlled by single nuclear gene D / d; the dominant allele D produces right-handed coiling, while its recessive allele d produces left-handed coiling. The direction of shell coiling in an individual is governed by the genotype of its female parent and not by its own genotype. As a result, reciprocal crosses show differences in coiling in F_1 and there is no phenotypic segregation in F_2 the phenotypic effect of segregation is observable in F_3 only.

Crosses between females with left-handed coil (dd) and males having right handed coil (DD) produce F1 progeny (Dd) with left-handed coil, since the genotype of the female parent is dd. In F_2 segregation of Dd produces three genotypes (DD, Dd, dd) in the ratio of 1:2:1 But the F_2 snails with DD, Dd as well as dd genotypes exhibit right-handed coiling since their female parent has the genotype Dd which determines right-handed coiling in the progeny (irrespective of the genotypes of the progeny). The F_3 progeny from the F_2 individuals with the genotypes DD and Dd will show right-handed coiling, while those from dd F_2 individuals will exhibit left-handed coiling of their shells; thus produces the typical 3:1 ratio in F_3

The reciprocal cross (DD x dd), on the other hand, yields right-handed coiling in the F_1 (Dd) as well as in the three genotypes, 1DD : 2Dd : 1dd, obtained in the F_2. But in F_3 2/3 of the progenies show right-handed coiling since they are derived from F_2 individuals having the genotypes DD and Dd. The remaining 1/3 of the F_3 progenies exhibit left-handed coiling since their female parents hand the genotype dd; this yields the typical monohybrid ration of 3 :1 in the F_3

The direction of coiling in this snail is determined by the plane or the direction of the first mitotic division of the zygote. The direction of the first mitotic division of the zygote. The plane of the first division, of the other hand, is determined by some substances already present in the egg cell. Obviously, these substances are produced by the female parent; as a result, they would produce the phenotype appropriate for the maternal genotype. Further

genotype of, the zygote itself has no effect of the plane of first division and consequently, on the direction of coiling since its gene products are not involved in determining this trait. As a result, the direction of coiling in an individual is governed by the genotype of its female parent. Therefore, phenotypes appear one generation later than the appearance of the concerned genotypes, producing delayed segregation in F_3

Inheritance of kappa particles in Paramecium

There are two types of strains in paramecium. One has kappa particles in its cytoplasm and other does not have such particles (Fig. 12.1). The presence of kappa particles in the cytoplasm leads to production of a toxin known as paramecin. This toxin can kill the strain *paramecium* that lacks kappa particle. Thus the strain with kappa particle is known as killer strain and that without kappa particle is called as sensitive strain.

The production of kappa particles is dependent on a dominant allele K, so that the killer strains are KK or Kk and sensitive strains are ordinarily kk. In the absence of dominant allele K, kappa particles can not multiply and in the absence of kappa particles dominant allele k cannot produce them de novo.

If the killer (KK) and sensitive (kk) and sensitive (kk) strains are allowed to conjugate, all exconjugants (the cells separating after conjugation) will have the same genotype Kk., The pheno9types of these exconjugants will however depend upon duration of which conjugation is allowed. If conjugation does not persist long enough for exchange of cytoplasm, heterozygote (Kk) exconjugants will only have parental phenotypes. It means that killers and sensitive will remain as sensitive after conjugation. If conjugation persists, sensitive strain will receive kappa particles and will become killer, so that exconjugants will be killers having genotype Kk.

Plastid inheritance

Plastids are minute cytoplasmic organelles in plant cells. Most important are the chloroplastids, which carry chlorophyll. Plastids arise from smaller cytoplasmic particles (plastid primordia) that contain DNA. They duplicate themselves independently. They are transmitted through the cytoplasm of the egg.

The Four- O' clock plant, *Mirabilis jalapa*, has branches that produce either green, white or mixed green-white (variegarted) leaves. In crosses between flowers of these branches the, offspring are all green if the maternal parent is a flower from a green branch. Such

offspring remain green throughout subsequent generations as long as the maternal plant is green. Similarly, as long as the maternal parent is from a white branch, the offspring are all white, when variegated branches are used as female source; both green and pale plastids are present in cells of female parent in cells of female parent. Therefore, female gametes may carry either green or pale plastids or both. Consequently, three kinds of plants namely green pale and variegated plants would be obtained (Table 12.1).

Table 12.1. Inheritance of leaf colour in *Mirabilis jalapa*

Egg source	Pollen Source	Progeny
White	Green White Variegated	White
Green	Green White Variegated	Green
Variegated	Green White Variegated	Green White Variegated

Plasmids

Plasmids are called episomes. They are extra chromosomal, circular, covalently closed double stranded DNA molecules found in bacteria. In effect, plasmids are accessory chromosomes. Plasmids can replicate autonomously of the host chromosome. The size of plasmid ranges from two to several hundred kilobases.

Plasmids carry genes for the inactivation of antibiotics, metabolism of natural products and production of toxins. The F factors are important plasmids of *Escherichia coli.*

Mitochondria (mt DNA)

Mitochondrion is present in living organisms arise from pre existing mitochondria. They are small cytoplasmic organelles present in animal and plant cells but not present in bacteria and viruses. Mitochondria provide cellular energy through oxidative phosphorilation. Mitochondria contain a small circular DNA molecule codes for limited number of structures and functions. The size of mtDNA ranges from about 16 kb in mammals upto several hundred kilo base pairs in higher plants (eg 570 kb in maize) and mt DNA usually found in multiple copies per organelle. The mtDNA play a significant role in crop improvement. Recent evidences showed hat the cytoplasmic genetic male sterility of mitochondrial genome to the nuclear genome.

Chloroplast DNA

Chloroplast of the plant cell contain circular DNA molecule which are self – replicating in nature. The isolated chloroplast found to capable of protein synthesis in the presence of light. The DNA analysis revealed that 30-60 copies of the chloroplast genome are found in each chloroplast of higher plants. The chloroplast genome contain herbicidal resistant and streptomycin resistant genes

DNA AS GENETIC MATERIAL

Introduction

Griffith experiment

The phenomenon of transformation *pnemococcus* bacterium was discovered Fredrick Griffith in1928. There are two types *pneumococcus* bacteria – *virulent* (pathogenic) and *Aviruler* (non-pathogenic) Virulent strains have polysaccharid capsules and give smooth colonies. *Avirulent* strains have capsules and give rough colonies. The virulent strain has antigenic property with serotype III and *Avirulent* has serotype II.

Virulence	Colony morphology	Serotype	Designation
Virulent	Smooth	III	III S
Avirulent	Rough	II	II R

Live II R and heat killed IIIS are not lethal when injected into the mice separately. But a mixture of live II R and heat killed IIIS was lethal to the mice. The blood of dead mice contained live IIIS. The heat killed IIIS had transformed live II R into live IIIS. Griffith (1928) called this phenomenon transformation. Thus the transformation of non virulent Type II R cells to virulent Type IIIS cells cannot be explained by mutation; rather some component of the dead Type IIIS cells must convert living Type II R cells to type IIIS.

The same phenomenon occurred in the test tube when live Type II R cells were grown in the presence of dead type III S or extract of Type IIIS cells. It was clearly shows stage for determining the chemical basis of hereditary in Pneumococcus.

Proof that the DNA is the genetic material

The first direct evidence showing that the genetic material is DNA rather than protein or RNA was published by O. T. Avery, C. M. Macleod and M. Mc Carty in 1944. The most definite experiments conducted by them provided that the DNA was the transforming princip (DNA is the genetic material) involved the use of enzymes that degrade DNA, RNA, or protein. In separate experiments highly purified DNA from Type IIIS cells was treated with (1) Deoxyribonucleic (DNA ase which degrades DNA) (2) Ribo nuclease (RNA ase which degrades RNA) or (3) Proteases (which degrade proteins) and then tested for its ability to

transform Type. II R cells to Type IIIS. Only Deoxyribonuclease had any effect on the transforming activity of the DNA preparation. It totally eliminated all transforming activity. The results obtained by Avery and co-workers clearly established that the genetic information in pneumococcus was present in DNA. We now know that the segment of DNA in the chromosome of pneumococcus that carries the genetic information specifying the synthesis of a Type III capsule is physically integrated into the chromosome of the Type II R recipient cell by a specific recombination process occurring during transformation.

The Hershey – Chase Experiment

The additional direct evidence indicating that DNA is the genetic material was published in 1952 by A. D Hershey (1969 Nobel Prize Winner) and M. Chase. These experiments showed that the genetic information of a particular bacterial virus (bacteriophage T_2) was present in DNA.

Viruses are the smallest living organisms. They never as such enters the cell' only the tail contacts the host and enzymatically cuts a small hote through the membrane and then the nucleic acid of the virus head flows to the cell. This idea was tested by Hershey and chase in the following way. Phase DNA was labeled with radio-isotope 32P in place of in place of normal isotope 31P where as protein coat was labeled with 35S in the place of normal isotope 32S. These labels are highly specific, because DNA does not contain sulphur and the protein coat is devoid of phosphorus. A sample of an E. coli culture as infected with labeled T2 phage. After a short incubation period, the suspension was spun for a few minutes in warring Blender at 10,000 rpm. This treatment served the connections between the viruses and bacteria. The resulting suspension was centrifuged. The pellet contained infected bacteria, where as supernatant conteined smaller particles. These fractions were analysed for 32P and 35S to determine the location of the phage DNA and the protein coat. The results of the experiment were:

1. Most of the phage DNA was found in the bacteria
2. Most of the phage protein was found in the supernatant
3. The blender treatment did not prevent the infection
4. The progeny of T phage contained the parental ^{32}P and not the parental ^{35}P.

5. This led to the conclusion that the phage has a genetic part-DNA and non-genetic protective part-protein. The protein coat serves as a vehicle. Only DNA carry necessary information for the new generation of phages.

RNA as genetic material in small viruses

In viruses the genetic information are present either in DNA or RNA. Tobacco mosaic virus (TMV) is an RNA virus. It consists of a single molecule of RNA surrounded by a protein coat (fig. 13.4). By using the appropriate chemical treatments, one can separate the protein coats of TMV from RNA. Moreover, this process is reversible; by mixing the proteins and RNA under appropriate conditions "reconstitution" will occur producing complete infective TMV particles.

Frankel-conrat and Singer (1957) took two different strain of TMV' separated the RNA from the protein coat and reconstituted mixed virus by mixing the proteins of one strain with the RNA of second strain and vice-versa. When these mixed virus were used to infect tobacco leaves, the progeny viruses produced were always found to be phenotypically and genotypically identical to the parent strain from which the RNA had been obtained. Thus the genetic information of TMV is stored in RNA and, not in protein.

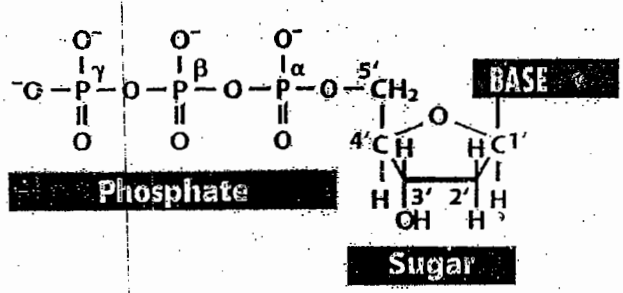
DNA

Introduction

Deoxy ribonucleic acid

DNA is a long thread like unbranched polymeric molecule of heredity. DNA molecule composed of repeating sub units called nucleotides. Each nucleotide composed of (i) a phosphate group (ii) a five carbon deoxyribose sugar (iii) cyclic nitrogen containing compound called nitrogenous base (Fig. 14.1).

(A) A nucleotide



(B) The four bases in DNA

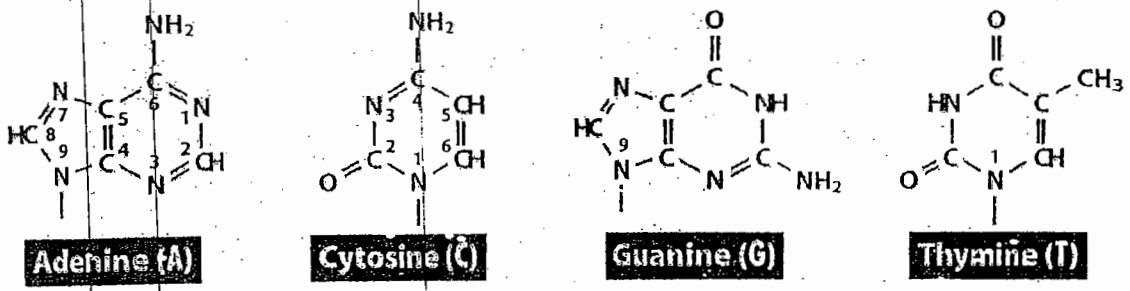


Fig.:Chemical structure of the different structural components of DNA

Structure of DNA molecule (Watson and Crick's DNA double helix model)

The correct structure of DNA was proposed by J.D. Watson and F.H.C. Crick (1953). The double helix model proposed by them is based on two evidences.

1. Chargaff's chemical analysis

Chargaff's (1950) found that a specific quantitative relationship is present between purines and pyrimidines of DNA molecule. In DNA molecule, the ratio of adenine to thymine and guanine to cytosine are 1:1 that is

Amount of purine	=	Amount of pyrimidine
A+G	=	T+C
A	=	T
G	=	C
AT	#	GC

It is called as Chargaff's rule.

2. Crystallographic studies by Wilkins and Franklins

The X-ray diffraction patterns and crystallographic data on DNA structure form studies of M.H.F. Wilkins and R. Franklins showed that DNA is highly ordered multiple strand structure with repeating subunit structures spaced in every 3.4 A^0 along the axis of the molecule.

Structure of DNA

On the basis of Chargaff's chemical data and crystallographic data by Wilkins and Franklins. Watson and Crick proposed the structure of DNA. The important features of their model are (Fig. 14.2.).

1. DNA exists in double helix in which two polynucleotide chain coiled about one another in spiral way.
2. Each polynucleotide chain consist of sequence of nucleotides linked together by phosphodiester bonds and two polynucleotide chains held together by hydrogen bonding between bases.
3. The base pairs are stacked between two chains perpendicular to the axis of the molecule similar to the steps of a spiral staircase. The base pairs in DNA stacked 3.4 A^0 apart with 10 base pairs per turn (360^0) of the double helix.
4. The base pairing in DNA molecule is specific ie Adenine pairs with Thymine (A = T) and Cytosine pair with guanine (C = G). So each base pair consists one purine and one pyrimidine (Fig. 14.3.)
5. The two strands of the DNA are complementary in nature (non-identical) *ie* once the sequences of bases is one strand is known, the sequences of bases in the other strand is also known because of specific base pairing. The complementary nature is very important for storing and transmitting the genetic information.

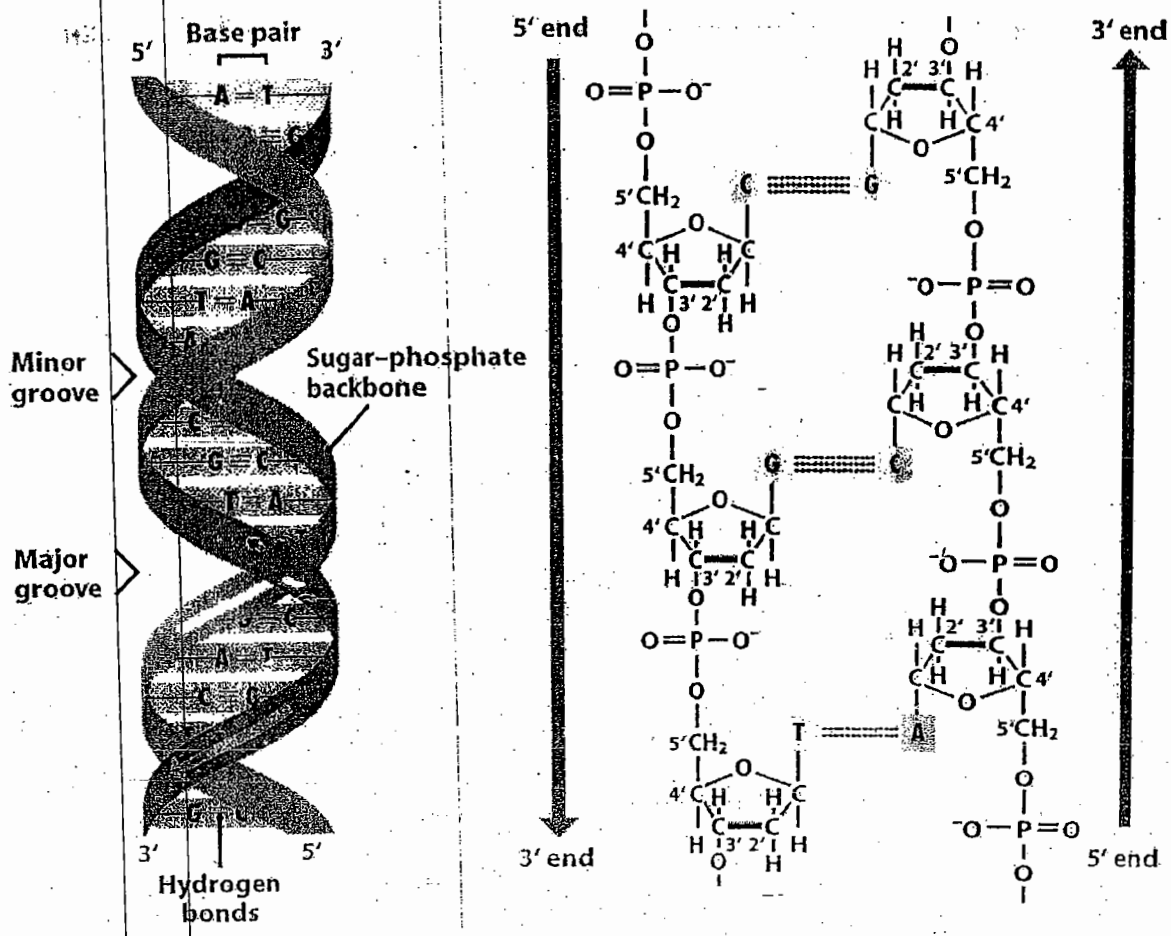


Fig.: The Watson Crick model of double stranded DNA molecule

6. The purine and pyrimidine bases are on the inside of the helix where as the phosphate and deoxyribose unit are on the outside. The sugar phosphate backbones of the two complementary strands are antiparallel. Among two strand of DNA, one strand go from 3' carbon of one nucleotide to 5' carbon of the adjacent nucleotide. Where as the complementary strand go from 5' to 3' carbon. This mechanism is very important in considering the mechanism of replication of DNA.
7. The high degree of stability of DNA is due to more number of hydrogen bonds.

DNA replication

Watson and Crick proposed that during DNA replication, the two strands of DNA molecule separate after the breakage of hydrogen bonds and each strand acts as a template for the synthesis of a new companion strand. Thus, resulting daughter DNA molecules each containing an old strand derived from parent DNA molecule and another strand newly synthesized. This type of distribution of parental strands is called as semi-conservative.

However in considering possible mechanisms of DNA replication, three different hypothetical modes are apparent in addition to semi conservative method of replication.

1. Semi-conservative method

In this method two strands of the parental DNA molecule separates and each strand act as a template for the synthesis of new complementary strand. Thus resulting daughter DNA molecules each contain one old strand derived from parental DNA molecule and another strand newly synthesis of new complementary strand. Thus resulting daughter DNA molecules each contain one old strand derived from parental DNA molecule and another strand newly synthesized

2. Conservative method

In this method, two parental DNA strand separates and each strand act as a template for synthesizing a complementary strand. The resulting progeny DNA molecule composed of two newly synthesized strand and parental DNA strands remain intact (totally conserved after replication)

3. Dispersive method

In this method a segments of parental strands and progeny or nascent strands become interspersed through some kind of fragmentation, synthesis and rejoining process.

Proof for semi-conservative method of DNA replication Meselson and Stahl's Experiment

E. coli were grown for many generations in a medium that contained ^{15}N, the heavy isotope as the sole nitrogen source (added as $^{15}NH_4CL$.) Their DNA thus labeled with ^{15}N, was denser than ordinary DNA. This density difference can be distinguished by 'cesium chloride density gradient equilibrium sedimentation technique' then these bacterial were transferred to ^{14}N medium. The bacterial cells were sampled at various times to ascertain the density of DNA. The sample time corresponded with the doubling of the cells. After one generation, the DNA of daughter bacteria had neither original ^{15}N density nor the pure ^{14}N density. Instead, this DNA had an intermediate (or) hybrid density. Instead, this DNA had an intermediate (or) hybrid density (precisely between ^{15}N – ^{14}N densities). The absence of ^{15}N DNA indicated that the parental DNA was not synthesized entirely *denova* (afresh). Of two strands of the daughter DNA molecule, one strand was derived from the parent ^{15}N DNA and

the other strand was newly synthesized from ^{14}N source. Hence, the daughter (^{15}N ^{14}N) gives an intermediate (or hybrid) density.

Meselson and Stahl concluded that during DNA duplications, each daughter molecule receives one strand from the parent molecule. This strand is conserved through much duplication. Their results agreed perfectly with the Watson – Crick hypothesis of DNA replication.

Mechanism of DNA replication

1. Relaxation of packed DNA

The highly packed super twisted DNA molecules are relaxed by the enzyme, DNA gyrase.

2. Unwinding of DNA strands

An enzyme rep protein (helicase) unwinds DNA helix. The unwinding is powered by hydrolysis of ATP. About two ATP are consumed for each base pair separation. The separated strands are stabilized by S-s binding proteins.

3. DNA polymerase

New DNA strand is synthesized by the enzyme DNA polymerase, with the old DNA strand acting as the template. There are three kinds of DNA polymerases – I, II and III. DNA polymerase I was discovered by Kornberg hence called Kornberg enzyme. DNA polymerases II and III were later discovered by De Lucia and Cairns. The catalytic rates of the three enzymes differ: 10 nucleotides per second are added by polymerase I. 0.5 per second by polymerase II and 150 per second by polymcrase III. DNA polymerase I catalyses the step-by-step addition of deoxyribonucleotides units to form a DNA chain. It adds deoxyribonucleotides to the 3' – hydroxyl terminus of a pre existing DNA (or RNA) strand. That, is a primer chain with a free 3' – OH group is required for the action of DNA polymerase I. In other words, DNA polymerase can not initiate the DNA chain but only elongates the chain.

4. DNA synthesis is primed by RNA

DNA polymerase requires a primer with a free 3' – OH group for the initiation of DNA synthesis. Hence, DNA polymerase can not initiate the DNA synthesis. Since RNA polymerase can start chains *denova*, RNA primes the synthesis of DNA.

5. Elongation o f new DNA strand

DNA Polymerase III begins to add deoxyribonucletides to the 3' – hydroxyl end of the RNA primer. The chain elongation reaction occurs by means of a nucleophilic attack of the 3' – OH terminus of the primer on the inner most phosphorus atom of the incoming deoxyribo nucleoside triphosphate a phosphor diester bridge is formed and pyrophosphate is released. The subsequent hydrolysis of pyrophosphate drives the polymerization forward. The elongation of the DNA chain proceeds in the 5→3 direction. The polymerization is processive that is, many nucleotides are added without the release of the enzyme from the template. DNA polymerase catalyses the formation of a phosphor-diester bond only if the base on the incoming nucleotide is complementary to the base on the template stand. Thus DNA polymerase is a template directed strand.

6. Proof reading by DNA polymerase-I

DNA polymerase-I have (i) 5' →3' polymerase activity (ii) 3' → 5'exonucleus activity and (iii) 5'→ 3' exonuclease activity.

i. *Function of 5'→3' exonuclease activity*

- Step by step addition of nucleotides in the chain elongation

ii *Function of 5'→3' polymerase activity*

- DNA repair: Removal of DNA segment damaged by UV – rays
- Removal of RNA primer: Which are used in DNA synthesis

iii *Function 3' → 5' exanucleose activity*

Used for proof reading and editing function.

7 **Continuous and dis-continuous synthesis of DNA strands**

RNA

Introduction

Ribonucleic acid

RNA is found in cells of all living organisms. It is found both in chromosomes in nucleus and ribosomes in cytoplasm. It contains ribose sugar, nitrogen bases and phosphate group. The nitrogen bases include adenine, guanine, cytosine and uracil and pairing occurs between AU and GC. The function of RNA is transfer of genetic messag- from nucleus to the cytoplasm and synthesis of protein in the ribosomes. In some viruses, RNA acts as the genetic material and regulates the gene action.

Flow of genetic information

DNA ⟶ RNA ⟶ Protein

(Transcription) (Translation)

Structure of RNA

RNA is a long unbranched polymer consists of nucleiotides joined by phosphor-diester bonds. RNA differs from DNA in two ways.

- RNA is single Stranded; DNA is double stranded.
- RNA contains ribose sugars where as DNA contains deoxyribose sugar.
- RNA contains the uracil in Place of thymine. Uracil lack Methyl group present in thymine.
- RNA molecules are generally much shorter than DNA molecules.

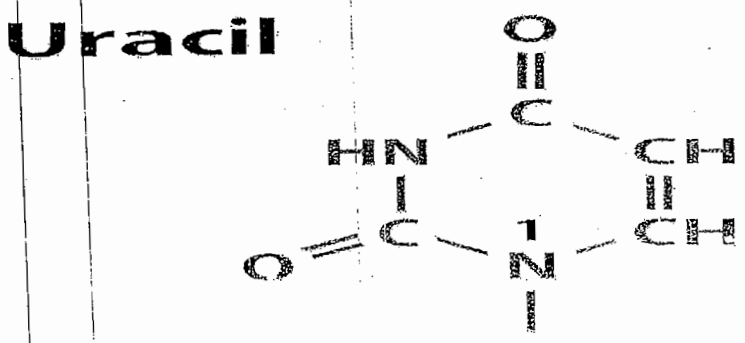


Fig.:Structure of Uracil

Three types RNA

1. Messenger RNA or mRNA

It is a kind of single strand RNA molecule which is complementary to sense strand of DNA molecule produced by transcription of structural genes in the DNA sequence. mRNA carries the genetic message from the chromosome to the site of protein synthesis i.e. ribosome. mRNA molecule corresponds to each gene that is expressed.

2. Transfer RNA or RNA

- tRNA is a kind of RNA molecule consists of 75 nucleotides and it become smallest of all RNA molecules.
- They carry the activated amino acids to the ribosome for protein synthesis.
- There is a specific tRNA for each of the twenty amino acids.
- The 5' end of tRNA have poly G and it is phosphorilated.
- tRNA contains many unusual bases between 7 and 15 per molecule.
- tRNA is folded into a clover leaf pattern. It has five following special region as follows.

I.CCA end

The base sequence in 3' end of all tRNA is CCA. The activated amino acid is attached to 3' hydroxyl group of the terminal adenosine.

ii.T Ψ C arm

Involved in the binding of the t-RNA to ribosome.

iii.Auticoden loop

It consists of seven bases with following sequences.

Pyrimidine – pyrimidine - X-Y-Z - Purine modified

Anticodon variable
(coden recogrition site)

Condon recognition site is complementary to codons of mRNA.

iv. DHUarm

It is the site for the recognition of amino acid activated enzymes.

v. Extra arm

Some tRNA have extra arm.

3. Ribosomal RNA or rRNA

i. It is a kind of RNA molecule serving as a major component of ribosomes.

ii. *E. coli* has three kind of rRNA i.e. 23s, 16s, 5 s

iii rRNA is transcripl of rRNA genes

GENETIC CODE

Introduction

As it becomes evident that the genes controlled the structure of polypeptides, attention focused on how the sequence of four base pairs in DNA control the sequence of 20 amino acids found in proteins.

Definition of genetic code

The genetic code is the relationship between the sequences of bases in DNA (or its mRNA) and the sequence of amino acid in the protein. The sequence of three nucleotides in DNA (or its mRNA) that specifies a particular amino acid in the protein synthesis is called genetic code.

Co-linearity between Gene and protein

Benzer revealed that there is a linear correspondence between a gene and its polypeptide products. This co-linearity gives the clue that specific arrangement of nitrogenous bases in DNA determines the specific sequence of amino acid in protein. So the genetic information is written by four-letter language of DNA nitrogen base.

Characteristic features of genetic code

1. Triplet code

There are 20 kinds of amino acids in the cytoplasm but only four kinds of nitrogenous bases A singlet code is inadequate because it codes for only 4 amino acids and also a doublet code is also inadequate since it codes for 4 x 4 = 16 amino acids only. A triplet code is only adequate since it codes for 64(4 x 4 x 4) amino acids (Table 16.1)

A group of three bases that codes for one amino acid is called codon.

2. Codons have degeneracy

The occurrence of more than one codon per amino acid is called degeneracy. The amino acids like methinine and tryptophan has only one codon each. Where as the amino acids leucine, serine and arginine have six codon each. The codons that specify the same amino acid are called synonyms. For example the codons CAU and CAC code for histidine and most of the synonyms differ only in the last base of the triplet. The degeneracy helps for minimizing the deleterious effect of mutation

Table 16.1. Amino acids coded by the 64 possible codons of the triplet code

First base position 5 end	*Second base position*				*Third base position 3 end*
	U	*C*	*A*	*G*	
U	UUU Phe	UCU Ser	UAU Tyr	UGC Cys	U
	UUC Phe	UCC Ser	UAC Tyr	UGC Cys	C
	UUA Leu	UCA Ser	UAA Stop	UGA Stop	A
	UUG Leu	UCG Ser	UAG Stop	UGG Trp	G
C	CUU Leu	CCU Pro	CAU His	CGU Arg	U
	CUC Leu	CCC Pro	CAC His	CGC Arg	C
	CUA Leu	CCA Pro	CAA Glu	CGA Arg	A
	CUG Leu	CCG Pro	CAG Glu	CGG Arg	G
A	AUU Ile	ACU Thr	AAU Asn	AGU Ser	U
	AUC Ile	ACC Thr	AAC Asn	AGC Ser	C
	AUA Ile	ACA Thr	AAA Lys	AGA Arg	A
	AUG Met	ACG Thr	AAG Lys	AGG Arg	G
G	GUU Val	GCU Ala	GAU Asp	GGU Gly	U
	GUC Val	GCC Ala	GAC Asp	GGC Gly	C
	GUA Val	GCA Ala	GAA Glu	GGA Gly	A
	GUG Val	GCG Ala	GAG Glu	GGG Gly	G

3. Codons have wobbling

The hydrogen bonding between the bases in the anticodon of tRNA and the codon of mRNA appears to follow strict base pairing rules only for the first two bases of the codon. The base pairing involving third third base of the codon is apparently less stringent, allowing wobbling at this site.

5' base in anticodon	3' base in codon in the Mrna
G	U or C
C	G
A	U
U	A or G
I (inosine)	A,U,C

4. Genetic code in non-overlapping

It means that no single base can take part in the formation of more than one codan

5. Genetic code is commaless

The genetic code is commaless, which means that no codon is reserved for punclautions.

6. Start and stop codon (initiation and termination codon)

UAG (Ochre), UAA (Amber) and UGA (opal) are only the three codons that donot specify amino acid. They designate chain termination. These codons are not read by tRNA molecules but by specific proteins called release factors. The codon AUG (methlomine) and GUG (valine) act as starting codon for translation.

7. Genetic code in universal

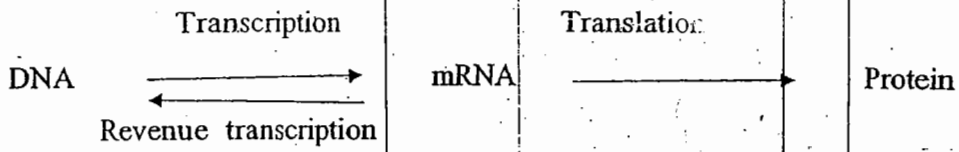
With one or two exceptions the genetic code is same or nearly same in all the organisms. The codon UGA code for tryptophan in human mitochondrial system. Whereas UGA is a termination codon for non-mitochondrial system. Mutation that produce chain – termination triplets within genes is called as non-sense mutation Whereas mis-sense mutation cause change a triplet to another triplet specifying a different amino acids.

PROTEIN SYNTHESIS

Introduction

I. The central dogma

The sequence of amino acids in a protein is determined by the base sequence of DNA. through the m RNA. The flow of genetic information is



II. Transcription

1. Only one strand of DNA is transcribed

RNA polymerase synthesized m RNA with DNA acting as temple. Synthesis of m RNA is called transcription. Only one strand of DNA is transcribed. The other strand is never used for transcription.

2. RNA polymerase

RNA polymerase is a holoenzyme, made up of six polypepildes and two of these polypeptides are identical and thus enzymes consists of five distinct sub unit 2α, β, β′, ώ and o. RNA polymerase without the sigma sub unit is called the core enzyme (2α, β, β′ , ώ). The β′ sub unit causes binding with DNA template and the β sub unit helps in the binding of ribonucleoside triphosphate. The σ sub unit participates in the selection of initiation sites for transcription. The synthesis of m RNA by RNA polymerase takes place in three stages (i) initiation of transcription (ii) elongation and (iii) termination.

i. Initiation of transcription

The transcription starts at specific sites, called promoters on the DNA template. The promoter sites consist of about 40 base pairs (about 140 A^0). The 2α, β, β′, ώ core of RNA polymerase is unable to start transcription at promoter sites. The 2α, β, β′, ώ holoenzyme is essential for specific initiation. In addition, sigma subunit dissociates from the holoenzyme after the new RNA chain is started. The core polymerase continues to transcribe the DNA template. The role of the holoenzyme is selection and initiation, whereas that of the core

enzyme is elongation. Repressors block the transcription by interfering with the binding of RNA polymerase. RNA chain has a triphosphate group at its 5′ terminus and a free hydroxyl group at its 3′ terminus. In contrast with DNA synthesis, primer is not needed. RNA chains can be formed *denovo*.

ii. Elongation of transcription

As in DNA synthesis, the growth of an RNA chain is in the 5′ → 3′ direction. The RNA polymerase moves along the DNA template strand in the 3′ → 5′ direction because the template strand is antiparallel to the newly synthesized RNA strand. The same RNA polymerase molecule synthesized an entire transcript that is, transcription is processive. The transcribed region of DNA remains its double – helical conformation as the next section of DNA unwinds. The maximum rate of elongation is about nucleotides per second.

In contrast with DNA polymerase, RNA polymerase does not edit the nascent polynucleotide chain. Hence, the fidelity of transcription is much lower than that of replication. The error rate of RNA synthesis is of the order of one mistake per 10^4 or 10^5. The lower fidelity of RNA synthesis can be tolerated because; a cell synthesized many RNA transcripts of a gene.

iii Termination of transcription

The DNA template contains stop signals for transcription. Before the termination site, GC rich region is followed by an AT rich sequence. The most distinctive feature of termination sequences is the two fold symmetry of their GC rich region. Hence, the RNA transcript of this region is self-complementary and so, it can base pair to form a hairpin structure. In addition, the nascent RNA chain ends with several U residues, which are specified by a series of A bases in the AT – rich region of the DNA template. These structural features cause RNA polymerase to pause when it encounters such a signal. The rho protein participates in the termination of transcription.

II. Translation

Synthesis of polypeptide chain from mRNA molecule is called translation.

1. Activation and linkage of amino acids to tRNA

The amino acids are activated by ATP. In the first step, the carboxyl group of the amino acyl adenylate and releasing pyrophosphate. This reaction is catalyzed by *amino acyl synthetase* in the presence of magnesium. The amino acyl synthetase is amino acid specific.

In the second step, the enzyme bound amino acyl adenylate reacts with tRNA and forms amino acyl – tRNA. The same enzyme acts to transfer the amino acid to tRNA. When amino acyl tRNA product is formed, adenosine monophosphate and the amino acyl synthetase are released.

The attachment of a amino acid to a tRNA is important because

i. Amino acids themselves cannot recognize the codons on mRNA.
ii. Amino acids are carried to the ribosomes by specific tRNA which recognizes codons on mRNA.
iii. Thus, the specific tRNA act as adaptor molecules.

Specificity of aminoacyl – tRNA synthetases

The correct translation the genetic message depends on the high degree of speciticity of amino acyl – tRNA synthetases. The amino-acyl tRNA synthetase contains two sites synthetic site and hydrolytic site. The synthetic site rejects amino acids that are larger than the correct one whereas the hydrolytic site destroys activated intermediates that are smaller than the correct species. In fact, the synthetase corrects its own errors.

2. Wobble hypothesis

A codon of m RNA is recognized by the anticodon of a tRNA bases of codon forms a Watson – Crick type of base pair with a complementary base on the anticodon. The codon and anticodon are antiparallel in b ase pairing. Some tRNA molecules can recognize more than one codon. For example, the yeast alanine tRNA binds to three codons GCU, GCC and GCA. This is explained by Wobble hypothesis.

3. Ribosomes – the site of protein synthesis S - Svedberg.

The E. coli ribosome has a sedimentation coefficient of 70 S. It can be dissociated into a large subumit (50 S) and a small subunit (30 S). These sub-units can be further split into their constituent proteins and RNAs. The 30 S subunit contains twenty one proteins and a 16 S RNA molecule. The 50 S subunit contains about thirty four proteins and two RNA molecules, a 23 S species and a 5 S species. The formation of a ribosome in vitro is a self - assembly process, That is, non-ribosomal factors are not needed.

The ribosome of eukaryotic cells is larger. It has sedimentation coefficient of 80 S. It dissociates into 60 S and 40 S sub units. Many ribosomes can simultaneously translate an mRNA. This increases the efficiency of utilization of the mRNA. The group of ribosomes

bound to an mRNA is called a pol ribosome or a polysome. In this unit, the ribosomes operate independently, each synthesizing a complete polypeptide chain. The maximum density of ribosomes on mRNA is about one ribosome per eighty nucleotides. Ribosomes near the 5′ end of the mRNA have shortest polyptidee chains, whereas those near the 3′ end have almost finished chains. Ribosomes dissociate into 30 S and 50 S subunits after the polypeptide product is released.

4. Initiation of translation

Protein synthesis is initiated by formyl meth onine tRNA

N-formyl methionine (f Met) is a modified methionine which has a formyl group attached to its terminal amino group. Ablocked amino acid like N-formyl methionine can be used only to start protein synthesis.

Protein synthesis starts with the association of mRNA, a 30 S ribosomal sub-unit and formylmethionyl – tRNA to form a 30 S initiation complex. The formation of this complex requires GTT and three protein factors called, 1F-1,1F-2, and 1F-3. 1F-3 mediates the binding of mRNA to a 30 S subunits forms coming together to form a dead end 70 S complex devoid o f m RNA. 1F-1 and 1F-2 enhance the binding of initiator tRNA to the mRNA – 30 S subunit complex.

A 50 S ribosomal subunit then joins a 30 S initiation complex to form a 70S initiation complex. The bound GTP is hydrolyzed in this step. The 70S initiation complex is ready for the elongation phase of protein synthesis. Ribosome contains two sites A and P. The Met – tRNA molecule occupies the P (peptidyl) site on the ribosome. The other site for a tRNA molecule on the ribosome, the A (aminoacyl) site is empty. The anticodon of f Met – tRNA pairs with the initiatin AUG (or GUA) codon on mRNA. Thus, the reading frame is defined by specific interactions of the ribosome and of Met RNA with RNA.

5. Elongation of Translation

The elongation cycle in protein synthesis consists of three steps:

(i) Binding of aminoacyl - tRNA (codon recognition)

(ii) Peptide bond formation

(iii) Translocation

(i) Binding of amino acyl tRNA in "A site"

The cycle begins with the insertion of an aminoacyl tRNA into the empty "A site" on the ribosome. The particular species inserted depends on the mRNA Codon that is positioned in the A site. The complementary amino acyl tRNA is delivered to the A site by a protein elongation factors called EF-Tu and EF-T. GTP bound to EF-Tu is hydrolyzed as the amino acyl – tRNA is precisely positioned on the ribosome "A site".

(ii) Formation of a peptide bound

When amino acyl – tRNA occupied the A site and fMet RNA occupies the P site. the stage is set for the formation of a peptide bond. This reaction is catalyzed by *peptidyl transferase* an enzyme that is integral part of the 50 s subunit. The activated formyl methionine unit of fMet – tRNA (in the p site) is transferred to the amino group of the aminoacyl RNA (in the A site) to form a depeptidyl – tRNA.

The tRNA occupies the "P site". Whereas a dipeptidyl tRNA occupies the "A" site, following the formation of a peptide bond (Fig. 17.2)

(iii) Translocation

Three movements occur the discharged tRNA leaves the "P site", and mRNA moves a distance of three nucleotides. The result is that the next codon is positioned in the "A site" for reading by the incoming aminoacyl – tRNA. Translocation requires a third elongation factor, EF-G (translocase). The GTP bound to EF-G is hydrolysed during translocation. After translocation, the "A site" is emply, ready to bind an aminoacyl – tRNA to start another round of elongation.

Normally at each translocation step, the mRNA template advances three nucleotides precisely. The movement of peptidyl tRNA from the "A site" to "P site" plus the mRNA three nucleotides distance correctly. That is the movement of a triplet codon is a comsequence of its binding to an anticodon in tRNA.

6. Termination of translation

Protein synthesis is terminated by realease factors. Aminoacyl – tRNA does not bind to the "A site" of a ribosome if the codon is UAA, UAG or UGA. In normal cells, tRNAs with anticodons complementary to these stop signals are absent. Instead, these stop signals are recognized by release factors, which are proteins. Release factor RF1 recognizes UAA or

UAG, RF2 recognizes UAA or UGA. Thus, proteins can recognize trinucleotide sequences with high specificity. The release factors are bound to GTP. Termination requires energy that is supplied by the splitting of GTP into GDP and phosphate.

The binding of release factor to a termination codon in the "A site" activates peptidyl transferease in such a way to hydrolyze the bond between the polypeptide and the tRNA in the "P site". The polypeptide chain then leaves the ribosome. The 70 S ribosome then dissociates into 30 S and 50 S subunits as the prelude to the synthesis of another protein molecule.

1. INTRODUCTION

Statistics, its meaning and definition

Statistics is a branch of applied mathematics and is concerned with observational data.

The word statistics is generally used in two different ways.

(1) When it is used in plural, it means the quantitative data affected to a marked extent by a multiplicity of causes. When we say `collect statistics' means collect the numerical data which are to be analyzed and interpreted. e.g.

(i) Wheat production affected by various causes.

(ii) Collect the data of height of the students of second semester.

(2) When it is used in singular, it means "the science of collecting, classifying and using the data for further statistical treatments". It involves the methods of analysis used in the analysis and interpretation of data and they are known as statistical methods.

Definitions:

(1) R. A. Fisher: It is a study of population, variation and the methods for reduction of the data.

(2) A. L. Bowley: It is a science of calculations and averages.

(3) Boddington : It is a science of estimate and probability.

All these definitions are not satisfactory because they cover only a part of the subject.

In general : Statistics may be defined as the science and art of collection, organization, presentation, analysis and interpretation of numerical data OR Statistics is concerned with scientific methods for collecting, organizing, summarizing, presenting and analyzing the data as well as drawing valid conclusions and making reasonable decisions on the basis of such analysis.

However, it is not used for all these purposes in all the fields e.g. in administrative and executive departments, statisticians are interested only in collecting and presentation of data. Such as crop yields, birth and death rates etc. On the other hand a researcher employs the methods which relate to design of experiments and analysis of experimental results.

Biometry: When the principles of statistics are applied on living thing or organisms, the science is called biometry.

Statistical methods: The methods by which statistical data are analyzed are called statistical methods.

Aims of studying statistics

(1) To study the population: The study of population of any kind is on the basis of sample data.

(2) To understand the nature of variability e.g. Height of plants. The biological phenomena observed under one set of conditions are never duplicated exactly under another set of similar conditions. Therefore, repetition of experiment is necessary to account all the factors causing variation. In biological phenomena were variation is a rule rather than exception it is this function that has wide application.

(3) To express the facts in summary form (the facts that are based on large number of observations) e. g. It is not possible for one to form a precise idea about the income position of the population of India from the records of individuals.

(4) To provide correct method(s) for taking sample (sampling).

(5) To provide proper method for comparison of two or more things.

(6) It helps in prediction/forecasting the yield of a particular crop for a particular year on the basis of the past records.

Limitations of Statistics

Statistics with its wide applications in almost every sphere of human activity is not without limitations. The following are the important limitations.

1) It does not deal with individual.
2) It deals only with quantitative characters.
3) Statistical results are true only on an average.
4) Statistics can be misused.
5) It does not reveal the entire story.
6) Expert knowledge is must to handle the statistical data.

(1) It does not study individuals

Statistics deals with an aggregate of objects and does not give any specific recognition to the individual items of a series. e. g. the individual figures of agricultural production of any country for a particular year are