COURSE GUIDE

BIO 402
CYTOGENETICS OF PLANTS (2 UNITS)

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CONTENTS **PAGE**

Introduction

Cytogenetics of Plants (Course Code 402) aims to take the learner through the fundamentals of basic cytology of plants. The course intends to expose the learner to the theoretical and practical aspects of plant genetics.

The course describes a wide range of topics starting from the history of plant cytogenetics, to characteristics of chromosomes and variations in chromosomes, their causes and consequences in genetics. It also introduces the learner to a range of knowledge in concepts of plant cytogenetics.

The course is therefore designed to equip those planning to work as geneticist. The course participants will be required to study online resource materials for a minimum of 2 hrs weekly and take assignments. Cytogenetics of plants is a 2 credit unit course.

The course guide tells you briefly what the course is all about, what course materials you will be using and how you can work your way through these materials. It gives you some guidance on your Tutor- Marked Assignments.

Course Competencies

Cytogenetics of plants, as a course, provides the candidate with an overall view of the different concepts of inheritance, genetic variation, its causes and the implication on plants. This course also provides information on the consequences of variations observed in animals and humans, citing pertinent examples, for better understanding of the subject matter.

Course Objectives

In addition to the aim of this course, the course sets an overall objective which must be achieved. In addition to the course objectives, each of the units has its own specific objectives. The learner is advised to read properly the specific objectives for each unit at the beginning of that unit. This is with a view to helping the learner to achieve the objectives. As you go through each unit, you should from time to time go back to these objectives to ascertain the level at which you have progressed. By the time you have finished going through this course, you should be able to discuss comfortably on:

- history of plant cytogenetics
- Concepts in Cytogenetics of Plant; and
- Causes and consequences of genetic variations

Working through this Course

In this course, you will be advised to devote your time in reading through the material. You would be required to do all that has been stipulated in the course: study the course units, read the recommended reference textbooks and do all the unit(s) self- assessment exercise(s) and at some points, you are required to submit your assignment (TMAs) for assessment purpose. You should therefore avail yourself of the opportunity of being present during the tutorial sessions so that you would be able to compare knowledge with your colleagues.

Study Units

This course is divided into 3 modules with a total of fifteen units which are divided as follows:

Module 1

Unit 1	Definition and History of Cytogenetics
Unit 2	Chromosome Theory of Inheritance
Unit 3	Chromosome Packaging
Unit 4	Chromosome Morphology
Unit 5	Chromosome Classification Based on Size and Other Attributes

Module 2

Unit 1	Variation in Chromosome Number: An Overview
Unit 2	Monoploidy
Unit 3	Diploidy and Introduction to polyploidy
Unit 4	Triploidy
Unit 5	Tetraploidy and Higher Polyploidy

Module 3

Unit 1	Aneuploidy: An Overview
Unit 2	Aneuploidy and its Genetic Consequences
Unit 3	Variation in Chromosome Structure
Unit 4	Inversion
Unit 5	Translocation

References and Further Readings

You would be required to do all that has been stipulated in the course: study the course units and read the recommended reference textbooks in each unit of the course material.

Presentation Schedule

Presentation schedule for this course will be uploaded on the online course page.

Assessment

You are required to submit your assignment (TMAs) for assessment purpose.

How to get the Most from the Course

The course comes with a list of recommended textbooks. These textbooks are supplement to the course materials so that you can avail yourself of reading further. Therefore, it is advisable you acquire some of these textbooks and read them to broaden your scope of understanding.

Online Facilitation

Online facilitation for this course will hold once in a week for the period of eight weeks. The time and day for the online facilitation will be one hour as indicated in the time table

Course Information

Course Code: BIO 402
Course Title: CYTOGENETICS OFPLANTS
Credit Unit: 2
Course Status: COMPULSORY
Course Blub: This course is designed to provide the students with an overall view of the different concepts of inheritance, genetic variation, its causes and the implication on plants. It also provides information on the consequences of variations observed in animals and humans, citing examples for better understanding of the subject matter.

Semester: SECOND SEMESTER
Course Duration: 13 WEEKS
Required Hours for Study: 65 HOURS

Ice Breaker

I am Prof. Babajide O. Odu, Lecturer in the Obafemi Awolowo University, Ile Ife and external facilitator in National Open University. I am currently the Director, University Research Office (URO) OAU. The links below are my research ID URL:
https://www.linkedin.com/in/babajide-odu-28b87352/
https://scholar.google.com/citations?user=96zKWmgAAAAJ&hl=en
https://scholar.google.com/scholar?scilib=1&scioq=Odu+Babajide&hl=en&as_sdt=0,5
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https://orcid.org/0000-0002-7085-6280
https://www.scopus.com/home.uri#

MAIN COURSE

CONTENTS

Module 1

Unit 1 Definition and History of Cytogenetics
Unit 2 Chromosome Theory of Inheritance
Unit 3 Chromosome Packaging
Unit 4 Chromosome Morphology
Unit 5 Chromosome Classification Based on Size and Other Attributes

Unit 1: Definition and History of Cytogenetics

CONTENT

 1.1. Introduction

Cytogenetics is the branch of genetics, cytology, and cell biology that analyses the nuclear genomes at the chromosome level. Cytogenetics makes the chromosome a substantial target in elementary plant cell biology and other fields such as mutagenesis and genotoxicity studies. Standard cytogenetic methods were, and are still, commonly used. Modern cytogenetic technologies involving advanced microscopy and imaging methods, that progress in the analyses on epigenetic DNA and histone modifications as well as DNA damage by using fluorescent antibodies benefit plant genome structure, dynamics, and evolution.

 1.2. Learning Outcomes

At the end of the class student must:
- be familiar with history of cytogenetics,
- be familiar with the contributors of cytogenetics, and
- understand the development of cytogenetics as a discipline

 1.3. History of Cytogenetics

Cytogenetics was developed from two originally separate sciences – cytology and genetics. To fully understand the development of cytogenetics as a discipline, one has to look into its history. The scientists chosen to be featured in this historical consideration made significant contributions to these sciences, and in this respect represent milestones.

Many other significant contributions were made by other men all of whom could not be mentioned in this account.

Johannes Sachariassen and Zacharias (1588-1631). Janssen – Two Dutch eyeglass makers, father and son, between the years 1591 and 1608 produced the first operational compound microscope. They combined two double convex lenses in a tube. The magnification was not more than ten times, but it nevertheless caused great excitement.

Robert Hooke (1635-1703)
An architect as well as a microscopist and the first curator of the Royal Society of London, in 1665 described cork and other cells and introduced the term **cell**. His was the first drawing ever made of cells.

Microscopes at that time magnified 100 to 200 times with a distortion of shape and color that increased with magnification. Nevertheless, these microscopes revealed many new things. Still, it was necessary to wait for better lenses to see anything more. Scientists waited for 160 years, and during this period they, naturally, argued about what they had seen.

Joseph Gottlieb Kolreuter (1733-1806)
German information about hybrids between plant varieties that might resemble one parent or the other or present a combination of their features. Camerarius was the first to experiment in this field. For a number of years Kolreuter crossed different types of tobacco with one another. Later, he crossed other plant genera such as pinks, *Aquilegia, Verbascum,* and others. One of his most valuable observations on reciprocal crosses showed the equality of contributions from the two parents. Thus, he provided clear evident that in reciprocal crosses, the hereditary contribution of the two parents to their offspring was equal.

Robert Brown (1773-1858)
A Scottish botanist, in 1828 discovered the cell nucleus in the flowering plant *Tradescantia*. Although he practiced medicine as a surgeon for five years, he later abandoned this and turned his efforts toward botanical sciences. He was libration to the Linnaean Society and curator at the British Museum. His remarkable account (1828) of the properties and behavior of the nucleus stand unmodified and without correction. He was a very skillful and careful observer. He also observed the random thermal motion of small particles, still known as **Brownian movement.**

Wilhelm August Oscar Hertwig (1849-1922)
A Professor of anatomy, in 1876 and 1877 studied reproduction in the sea urchin, *Paracentrotus lividus,* and concluded that fertilization involves the union of sperm and egg. This study initiated the period of experimental cytology.

Walter Flemming (1843-1915)
An Austrian cytologist, in 1882 proposed the term mitosis. He showed that the chromosomes split longitudinally during nuclear division and the formation of daughter nuclei. He also applied the name **chromatin** to the stainable portion of the nucleus. He was a distinguished observer, technician, and teacher.

August Weismann (1834-1914)
A German biologist, in his essays of 1883 and 1885 put forth his **germplasm theory**, which was an alternative explanation to Lamarck's theory of acquired characteristics. Weismann speculated that the chromosomes of the sex cells were the carriers of his germsplasm, but he erred in assuming that each chromosome could contain all hereditary material. He also postulated that a periodic reduction in chromosome number must occur in all sexual organisms and that during fertilization a new combination of chromosomes and hereditary factors takes place. His theory was that the alternation of reduction and fertilization is necessary for maintaining constant chromosome numbers for sexual reproduction. At that time this process had not been observed under the microscope, and its mechanism was a matter of speculation.

Wilhelm Roux (1850-1924)
A German zoologist, in 1883 proposed that it was the chromosomes that contain the units of heredity. He speculated on the question of how the hereditary units could behave in such a way that each daughter cell receives all that is in the parent cell and becomes a complete cell and not half a cell or only part of a parent cell.

The most likely constituents of the nucleus to fill these requirements were the chromosomes. His hypothesis was that not only the chromosomes but individual parts of each chromosome were important in determining the individual's development, physiology, and morphology. Proof of this hypothesis was not given until later. This was in direct contrast to Weismann's idea, that each chromosome could contain all hereditary material.

Edouard van Beneden (1845-1910)
He showed that in the round worm, *Ascaris megalocephala*, the number of chromosomes in the games is half the number that is in the body cells, and that in fertilization, the chromosome contributions of egg and sperm to the zygote are numerically equal. Through this observation he confirmed Weismann's theory on reduction and fertilization.

Edward Strasburger (1844-1912)
Strasburger demonstrated that the principles of fertilization developed by Oscar Hertwig for animals held also for plants.

Strasburger made reciprocal crosses between different plant species and found that the results were similar. Since the egg and sperm were unequal with respect to size and amount of cytoplasm carried, he suggested that the cytoplasm was not responsible for hereditary differences between species. Consequently, he came to the conclusion that the nucleus and its chromosomes are the material basis of hereditary and, at the same time, the material governing development.

Theodor Boveri (1862-1915)
By shaking sea urchin eggs at a critical time in their development, he produced some eggs without nuclei and some with nuclei as usual. Each of these kinds of eggs were fertilized by a normal sperm from another species of sea urchin. Eggs lacking a nucleus produced larvae resembling the species from which the sperms were obtained, but those with nuclei developed into hybrids, showing the characteristics of both species. The cytoplasm in the two kinds of eggs had not been altered and it was therefore presumed that the nucleus and not the cytoplasm was responsible for the transmission not hereditary traits.

With his experiments on the double fertilization of sea urchin eggs, *Toxopneustes* (1902, 1904, 1907), Boveri also contributed to the formulation of the **chromosome theory of inheritance,** which will be discussed later.

Edmund Beecher Wilson (1856-1939)
The beginning of cytogenetics and of the chromosome theory of inheritance were clearly outlined by Wilson's statement that the visible chromomeres on the chromosomes were in all probability much larger than the **ultimate dividing units** and that these units must be capable of assimilation, growth, and division without loss of their specific characteristics.

Walter S. Sutton (1877-1916)
He showed the significance of reduction division and proposed the **chromosome theory of heredity.** He independently recognized a parallelism between the behavior of chromosomes and the Mendelian segregation of genes. His most significant contribution to present-day biology was his theory that the Mendelian laws of inheritance could be applied to chromosomes at the cellular level of living organisms. This is now known as the Boveri-Sutton chromosome theory. He furthermore provided the first conclusive evidence that chromosomes carry the units of inheritance and occur in distinct pairs.

The first paper (1902) contained the earliest detailed demonstration that the somatic chromosomes of the lubber grasshopper, *Brachystola magna*, occur in definite distinctly different pairs of like chromosomes. He knew

of Boveri's first paper (1902) on dispermic eggs). His 1903 paper contains a full elaboration of his hypothesis, including the view that the different chromosome pairs orient at random on the meiotic spindles, thus accounting for the independent segregation of separate pairs of genes seen by Mendel. This cytological basis for genetics theory is also often called the *Sutton-Bovgeri theory of chromosomal inheritance.* From then on cytology and genetics began to have strong effects on each other, and this is generally considered the birth of cytogenetics.

Thomas Hunt Morgan (1866-1945)
He discovered the mutant white eye and consequently sex linkage in *Drosophila.* With this discovery, *Drosophila* genetics had its beginning. Morgan was concerned about the exceptions to Mendel's second law of independent assortment. This law implies that an organism cannot possess more gene pairs than the number of chromosomes in a haploid set, if it is granted that the genes are borne on chromosomes. Within the first decade after the rediscovery of Mendelism, this logical consequence of the theory was sharply contracted by experience.

Cyril Dean Darlington (b.1903)
In an attempt to explain meiosis, he advanced the precocity theory. He assumed that the chromosomes have a tendency to be in a paired state at all times. In mitosis this condition is met in that the chromosomes entering prophase are already double. According to this theory meiotic prophase is assumed to start precociously with chromosomes that have not yet split, and this is held responsible for chromosome pairing.

Darlington said that the chromosomes are in an unsatisfied, or unsaturated, state electrostatically. To become saturated they must pair homologously. When the chromosomes become double in late pachytene, the satisfied state is between system chromatids instead of homologus chromosomes. The paired homologues consequently fall apart and diplotene is initiated. This theory was logically beautiful in superficially explaining the genetic implications of meiosis.

Emil Heitz (b. 1892)
Together with Bauer discovered the importance of the **giant chromosomes** in the salivary gland cells of dipteran insect species as important objects in cytogenetic research. These structures had been discovered prior to this in 1881, but had not been identified as chromosomes. They represent bundles of chromosome subunits or chromatids.

In 1928 and 1929 Heitz was the first to distinguish two types of chromatin, which he named **euchromatin** and **heterochromatin**. Euchromatin stains lightly or not at all in interphase and prophase, while

heterochromatin stains darkly in these stages. Heterochromatin is an extremely helpful marker for chromosome mapping in the pachytene stage of meiotic prophase. In 1931 Heitz showed a correlation between the number of nucleoli in the interphase nucleous and the number of a particular type of chromosome, now called the **nucleolus organizer chromosome**. A study of these chromosomes indicated that the nucleolus is organized at a specific site on the chromosome.

Sol Spiegelman (b. 1914)

In 1965 Spiegelman together with Ritossa showed that the genes producing the ribosomal RNA of *Drosophila* are located in the nucleolus organizer regions of the chromosomes. It appears now that the precursor material or ribosomal RNA is manufactured by the nucleolus, or organizer, and is then transferred to the nucleolus for final assembly into ribosomes. These findings are in line with recent research that indicates that living organisms cannot exist without nucleolar organizer chromosomes. Define cytogenetics.

Cytogenetics is a science concerned with the structure, number, function, and movement of chromosomes and the variation of the properties as they relate to the transmission, recombination and expression of the genes.

Self Assessment Exercise

Provide answers to the following questions in 10 minutes

Outline the contributions of the following scientists to the development of cytogenetics as a discipline

(i) Walter Flemming
(ii) (ii) Thomas Morgan
(iii) (iii) Emil Heitz

1.4. Summary

Cytogenetics is a union of cytology and genetics. Several contributors play significant roles in the development of cytogenetics as a discipline. It is defined as a science concerned with the structure, number, function, and movement of chromosomes and the numerous variations of these properties as they relate to the transmission, recombination and expression of the genes.

1.5. References/Further Readings/ Web Sources

Fuchs, J. and Schubert, I. (2012). Chromosomal Distribution and Functional Interpretation of Epigenetic Histone Marks in Plants. In: Bass, H., Birchler, J. (eds) Plant Cytogenetics. Plant Genetics

and Genomics: Crops and Models, vol 4. Springer, New York, NY. https://doi.org/10.1007/978-0-387-70869-0_9

Lamb, J.C., Shakirov, E.V. and Shippen, D.E. (2012). Plant Telomeres. In: Bass, H., Birchler, J. (eds) Plant Cytogenetics. Plant Genetics and Genomics: Crops and Models, vol 4. Springer, New York, NY. https://doi.org/10.1007/978-0-387-70869-0_7

Murphy, S.P. and Bass, H.W. (2012). Genetics and Cytology of Meiotic Chromosome Behavior in Plants. In: Bass, H., Birchler, J. (eds) Plant Cytogenetics. Plant Genetics and Genomics: Crops and Models, vol 4. Springer, New York, NY. https://doi.org/10.1007/978-0-387-70869-0_8

https://www.onlinebiologynotes.com/terminology-used-in-mendelian-genetics%ef%bb%bf/

https://www.onlinebiologynotes.com/physical-behaviour-of-chromosomes-during-meiosis/

https://www.onlinebiologynotes.com/karyotype-and-idiogram/

https://www.onlinebiologynotes.com/mendels-genetics-selection-of-pea-plant-and-reasons-for-mendels-success/

Chromosome Structure and Organization - YouTube

https://youtu.be/nXTbY8hEQY4

https://youtu.be/o_-6JXLYS-k

https://www.youtube.com/watch?v=8m6hHRlKwxY&vl=en

https://www.khanacademy.org/science/ap-biology/heredity/mendelian-genetics-ap/v/introduction-to-heredity

 1.6. Possible Answers to SAEs

SAE

(i) Walter Flemming – He showed that the chromosomes split longitudinally during cell division and first applied the name chromatin.

(ii) Thomas Morgan – He discovered sex linkage working with Drosophila.

(iii) Emil Heitz – He discovered giant chromosomes in the salivary gland cells of diptherian insects.

Unit 2: Chromosome Theory of Inheritance

CONTENTS

 2.1. Introduction

The experiment of Theodore Boveri and other scientists are central to the development of chromosome theory of inheritance. It is necessary to understand these experiments and the line of reasoning of the scientists to fully appreciate how the chromosome theory of inheritance was developed. The chromosomal theory of inheritance was given by Boveri and Sutton in the early 1900s. It is the fundamental theory of genetics. According to this theory, **genes are the units of heredity and are found in the chromosomes**. Chromosomal Theory of Inheritance came into existence long after Mendelian genetics.

 2.2. Learning Outcomes

At the end of the class, student must have fully understood the experiments of Theodore Boveri and another scientist in the development of chromosome theory of inheritance.

 2.3. Theories of Inheritance

2.3.1. Theodore Boveri's Experiment

He performed his experiment using sea urchin eggs. Usually double fertilization does not occur in animals unlike in flowering plants where double fertilization animals, a membrane is formed covering the egg to prevent other sperms from entering the fertilized egg. This is called monospermy. However, an egg may sometimes be fertilized by two sperms, this is called dispermy. With experiments on double fertilization of sea urchin eggs, Boveri contributed significantly to the development of Chromosome Theory of Inheritance.

He found eggs that had been fertilized by two spermatozoa. Since each sperm introduced a centrosome into the egg, and each centrosome divided in anticipation of the first cleavage division, the initial metaphases and anaphases were often characterized by a **tetraster,** which is a spindle with four poles. Since the dividing nucleus was triploid, the distribution of the chromosomes to four poles in anaphase was irregular. Boveri isolated many of the first-division blastomeres from these dispermic eggs and demonstrated that most were abnormal in development, but that all were not alike in their abnormalities. He concluded that abnormal development resulted from the irregular distribution of chromosomes brought on by the multipolar division. Each chromosome must consequently have possessed a certain individual quality that expressed itself in development.

What is monospermy?
Ans. Double fertilization in animals whereby a membrane is formed covering the egg to prevent other sperms from entering the fertilized egg.

2.3.2. Edmund Beecher Wilson's Principle of Chromosome Theory of Inheritance
Wilson's three editions of The Cell in Development and Inheritance (or Heredity) in 1896, 1900, and 1925 introduced generations of students to cell biology. Edmund Beecher Wilson and Nettie Maria Stevens both independently developed the idea of sex determination by chromosomes. Their work established the importance of chromosomes in heredity and helped Thomas Hunt Morgan interpret the early genetic results from Drosophila.

Four principles were laid down by Wilson as the foundation of the chromosome Theory of Inheritance namely:

1. The exact lengthwise division of the chromosomes at mitosis allows for the equal distribution of linearly arranged particles to the daughter cells.
2. The assumed material existence of the chromosomes in the nucleus between mitoses gives the genetic continuity necessary for the organs of heredity.
3. The fact that the nucleus goes where things are happening shows its governing position in the work of the cell.
4. The quality of the chromosomes of the fusing germ cells corresponds to the equality of male and female in heredity.

These arguments had long been known but were still widely disputed or misunderstand at this time.
What did Edmund Beecher Wilson's Principle of chromosome theory of inheritance establish?

The theory established the importance of chromosome in heredity

2.3.3. Thomas Hunt Morgan's experiments on *Drosophila melanogaster*

Morgan's experiments on *Dorsophila melanogaster* **to explain chromosomal theory of inheritance**

- In 1910, **Thomas Hunt Morgan** performed experiment on *Drosophila melanogaster*
- *Dorsophila melanogaster* is commonly known as a fruit fly. It has only four pair of chromosomes (three pair of autosomes and a pair of sex chromosome). At the time of experiment, it was established that X and Y chromosomes are associated with gender. This experiment of Morgan establish that gene for eye color in Drosophila is on X-chromosome and it is the first X-linked trait to be identified.

I. Cross between Red eyed female (Xw+ Xw+) and white eyed male (Xw Y)

- In *Drosophila*, normal flies have red eyes and mutated flies have white eyes.
- In the experiment, normal flies with red eyes and mutated flies with white eyes are crossed and offspring are observed.
- Dominant allele: Red eye color.
- Recessive allele (mutated): white eye
- When **a red eyed female (Xw+ Xw+)** and **white eyed male (Xw Y)** were mated, all the progeny in F_1 generation had **red eyes**
- According to Dominant and recessive inheritance pattern, this result makes perfect sense.

II. Reciprocal cross- red eyed male (Xw+ Y) and white eyed female (Xw Xw)

- A surprising result was obtained when the reciprocal cross was performed by mating white eyed females to red eyed males.
- In F1 generation, instead of obtaining all the progeny with red eye, the result showed that all the progeny females had red eyes and all the progeny males had white eyes.
- This result shows traits for gender and eye color are linked, which is not accordance with Mendel's law of independent assortment.
- This result can be explained only if gene for color of eye is present on X chromosome and are linked.
- Thus, this experiment on eye color of *Drosophila* supports the chromosome theory of inheritance. Now, all the patterns of inheritance observed by Mendel, including the principle of segregation and the principle of independent assortment can be explained by the behavior of chromosomes during meiosis and chromosome theory of inheritance.

Note: Males are known as Hemizygous because they have only one allele for any X-linked traits. Therefore, a male with the recessive allele will always show the recessive trait, because he only has one copy of the allele.

- We know, most genes are located on autosomes.

What did Morgan's experiment establish?
Morgan's experiment established that gene for eye color in Drosophila is on X-chromosome and it is the first X-linked trait to be identified.

Self-Assessment Exercise 1

Provide concise answer to the following question giving illustrations where necessary in 20 minutes

1. What are the foundations upon which the chromosome theory of inheritance was laid as established by Edmund Beech Wilson?
2. What do you consider as the main idea of the chromosome theory of inheritance?
3. What was Thomas Morgan's contribution to the chromosome theory of inheritance?

2.4. ***NON disjunction as proof of chromosomal theory of Inheritance:***

- Morgan showed that a gene for eye color was on the X chromosome of *Drosophila*
- One of his students, **C. B. Bridges,** who secured proof of the chromosome theory by showing that exceptions to the rules of inheritance could also be explained by chromosome behavior.
- **Bridges** performed one of Morgan's experiments on a larger scale.
- In his experiment**,** white-eyed female *Drosophila* **(Xw Xw)** crossed with red-eyed males **(Xw+ Y)** and examined many F1 progeny.
- In F1 generation, following results are obtained;
- Almost all of the F1 progeny flies are either **red eyed females** or **white eyed males** which are normal
- But few flies such as **white-eyed females** and **red-eyed males** are also obtained which are exceptional.
- When he tried to crossed red eyed F1 progeny males with normal white eyed females, it was found that all the F1 progeny red eyed males were sterile.
- However, the F1 progeny white eyed females were fertile. When these white eyed females were crossed with normal red eyed males, many F2 progeny were obtained as white eyed females and red eyed males.
- It seems, the exceptional F1 progeny females produced large number of exceptional progeny in successive generation.

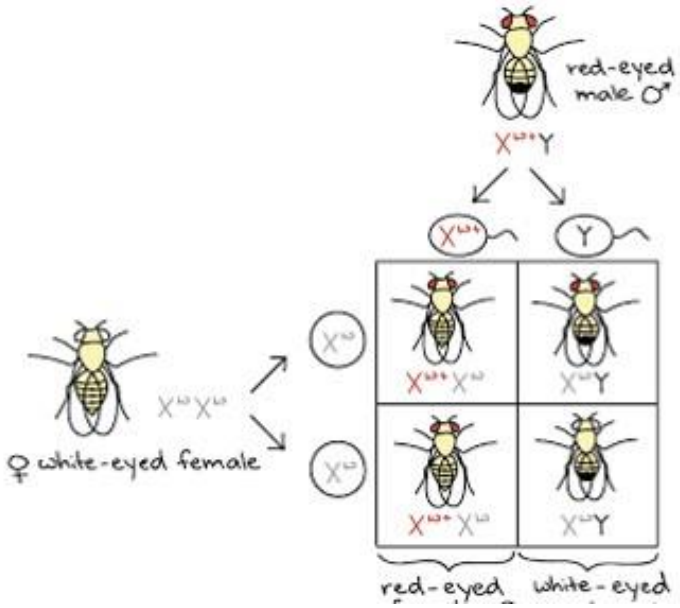

Figure 1. Bridge's **proof of chromosomal theory of Inheritance**

Explanation of result:

- Bridges explained these results by proposing that the exceptional F1 flies were the result of **abnormal X chromosome behavior** during meiosis in the females of the P generation.
- Normally the X chromosome in females separates or disjoin during gametogenesis but Occasionally, they might fail to separate, producing an egg with two X chromosomes (**diplo-X**) or an egg with no X chromosome at all (**nullo-X**) known as **NON-DISJUNCTION** (Figure 1)
- Fertilization of such abnormal eggs by normal sperm would produce zygotes with an abnormal number of sex chromosomes.
- If an egg with two X chromosomes (XwXw) is fertilized by normal Y chromosome of sperm (Y) - the zygote will be **XwXwY.** Since each of the X chromosomes in this zygote carries a mutant Xw allele, the resulting fly will have **white eyes**.
- If an egg without an X chromosome (O) is fertilized by an X-bearing sperm (Xw+), the zygote will be Xw+ O. (O- denotes absence of a chromosome.) Because the single Xw+ in this zygote carries mutant Xw+ allele, the zygote will develop into a **red-eyed fly**
- It is inferred that **XXY flies were female** and that **XO flies were male.** The exceptional F1 **white-eyed females** that were observed were therefore **XwXwY,** and the exceptional F1 **red-eyed males** were **Xw+O.**
- Bridges confirmed the chromosome constitutions of these exceptional flies by direct cytological observation.
- Because the XO animals were male, Bridges concluded that in ***Drosophila*** the Y chromosome has nothing to do with the determination of the sexual phenotype. However, because the XO

males were always sterile, he realized that this chromosome must be important for male sexual function.

- Bridges called the abnormally **nondisjunctio**n because it involved a failure of the chromosomes to disjoin during meiotic divisions.

2.4.1. Causes of nondisjunction:

i. faulty chromosome movement
   a. imprecise or incomplete homologous pairing,
   b. centromere malfunction.
ii. From Bridges experiment, in F2 generation, exceptional **XwXwY** females produces high frequency of exceptional progeny, it means, the **nondisjunction** of sex chromosome goes on **disjunction.**
iii. **XXY Sex chromosomes can disjoin in different ways:**
iv. Either X chromosomes can disjoin from each other form to form: X and XY
v. OR X chromosome can disjoin from the Y to form: XX and Y: In the latter case, a diplo- or nullo-X egg is produced because the X that does not disjoin from the Y is free to move to either pole during the first meiotic division. When fertilized by normal sperm, these abnormal eggs will produce exceptional zygotes.
vi. These early studies with *Drosophila*—primarily the work of Morgan and Bridges greatly strengthened the view that all genes were located on chromosomes. Thus chromosome are the genetic material for inheritance.

What did Morgan and Bridge's works on *Drosophila melanogester* establish?

Morgan and Bridge's experiments established that gene for eye color in Drosophila is on X-chromosome and it is the first X-linked trait to be identified. Their works greatly strengthened the view that all genes are located on chromosomes and that they are the genetic material for inheritance.

Self-Assessment Exercise 2

Provide concise answer to the following question giving illustrations where necessary in 20 minutes

1. What are the causes of Nondisjunction of chromosomes?
2. What is non disjunction as it relates to chromosome theories of inheritance?

2.5. Summary

Sutton and Boveri were credited with initiating the chromosome theory of inheritance, the idea that the genetic material in living organisms are contained in the chromosomes. Work by others like Thomas H. Morgan,

Alfred H. Sturtevant, calvin Bridges and other workers established beyond a reasonable doubt that Sutton's and Boveri's hypothesis was correct.

2.6. References/Further readings/Web Sources

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2.7. Possible Answers to Self-Assessment Exercises

SAE 1

1. Four principles were laid down by Wilson as the foundation of the chromosome Theory of Inheritance namely:

a. The exact lengthwise division of the chromosomes at mitosis allows for the equal distribution of linearly arranged particles to the daughter cells.
b. The assumed material existence of the chromosomes in the nucleus between mitoses gives the genetic continuity necessary for the organs of heredity.
c. The fact that the nucleus goes where things are happening shows its governing position in the work of the cell.
d. The quality of the chromosomes of the fusing germ cells corresponds to the equality of male and female in heredity.

2. The main idea of the theory is that genes are arranged linearly on the chromosomes.

3. He discovered sex-linkage. By this discovery he was able to show that sex- linked characters are present on X chromosome thereby contributing to the development of chromosome theory of inheritance.

SAE 2

1. Causes of nondisjunction:

i. faulty chromosome movement
   a. imprecise or incomplete homologous pairing,
   b. centromere malfunction.

ii. From Bridges experiment, in F2 generation, exceptional **XwXwY** females produces high frequency of exceptional progeny, it means, the **nondisjunction** of sex chromosome goes on **disjunction.**

iii. **XXY Sex chromosomes can disjoin in different ways:**

iv. Either X chromosomes can disjoin from each other form to form: X and XY

v. OR X chromosome can disjoin from the Y to form: XX and Y: In the latter case, a diplo- or nullo-X egg is produced because the X that does not disjoin from the Y is free to move to either pole during the first meiotic division. When fertilized by normal sperm, these abnormal eggs will produce exceptional zygotes.

vi. These early studies with *Drosophila*—primarily the work of Morgan and Bridges greatly strengthened the view that all genes were located on chromosomes. Thus chromosome are the genetic material for inheritance.

2. Nondisjunction is the failure of the chromosomes to separate, which produces daughter cells with abnormal numbers of chromosomes. Simply put, it is the failure of homologous chromosomes or sister chromatids to separate properly during cell division

Unit 3: Chromosome Packaging

CONTENTS

 3.1. Introduction

Chromosomal DNA is packaged inside microscopic nuclei with the help of histones. These are positively-charged proteins that strongly adhere to negatively-charged DNA and form complexes called nucleosomes. Each nucleosome is composed of DNA wound 1.65 times around eight histone proteins. The importance of the organization of DNA into chromatin and of chromatin into mitotic chromosomes can be illustrated by considering that a human cell stores its genetic material in a nucleus about 5 to 10 μm in diameter.

In the overall transition from a fully extended DNA helix to the extremely condensed status of the mitotic chromosome, a packing ratio (the ratio of DNA length to the length of the structure containing it) of about 500 to 1 must be achieved.

 3.2. Learning Outcomes

To understand how DNA is organize into chromatin and how chromatin is in turn organized into metaphase chromosome by repeated coiling.

 3.3. Chromosome Packaging

3.3.1. First Level Packaging

During interphase, chromosomes exist as chromatin fibers composed of DNA and proteins (positively charged histones and less positively charged nonhistones). The chromatin fibers resemble beads (~100A0 diameter) on a string (~20A0 diameter). The string is DNA while the bead is 147 bp length of DNA coiled 1.7 turns round a core composed of 8 histones (octamer) – two each of H2A, H2B, H3 and H4. The fifth type of histone, H1 lies outside the core.

3.3.2. Second Level Packaging

Further packaging of several (6) $100A^0$-nucleosomes gives a solenoid of about 300A0. This further reduces the chromosomes length by 1/6.

3.3.3. Third Level Packaging

In transition to mitosis, the 300A0 structure forms a series of looped domains of about 3,000A0 in diameter.

3.3.4. Fourth Level Packaging

The condensed fibers are further coiled into chromosome arms (of about 7000A0) that constitute a chromatid seen at mitotic metaphase. At metaphase, the chromosomes are at their highest level of coiling and therefore appear more condensed, shorter and thicker than in any other stage. This makes chromosomes most ideal for cytological study at metaphase because, they are most sharply defined at this stage (Figures 2 and 3)

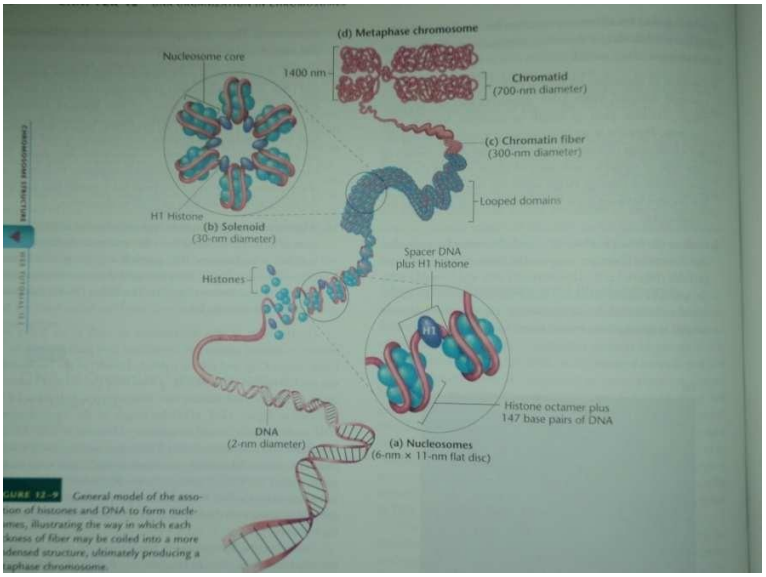

Figure 2. Successive Levels of Chromosome Packaging

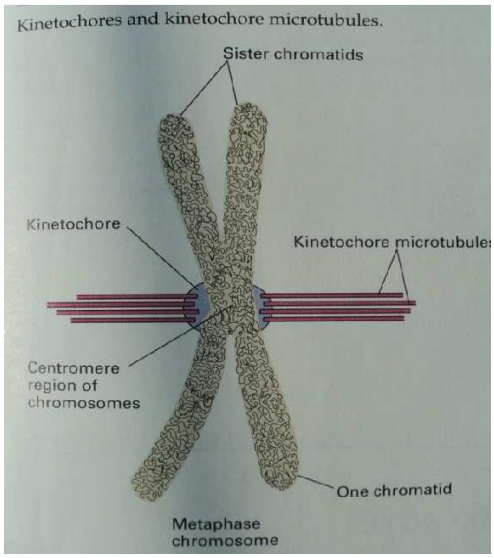

Figure 3. A Metaphase Chromosomes

Why do we need chromosome packaging?
The entire DNA strand must fit within the nucleus of a cell, so it must be very tightly packaged to fit. This is accomplished by wrapping the DNA around structural histone proteins, which act as scaffolding for the DNA to be coiled around.

Self-Assessment Exercise

Provide answer to the following question in 20 minutes

1. What is chromosome packaging?
2. What do you understand by DNA packaging?
3. What is the importance of DNA packaging?
4. How is DNA organized into chromatin?

3.4. Summary

Eukaryotic chromatin is a nucleoprotein organized into repeating units called nucleosomes. Composed of 200 base pairs of DNA, an octamer of four types of histones, plus one linker histone, the nucleosome is important for condensing the extensive chromatin fiber within the interphase nucleus into the highly condensed chromosome seen in mitosis.

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 3.6. Possible Answers to Self-Assessment Exercises SAE

1. Chromosomal DNA is packaged inside microscopic nuclei with the help of histones. These are positively-charged proteins that strongly adhere to negatively-charged DNA and form complexes called nucleosomes. Each nuclesome is composed of DNA wound 1.65 times around eight histone proteins.

2. DNA packaging is the folding of an organism's DNA into a compact structure that can fit within the nucleus of a cell.

3. DNA packaging is important because the DNA is very long. In order to fit the DNA into the nucleus, it needs to be packaged properly.

4. DNA is negatively charged; it is therefore found associated with positively charged histones and other non-histone proteins to form the chromatin

Unit 4: Chromosome Morphology

CONTENTS

 4.1. Introduction

Chromosome morphology deals with chromosome structure. This has extensive applications in identifying chromosomal aberrations, genetic diseases such as Down's syndrome, klinefelter's syndrome and other genetic diseases.

 4.2. Learning Outcomes

At the end of the class, student must be familiar and have understood chromosome morphology in terms of centromere location, chromosome sizes etc.

 4.3. The Centromere

Each centromere contains a constricted region (primary constriction). Centromeric region is heterochromatic. A region of chromosome is described as heterochromatic if the region is made up of heterochromatin. As we shall see later, heterochromatin are made up of inactive DNA.

4.3.1. Functions of the Centromere

*Maintenance of cohesion of sister chromatids: sister chromatids remain attached at the centromere before anaphase.

* Mediation of chromosome movement during anaphase: The microtubules of the spindle fibres are attached to the kinetochore of the

proteinaceous platform that attaches to the microtubules of spindle fibers during anaphase.

The critical region that supports the second function above is the CEN region of the centromere. In yeast, the CEN region consists of 100 bp of repetitive sequences divided into 3 regions. Mutational analysis revealed that Regions I and II affect segregational activity, but mutation in the central CCG triplet within region III completely inactivates centromere function.

What are two functions of a centromere?
The main functions include the attachment of sister chromatids, and it is the site for the attachment of spindle fibre. Centromeres help in the proper alignment and segregation of the chromosomes during the process of cell division in eukaryotic cells.

Self-Assessment Exercise 1

Provide answer to the following question in 5 minutes

What are chromosomes explain the morphology and classification of chromosomes in detail?

4.4. Chromosome classification based on the number of centromeres

4.4.1. Acentric Chromosomes

This refers to chromosomes that lack a centromere. In fact such chromosomes do not exist in nature in view of the crucial functions of centromeres in chromosome behaviour. They are formed as a result of structural aberrations. Such chromosomes are more appropriately referred to as acentric fragments.

What is meant by acentric chromosome?
A fragment of a chromosome (one of the microscopically visible carriers of the genetic material DNA) that is lacking a centromere (the "waist" of the chromosome essential for the division and the retention of the chromosome in the cell) and so is lost when the cell divides.

4.4.2. Monocentric Chromosomes

Chromosomes having one centromere per chromosome are known as monocentrics. This is the usual case. Thus, normal chromosomes are usually monocentric.
What is meant by monocentric chromosome?
Chromosomes with a single centromere are termed as monocentric chromosomes.

4.4.3. Dicentric Chromosomes

These are chromosomes having two centromeres per chromosome. They arise from single chromosomal breaks to give a dicentric and acentric fragments. The acentric fragment is not incorporated into any daughter cells and is lost. The dicentric fragment has centromeres on 2 sister chromatids, it forms an anaphase bridge. What is meant by Dicentric chromosome?

chromosomes having two centromeres per chromosome are termed as dicentric chromosomes.

Self-Assessment Exercise 2

Provide answer to the following question in 10 minutes

i. What causes dicentric chromosomes?

ii. What is acentric chromosome? give an example.

4.5. Chromosome Classification Based on Centromere Location

4.5.1. Centromere Located in the Middle of the Chromosome

Such centromeres are referred to as median centromeres and the chromosome is described as the metacentric chromosome.

4.5.2. Centromere Located near the Middle of the Chromosome

This is referred to as sub-median centromere, and the chromosome is called a submetacentric chromosome. What type of chromosome is located on the centromere?

Metacentric chromosome: The chromosomes in which the centromere is present in the middle divide the chromosome into two equal arms.

4.5.3 Centromere Located near the end of the Chromosome

Such centromeres are referred to as sub-terminal centromere to give acrocentric chromosome.

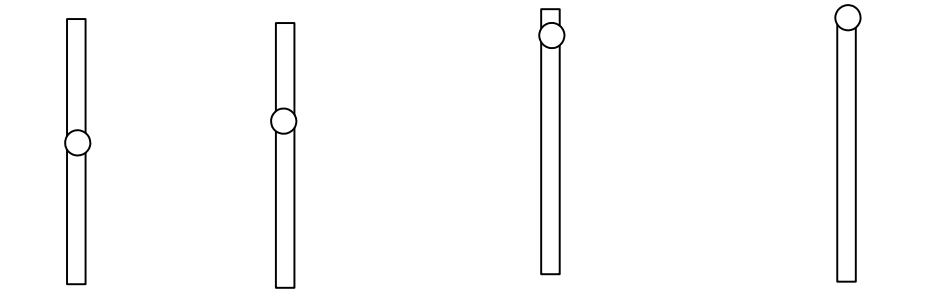
What are acrocentric chromosomes?

A chromosome where the centromere is not central and is instead located near the end of the chromosome.

4.5.4 Centromere Located at the end of the Chromosome

Such centromeres are refer to as terminal centromere. And the chromosomes are described as telocentric.

Telocentric chromosomes may arise by centromere misdivision or breakage induced within the centromere. Telocentrics are unstable because their formation involves fracturing of the centromere (Figure 4). Thus, they are rare in nature



Metacentric Chromosome | Sub-metacentric chromosome | Sub-eloncentric chromosome | Talocentric chromosome

Figure 4. Different types and locations of Chromosomes

What is the position of centromere in Metacentric chromosome?
Metacentric chromosomes have the centromere located midway between the ends of the chromosome, separating the two arms of the chromosome.

Self-Assessment Exercise 3

Provide answer to the following question in 20 minutes

What determines position of centromere?

4.6. Shapes of Chromosomes during Anaphase
During anaphase chromosome moves to the opposite poles. The spindle fibres originating from the poles are attached to the chromosomes at their centromeres. Thus, the chromosomes assume characteristics shapes because the centromere is in the lead while the arms trail behind.

Metacentric : V-shape
Acrocentric: rod shape
Sum-metacentric: J-shape

What are the different morphological types of chromosomes?
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 position on the chromosomes respectively.

Self-Assessment Exercise 4

Provide answer to the following question in 5 minutes

Complete the Table

Centromere location	Chromosome type	Shape at anaphase
terminal	(i)	(ii)
(iii)	metacentric	(iv)
(v)	acrocentric	(vi)

4.7. Summary

Chromosomes can be classified based on centromere location and size. Based on centromere location, we may have metacentric, sub-metacentric, sub- telocentric and telocentric chromosomes. Existence of telocentric chromosomes is still being debated.

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4.8. Possible Answers to Self-Assessment Exercises

SAE 1

A chromosome is a long DNA molecule with part or all of the genetic material of an organism. In most chromosomes the very long thin DNA fibers are coated with packaging proteins; in eukaryotic cells the most important of these proteins are the histones.

SAE 2

i. Dicentric chromosomes are formed by the fusion of two chromosome ends, which then initiates an ongoing chromosomal instability via breakage-fusion-bridge cycles (BFB).

ii. Acentric chromosome: A fragment of a chromosome (one of the microscopically visible carriers of the genetic material DNA) that is lacking a centromere (the "waist" of the chromosome essential for the division and the retention of the chromosome in the cell) and so is lost when the cell divides.

SAE 3

DNA sequences are both necessary and sufficient to specify centromere identity and function in organisms with point centromeres. In budding yeasts, the centromere region is relatively small (about 125 bp DNA) and contains two highly conserved DNA sequences that serve as binding sites for essential kinetochore proteins.

SAE 4

Centromere location	Chromosome type	Shape at anaphase
Terminal	(i) telocentric	(ii) rod shaped
(iii) median	metacentric	(iv) v-shape
(v) sub-terminal	acrocentric	(vi) rod-shaped

Unit 5: Chromosome Classification Based On Size and Other Attributes

CONTENTS

 5.1. Introduction

The most important parameter for describing chromosome is the centromere location. However, the size of chromosome is also very important in their morphological description. Therefore, it is generally agreed that the two most important parameters for morphological description of chromosomes are the centromere location and the size of the chromosomes.

 5.2. Learning Outcomes

At the end of the class, student must be familiar and have understood the following:

- types of chromosome based on size, presence of secondary constriction;
- the functions of satellite chromosomes; and
- what heterochromatin and euchromatin are.

 5.3. Size of Chromosomes

Chromosomes can be qualitatively described as long, short or medium. Mitotic metaphase chromosomes usually range from about 0.5 µm to 30 µm in length and from 0.2 µm to 3.0 µm. On the average, plants have larger chromosomes than animals.

Cytogeneticists use karyotypes and ideograms to demonstrate such characteristics. The total chromosomal complement of a cell is refer to as the karyotype (Figure 5). The complement can be photographed during mitosis and rearranged in pairs to produce a picture refer to as karyotype. An ideogram is a diagrammatic representation of the gametic chromosome set (n) of a given species. The longest chromosomes occur

in plant genus *Trillium a*nd are longer than 30 µm. The shortest chromosomes are less than 1.0 µm.

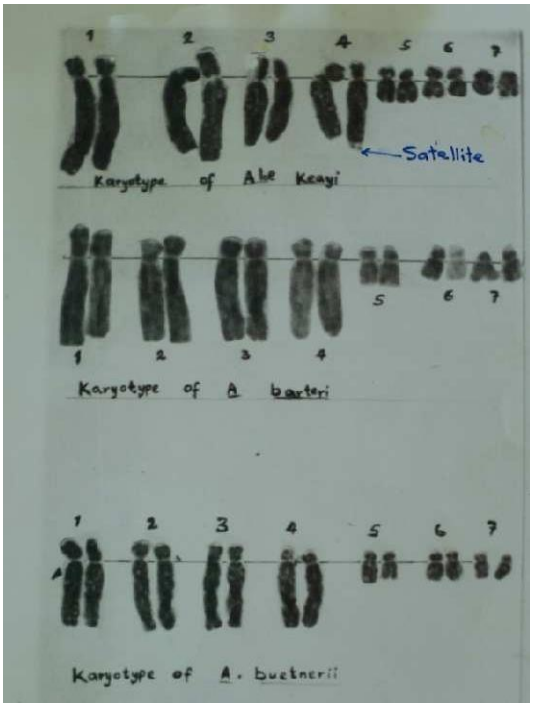

Figure 5: Karyotypes

Which is the largest size chromosome?
Chromosome 1 is the largest human chromosome, spanning about 249 million DNA building blocks (base pairs) and representing approximately 8 percent of the total DNA in cells.

Self-Assessment Exercise 1

Provide answer to the following question in 10 minutes

What is the size of chromosomal DNA?

5.4. Satellite Chromosomes

These are also refer to as nucleolar organizer chromosomes (NOC). These are chromosome that have secondary constriction in addition to the primary constriction, the centromere, that we have dealt with before. The region of secondary constriction is referred to as nucleolar organizer region (NOR). The region is so called because nucleolus is found associated with the region during interphase and prophase, and it is responsible for the formation of the nucleolus during telophase. Thus, NOR is actually not a constriction, but it is negatively heteropycnotic such that the remaining portion of the chromosome appears removed from the

chromosome like a fragment: the portion that appears removed is called satellite (Figure 6). Thus NOC can also be called satellite chromosome. Large satellites can possess a separate constriction and are called tandem satellite.

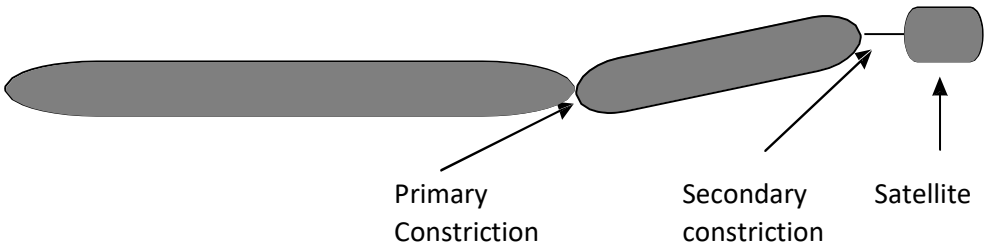

Figure 6: Satellite Chromosome

5.4.1. **Molecular Explanation for the Function of NOR**

NOR contains genes that are responsible for rRNA production. After production, rRNA are transferred to the nucleolus for final assembly into ribosomes.

What is satellite chromosome function?

Satellite DNA contributes to the essential processes of formation of crucial chromosome structures, heterochromatin establishment, dosage compensation, reproductive isolation, genome stability and development.

Self-Assessment Exercise 2

Provide answer to the following question in 10 minutes

What is the difference between satellite DNA and satellite chromosome?

5.5. Euchromatin and Heterochromatin

Staining the interphase nucleus by various chemical dyes reveals a network of nucleoprotein material called the chromatin. These structures are organized into chromosomes during nuclear division. Chromatin possesses differential staining properties. Those that stain very darkly are called heterochromatin while those that stain relatively lightly are called euchromatin. Heterochromatin is considered to be genetically inactive while euchromatin is associated with intense genetic activity before the term heterochromatin was coined, Montgomery (1904, 1906) and Gutherz (1907) described the concept of heteropycnosis whereby some chromosomes or chromosome regions are out of phase in respect to their coiling cycle and staining properties. These chromosomes or chromosome regions were later described as heterochromatic. Costitutive

heterochromatin usually does not change its nature. It is found at the proximal to centromere, telomere, and in the NOR and satellites. Facultative heterochromatinis euchromatin that has been heterochromatized.
Distinguish between heterochromatin and euchromatin.
Heterochromatin stains darkly, and they are genetically inactive while euchromatin stains lightly and are genetically active.

Self-Assessment Exercise 3

Provide answer to the following question in 20 minutes

i. What is difference between heterochromatin and euchromatin?

ii. What is the color of heterochromatin?

5.6. Telomeres

Telomeres are present at the ends of chromosomes. They act like caps preventing chromosome ends from joining. When chromosome breaks, absence of telomeres at the broken ends can make chromosome to join each other and cause aberrations. Moreover, telomere prevents chromosome shortening after replication.
Functions of the Telomere

i. Prevention of joining of chromosomes
ii. Prevention of digestion of chromosome ends from digestion by enzymes that can digest double-stranded chromosome ends.

What shortens telomeres?
Telomere length shortens with age. Rate of telomere shortening may indicate the pace of aging. Lifestyle factors such as smoking, lack of physical activity, obesity, stress, exposure to pollution, etc. can potentially increase the rate of telomere shortening, cancer risk, and pace of aging.

Self-Assessment Exercise 4

Provide answer to the following question in 10 minutes

What are telomeres? What are their cytogenetic importance?

5.7 Summary

Chromosomes can be classified as long, medium and small based on their sizes. Apart from the centromere (primary constriction) present on chromosomes, some also have a region of secondary chromosomes. Such

chromosomes are called satellite chromosomes. They are important in ribosome production. Heterochromatin are genetically inactive regions while euchromatin are active regions.

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 5.9. Possible Answers to Self-Assessment Exercises

SAE 1

The male nuclear diploid genome extends for 6.27 Gigabase pairs (Gbp), is 205.00 cm (cm) long and weighs 6.41 picograms (pg). Female values are 6.37 Gbp, 208.23 cm, 6.51 pg. The individual variability and the implication for the DNA informational density in terms of bits/volume were discussed.

SAE 2

Satellite DNA is located at very specific spots in the genome. Satellite DNA is highly repetitive DNA sequences found in heterochromatin. Satellites of chromosomes have repetitive, heterochromatic DNA sequences. During the replication of the chromosomes the ends of chromosomal sequences are truncated.

SAE 3

i. Heterochromatin is defined as the area of the chromosome which is darkly stained with a DNA-specific stain and is in comparatively condensed form. Euchromatin is defined as the area of the chromosome which is rich in genes that actively participate in the transcription process.

ii. DNA can be divided into euchromatin and heterochromatin. Shown is a representative acrocentric chromosome containing both condensed heterochromatic (dark gray) and less condensed euchromatic regions

(light gray). Beside each region are characteristics typical for each type of chromatin.

SAE 4

Telomeres are special DNA sequences present at the ends of chromosomes. They act like caps for chromosomes.

Importance of telomeres:

i. They prevent one chromosome joining the other.
ii. They help proper replication of chromosome ends in order to prevent shortening of chromosomes due to replication.

Glossary

BFB - breakage-fusion-bridge cycles
bp – Basepairs
CCG - cytosine-cytosine-guanine region
CEN – centromere
Cm - centimetre
DNA – Deoxyribonucleic acid
F1 – First Filial Generation
F2 – Second Filial Generation
Gbp - Gigabase
H1 – Histone variant 1
H2A – Histone variant 2A
H2B – Histone variant 2B
H3 – Histone variant 3
H4 – Histone variant 4
NOC - nucleolar organizer chromosomes
NOR - nucleolar organizer region
pg - picograms
RNA - Ribonucleic acid
rRNA – Ribosomal Ribonucleic acid
µm – micrometer

End of Module Questions

What are the differences among monocetric, dicentric and polycentric chromosomes?

Answer:

Chromosomes are thread-like structures that are packed with long DNA molecules, along with histone and nonhistone proteins inside a nucleus. Each chromosome is composed of two chromatid sisters that are joined by a centromere. Centromeres are important structures on a chromosome. They are the sites of attachment of kinetochores that assemble microtubules and spindle fibres for the process of cellular division.

The chromosomes are of three different types, based on the number of centromeres they possess. The three types are monocentric, dicentric, and polycentric chromosomes.

Monocentric Chromosomes

The chromosomes that have only one centromere joining the two sister chromatids are known as monocentric chromosomes. They occur commonly in all types of plants and animals.

Based on the position of the centromere, they can be of different types such as acrocentric, telocentric, submetacentric, and metacentric chromosomes.

Dicentric Chromosomes

Dicentric chromosomes are the ones that have two centromeres in their chromosomes. Translocation and paracentric inversion are two methods that are known to form dicentric chromosomes. When two chromatid segments, each with a centromere attach with each other, they form dicentric chromosomes.

Dicentric chromosomes can be used clinically to study abnormalities in human genomes. They can be used as biomarkers to study genetic syndromes. They also find applications in cytogenetics.

Polycentric Chromosomes

A chromosome featuring multiple centromeres is called polycentric chromosome. They are produced as a result of a chromosomal anomaly such as translocation, deletion or duplication. Cells with such a type of chromosome tend to die because they cannot separate at the anaphase stage. The chromosomes are fragmented, and as a result, the cell dies.

However, polycentric chromosomes are a common occurrence in certain algal species.

Monocentric vs Dicentric vs Polycentric Chromosomes

Monocentric Chromosomes	**Dicentric Chromosomes**	**Polycentric Chromosomes**
Description		
Chromosomes with a single centromere are termed as monocentric chromosomes.	Chromosomes with two centromeres are termed dicentric chromosomes.	Chromosomes with multiple centromeres are referred to as polycentric chromosomes.
Formation		
They exist naturally in the cells.	They are formed as a result of translocation and paracentric inversion.	They are formed as a result of deletion, duplication and translocation.
Number of Centromeres		
1	2	Multiple
Found in		
They are commonly found in plant and animal cells.	They are rarely found in plant and animal cells.	They are a common occurrence in algal species.
Clinical Significance		
The centromere acts as a site for the assembly of cell division machinery.	It can be used to study genetic abnormalities in the genome.	No such significance, because the cells tend to die.

Module 2

Unit 1	Variation in Chromosome Number: An Overview
Unit 2	Monoploidy
Unit 3	Diploidy and Introduction to polyploidy
Unit 4	Triploidy
Unit 5	Tetraploidy and Higher Polyploidy

Unit 1: Variation in Chromosome Number: An Overview

CONTENTS

 1.1 Introduction

In this module, phenotypic variation that results from changes that are more substantial than alterations of individual genes are considered. Such alterations cause modifications at the level of the chromosome.

Most members of diploid species normally contain precisely two haploid chromosome sets, many known cases vary from this pattern. Modifications that affect changes in the number of chromosomes as opposed to those that affect the structure of chromosomes will be considered in this module.

Changes in chromosome number can occur by the addition of all or part of a chromosome (aneuploidy), the loss of an entire set of chromosomes (monoploidy) or the gain of one or more complete sets of chromosomes (euploidy). Each of these conditions is a variation on the normal diploid number of chromosomes.

 1.2. Learning Outcomes

At the end of the class, student must be familiar and have understood nature and consequence of variation in chromosome numbers.

1.3. Aneuploidy and Euploidy

In aneuploidy, an organism has gained or lost one or more chromosomes but not a complete set. The absence of a single chromosome from an otherwise diploid genome is called *monosomy.* The gain of one extra chromosome results in *trisomy.* Such changes are contrasted with the condition of **euploidy,** where all chromosomes belong to complete haploid sets. If more than two sets are present, the term **polypoloidy** applies. Organisms with three sets are specifically triploid; those with four sets are *tetraploid.* Table 1 provides an organizational framework for you to follow.

Table 1. Terminology of Variation in Chromosome Numbers

Term	Explanation
Aneuploidy	2n ± x; chromosomes
Monosomy	2n – 1
Disomy	2n
Trisomy, Pentasomy, e.t.c.	2n + 1
Euploidy	2n + 2, 2n + 3, e.t.c.
Diploidy	2n
Polyploidy	3n, 4n, 5n
Tetraploidy, Pentaploidy e.t.c.	3n
Autopolyploidy	Multiples of the same genome
Allopolyploidy (Amphidiploidy)	Multiples of closely related genomes

What causes chromosome variation?

Abnormal chromosomes most often happen as a result of an error during cell division. Chromosome abnormalities often happen due to 1 or more of these: Errors during dividing of sex cells (meiosis) Errors during dividing of other cells (mitosis)

Self-Assessment Exercises 1

Provide answer to the following question in 10 minutes

i. What is the difference between aneuploidy and polyploidy?

ii. Is Turner syndrome aneuploidy?

1.4. Origin of Variations in Chromosome Number

As cases that result from the gain or loss of chromosomes, it is useful to examine how such aberrations operate. Such chromosomal variation originates as a random error during the production of gametes, a phenomenon referred to as **nondisjunction**, whereby paired homologs fail to disjoin during segregation. This process disrupts the normal distribution of chromosomes into gametes. The results of nondisjunction during meiosis I and meiosis II for a single chromosome of a diploid organism, abnormal games can form containing either two members of the affected chromosome or none at all. Fertilizing these with a normal haploid gamete produces a zygote with either three members (trisomy) or only one member (monosomy) of this chromosome. Nondisjunction leads to a variety of aneuploid conditions in plants and human and other organisms.

How do variations in chromosome structure arise?

Structural Abnormalities: A chromosome's structure can be altered in several ways. Deletions: A portion of the chromosome is missing or deleted. Duplications: A portion of the chromosome is duplicated, resulting in extra genetic material. Translocations: A portion of one chromosome is transferred to another chromosome. Changes in chromosome number can occur by the addition of all or part of a chromosome (aneuploidy), the loss of an entire set of chromosomes (monoploidy) or the gain of one or more complete sets of chromosomes (euploidy). Each of these conditions is a variation on the normal diploid number of chromosomes.

Self-Assessment Exercises 2

Provide answer to the following question in 10 minutes

What are sources of variation

1.5. Concept of the Basic Chromosome Number

The number of chromosomes in a basic set is called the monoploid number (x). The haploid number (n) refers to the number of chromosomes in gametes. In most plants and animals haploid number and monoploid number are the same so n = x or 2n = 2x. However, in some plants like the hexaploid wheat 2n = 6x = 42. Hence n = 21, but x = 7.

What is the importance of chromosome number?

Having the correct number of chromosomes is critically important to having a successful pregnancy. If your embryo does not have the correct number of chromosomes then your baby may fail to develop properly.

Self-Assessment Exercises 3

Provide answer to the following question in 10 minutes

i. What determine number of chromosomes?
ii. What is chromosome and its function?

1.6. Genome Formula

This is a way of representing the chromosome complement of an organisms using upper case letters. Thus wheat (6x) is represented as AABBCC-a hybrid of 3 different species. Unlike another hexaploid which can be AAAAAA, a hexaploid of the same genome.

How do you calculate genome?

Calculation of the genome sizes: The genome size, i.e. the number of base pairs per genome, is given by $\mathbf{\Gamma = C \times N_A \times M_{Bp}^{-1}}$, where N_A is Avogadro's number (6.022×10^{23} mol^{-1}) and M_{Bp} is the mean molar mass of a base pair (660 g mol^{-1}).

Self-Assessment Exercises 4

Provide answer to the following question in 10 minutes

How do you calculate 1 ug of DNA?

1.7. Summary

There are two broad categories of numerical chromosomal aberration in plants and other organisms: Aneuploidy and Polyploidy. Aneuploidy is when the change does not involve the whole set, if the change affects the whole set, it is known as euploidy.

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 1.8. Possible Answers to Self-Assessment Exercises

SAE 1

i. Aneuploidy is usually defined as the condition which is characterised by having an abnormal number of chromosomes in a haploid set. When a complete new set of chromosomes get added it is called polyploidy. They cause chromosomal disorders which are lethal.

ii. Monosomy is another type of aneuploidy in which there is a missing chromosome. A common monosomy is Turner syndrome, in which a female has a missing or damaged X chromosome.

SAE 2

Mutations, recombinations and deletions all are the sources of genetic variation. A Mutation occurs when a DNA gene is damaged or changed in such a way as to alter the genetic message carried by that gene.

SAE 3

i. For instance, the number of chromosomes is based on how the organism happens to divide up its DNA. Whether the DNA is in 6, 46, or 1260 pieces, it doesn't actually mean there's more information. It just means the information is in many more pieces.

ii. Chromosomes are the highest level of organisation of DNA and proteins. The main function of chromosomes is to carry the DNA and transfer the genetic information from parents to offspring. Chromosomes play an important role during cell division. They protect the DNA from getting tangled and damaged.

SAE 4

To find the DNA yield from its concentration, use the following equation: DNA yield [µg] = DNA concentration [µg/mL] * total sample volume [mL]

Unit 2: Monoploidy

CONTENTS

 2.1. Introduction

Organisms with one chromosome set sometimes arise as variants of diploids; such variants are called monoploids (1x). In some plants like the ferns, monoploid stages are part of the regular life cycle, but other monoploids are spontaneous aberrations. The haploid number (n), which we have been using in your study of genetics refers strictly to the number of chromosomes in gametes.

 2.2. Learning Outcomes

At the end of the class, student must have fully understood

- what monoploids are,
- how monoploids arise, and
- the difference between monoploidy and haploidy.

 2.3. Occurrence of Monoploids

In the plant kingdom, spontaneous monoploids have been found in tomatoes and onion and more recently in coffee, barley, coconut and wheat among other plants.

What is monoploids? What is meant by monoploidy?

The state of being monoploid, that is having one set of the chromosomes. Supplement. Ploidy refers to the number of sets of homologous chromosomes in the genome of a cell or an organism.

Self-Assessment Exercises 1

Provide answers to the following questions in 10 minutes

What is monoploidy mutation?

2.4. Production of Monoploids

2.4.1. Interspecific and Intergeneric Hybridization

Jorgensen (1928) performed the cross *Solanum nigrum* x *S. luteum* and the offspring were *S. nigrum* haploids. Here the embryos developed directly from the egg without fertilization (parthenogenesis).

Normal plant are pollinated with gamma-irradiated pollen making them to lose their viability to fertilize. The unfertilized egg is stimulated to develop parthenogenetically. Pollen can also be treated with chemicals such as iodine blue to achieve similar effect.

2.4.2. Twin seedling

Twin seedling result from polyembryonic seed. Polyembryonic seed can produce diploid-diploid, diploid-haploid or haploid-haploid twins. Twin seedling is controlled by the female genotype.

2.4.3. Anther and Pollen Culture

Pioneering work in *Datura innoxia* showed that culturing anthers can yield haploid plants either by the direct format of embryo-like structures from pollen grains or by the formation of callus and subsequent plant regeneration.

2.4.4. Chromosome Elimination

Kasha and Kao (1970) crossed cultivated barley, *Hordeum vulgare* (2 x = 14) with its wild relative *H. bulbosum* (2x = 14) to give a hybrid. Subsequent mitotic elimination of the *H. bulbosum* chromosomes in the developing embryo gave a haploid.

How are Monoploids formed?

Monoploids show only one essential set of chromosomes and can be formed by an unfertilized egg's spontaneous growth.

Self-Assessment Exercises 2

Provide answers to the following questions in 10 minutes

Why are monoploids useful in plant breeding?

2.5. Meiotic Behaviour in Monoploids

In order to have normal meiosis, there must be two homologous chromosomes present. Monoploids have only one basic genome (x) and

are therefore meiotically irregular. The chromosomes appear mostly as univalents at diakinesis. However, bivalents and multivalent were occasionally observed (intragonic pairing = pairing of chromosome in monohaploids). The pairing mechanismin monohaploids is not yet clear. Rieger (1957) put forward a theory that all chromosomes have certain tendency for pairing. If homologous chromosomes are present, they are prefentially paired. If absent, certain forces unite nonhomologous chromosomes. This agrees with precocity theory of Darlington (1932) that single chromosomes are in an unsaturated state electrostatically, and in order to become saturated, they must pair. Precocity theory is weakened by the fact that chromosoma had been duplicated at the S phase. Meiotic pairing in the monoploids indicated the presence of three or more duplications that involved at least six of the nine chromosomes in the basic genome. Chromosomes of monoploid sugarbeet usually were condense and seldom congregated at metaphase plates.

What is the Behaviour of chromosomes during meiosis?
The typical behavior of chromosomes in meiosis is that homologous pairs synapse, recombine, and then separate at anaphase I. At anaphase II, sister chromatids separate. However, studies of small chromosomes in maize derived from a variety of sources typically have failure of sister chromatid cohesion at anaphase I.

Self-Assessment Exercises 3

Provide answers to the following questions in 10 minutes

Compare the following hypothetical monoploids for their degree of infertility. A: x = 3; B: x= 5.

2.6. Fertility in Monoploids

Fertilization is defined as the process of union of two gametes, eggs and sperm. When mammalian eggs and sperm come into contact in the female oviduct, a series of steps is set in motion that can lead to fertilization and ultimately to development of new individuals. The germ cells of a monoploid cannot proceed through meiosis normally, because the chromosomes have no pairing partners. Thus, monoploids are characteristically sterile. Male bees, wasps, and ants bypass meiosis in forming gametes; here, mitosis produces the gametes. If a monoploid cell does undergo meiosis, the single chromosomes segregate randomly, and the probability of all chromosomes going to one pole is (1/2)x-1, where x is the number of chromosomes. This formula estimates the frequency of viable (whole-set) gametes, which is a small number if x is large.
What are the 4 steps of fertilization?

The stages of fertilization can be divided into four processes: 1) sperm preparation, 2) sperm-egg recognition and binding, 3) sperm-egg fusion and 4) fusion of sperm and egg pronuclei and activation of the zygote.

Self-Assessment Exercises 4

Provide answers to the following questions in 10 minutes

Differentiate between the terms monoploid and haploid numbers.

2.7. Summary

A monoploid is an organism with a haploid set of chromosomes. The method for their production include interspecific and intergeneric hybridization, anther and pollen culture, twin seedling and chromosome elimination.

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2.9. Possible Answers to Self-Assessment Exercises

SAE 1

An individual that contains one half the normal number of chromosomes is a monoploid and exhibits monoploidy. Monoploids are very rare in nature because recessive lethal mutations become unmasked, and thus they die before they are detected.

SAE 2

Monoploidy has been applied in plant biotechnology to rapidly develop plants from anthers that have a fixed genotype. F1 plants derived from a

cross of two parents are grown and anther tissue is used to regenerate new plants using tissue culture techniques.

SAE 3
A is expected to be more fertile than B because the monoploid number of A = 3 is less than that of B = 5. Fertility increases with increasing monoploid number based on the formular of probability of a balanced gamete = (1/2)x-1.

SAE 4
Monoploid number is the basic chromosome number, and it is represented as x, while haploid number is the number of chromosome in a gamete and it is represented as n.

Unit 3: Diploidy and Introduction to Polyploidy

CONTENTS

 3.1. Introduction

Diploids especially and monoploids are cases of normal euploidy. Most living organism exist normally as diploids. Euploid types that have more than two sets of chromosomes are called polyploids. The polyploidy types are named triploids, 3x), tetraploids (4x), pentaploids (5x), hexaploids (6x) and so forth. Polyploids may arise as spontaneous chromosomal mutation and, as such, they must be considered aberrations because they differ from the well-known diploid norm. However, many plant species have clearly arisen through polyploidy. So evidently evolution can take advantage of polyploidy when it arises.

 3.2. Learning Outcomes

At the end of the class, student must have fully understood the diploidy as cases of euploidy, and know the difference between diploidy and polyploidy in terms of their level of ploidy.

 3.3. Diploidy

This is when an organism has 2 basic homologous chromosome sets (2x). Some polyploids especially allopolyphoids (amphiploids) behave as "good diploids" after a selection process that allows polyploids that are originally meiotically irregular to become meiotically regular. This process is called diploidization. For instance, wheat is a hexaploid AABBDD (6x = 48) but it behaves as a good diploid' 2n = 48.

Why are diploids usually fertile without experiencing the problems of fertility associated with other states of euploidy?

All things being equal, diploids usually undergo normal meiosis whereby bivalents are formed and segregation of chromosomes is normal. Thus, normal haploid gametes are formed.

Self-Assessment Exercises 1

Provide answers to the following questions in 10 minutes

Differentiate between the terms monoploid and haploid numbers.

3.4. Meiotic Behavior and Fertility in Diploids

Normal bivalents are formed at meiosis and therefore balanced gametes are produced. Meiosis is a seemingly paradoxical process in which universality and uniqueness are harmoniously combined. All organisms, irrespective of their evolved complexity, meiotically reduce the chromosome number at the start of sexual reproduction, compensating for fertilization and maintaining the diploid chromosome set from generation to generation. Meiosis is a seemingly paradoxical process in which universality and uniqueness are harmoniously combined. All organisms, irrespective of their evolved complexity, meiotically reduce the chromosome number at the start of sexual reproduction, compensating for fertilization and maintaining the diploid chromosome set from generation to generation.

What is the chromosome behavior in meiosis?

The typical behavior of chromosomes in meiosis is that homologous pairs synapse, recombine, and then separate at anaphase I. At anaphase II, sister chromatids separate. However, studies of small chromosomes in maize derived from a variety of sources typically have failure of sister chromatid cohesion at anaphase I.

Self-Assessment Exercise 2

Provide Answers to the following question in 10 minutes

i. What is the importance of meiosis in plant breeding?

ii. Is meiosis a haploid of diploid?

3.5. Summary

Organisms at ploidy level of 1 are monoploids, those with chromosome complements with ploidy level of 2 are diploids. A ploidy of 3 and above are called polyploids. Meiosis is regular in diploids and therefore there is normal fertility unlike in monoploids and some polyploids.

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3.7. Possible Answers to Self-Assessment Exercises

SAE 1

The number of chromosomes found in a single complete set of chromosomes is called the monoploid number (x). The haploid number (n) refers to the total number of chromosomes found in a gamete (a sperm or egg cell produced by meiosis in preparation for sexual reproduction).

SAE 2

i. Meiosis, a specialized cell division producing haploid gametes to maintain somatic diploidy following their fusion, assures genetic variation by regulated genetic exchange through homologous recombination.

ii. Meiosis is the process by which a haploid cell is formed from a diploid cell. The difference between haploid cells and diploid cells is that haploid cells contain one complete set of chromosomes, whereas diploid cells contain two complete sets of chromosomes. Meiosis involves the division of a diploid (2n) parent cell.

Unit 4: Triploidy

CONTENTS

 4.1. Introduction

Triploids (3x) are the first category of plants among the polyploids. Triploids have chromosome complement 3x implying that chromosome complement is at ploidy level of 3. They are the first category of organisms that are polyploids. Recall that organisms with ploidy levels greater than two are polyploids. Therefore, polyploids start from triploids.

Triploidy is a rare chromosomal abnormality. Triploidy is the presence of an additional set of chromosomes in the cell for a total of 69 chromosomes rather than the normal 46 chromosomes per cell. The extra set of chromosomes originates either from the father or the mother during fertilization.

 4.2. Learning Outcomes

At the end of the class, student must be familiar and have understood triploidy as the first set of polyploids, its qualities, and the difference between autoploids and allopolyploids.

 4.3. **Autopolyploids and Allopolyploids**

In the realm of polyploids, we must distinguish between autopolyploids and allopolyploids. Autopolyploids are composed of multiple sets from within one species. Allopolyploids are composed of sets from different species. An important implication of the above is that allopolyploids arise

through hybridization. Hybridization is the formation of a progeny called hybrid from mating between genetically unrelated parents.
Important: Triploids are usually autotriploids.

When we consider allopolyploidy, the degree of unrelatedness matters. Genome allopolyploids contain clearly different basic genomes derived from different species. The genome formula is represented as AAB. Segmental allopolyploids contain genomes that are not strikingly dissimilar because the combining species are slightly related. The genome formula is represented as AAA1.

Autopolyploidy appears when an individual has more than two sets of chromosomes, both of which from the same parental species. Allopolyploidy, on the other hand, occurs when the individual has more than two copies but these copies, come from different species.
What is meant by allopolyploidy? Define Autopolyploid?
A polyploid individual or strain having a chromosome set composed of two or more chromosome sets derived more or less complete from different species.

An individual or strain whose chromosome complement consists of more than two complete copies of the genome of a single ancestral species.

Self-Assessment Exercise 1

Provide answers to the following questions in 20 minutes
Why is allopolyploid important?

4.4. Production of Triploids

4.4.1. Genetic Non-Reduction

This occurs when a normal gamete is fertilized by an unreduced pollen (Figure 7).

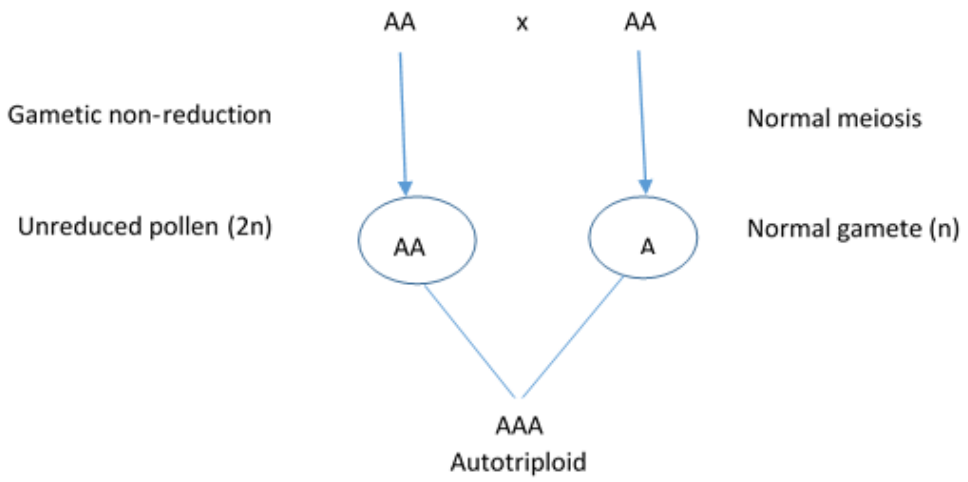

Figure 7. Gametic representation of crosses between a normal gamete and unreduced pollen

Errors occur sometimes during meiosis in regular diploid plants and chromosomes fail to segregate properly to the daughter cells. Such an unreduced 2n gamete can unite with a normal, haploid gamete, resulting in a triploid zygote that may develop into a triploid plant.

How are Triploids produced?
Triploidy can result either from the fusion of a 2n gamete to a regular reduced gamete (n), with both produced by diploid individuals, or from crosses between diploid and tetraploid individuals.

Self-Assessment Exercise 2

Provide answer to the following question in 20 minutes

How are triploid plants produced?

4.5. Fertility in Triploids
4.5.1. Meiosis in Triploids
Triploids contain odd number of chromosome set or ploidy (i.e. 3x). Thus each chromosome occurs in triplicate. The three homologous chromosomes experience pairing problems: normal bivalents characteristic of normal meiosis does not occur. Thus univalent-bivalent and trivalent pairing can occur. Possible trivalent configurations formed in triploids are shown in the Figure 8 below:

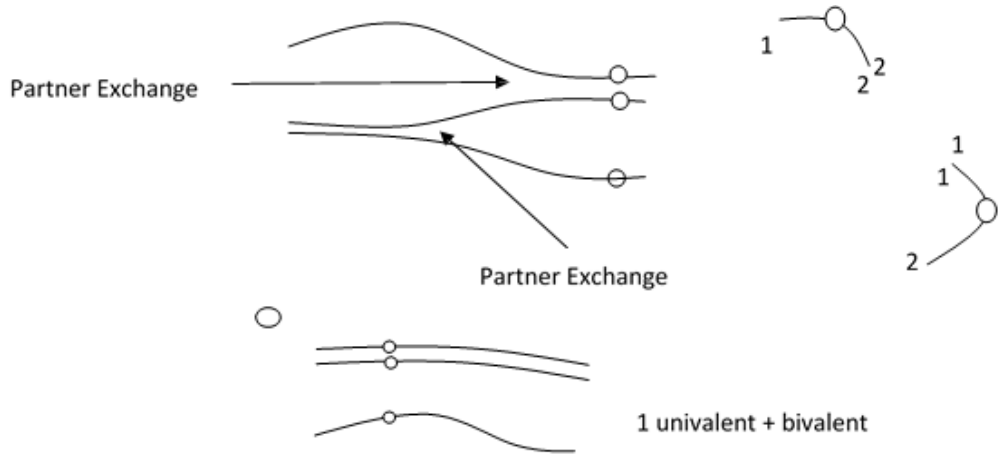

Figure 8. Principle of Partner Exchange
Note that in the trivalents, meiotic pairing in any region is united to only two homologues at a time. Regions where the chromosomes change their pairing association from one pairing partner to another is called partner exchange.

Why are Triploids sterile meiosis?
Triploids, in particular, are characterized by the problems of chromosomal pairing and segregation during meiosis, which may cause aneuploid gametes and results in sterility. Thus, they are generally considered to reproduce only asexually.

Self-Assessment Exercise 3

Provide answer to the following question in 20 minutes

How many chromosomes do triploid cells have?

4.5.2. Gamete Production
Since chromosomes occur in triplicates in triploids, there is no way to ensure that the resulting gametes obtain a complete (or balanced) chromosome complements of x or 2x. Unbalanced gametes are produced and triploids have no seeds. Thus they are sterile. Examples are banana and triploid watermelon.

The only way to ensure fertility in triploids is when all the single chromosomes pass to the same pole and simultaneously the other two chromosomes pass to the opposite pole, then the gametes formed will be balanced having haploid (x and diploid (2x) chromosome complements respectively. The probability of this type of meiosis will be (1/2)x-1.

How are Triploids produced?
Triploidy can result either from the fusion of a 2n gamete to a regular reduced gamete (n), with both produced by diploid individuals, or from crosses between diploid and tetraploid individuals.

Self-Assessment Exercise 4

Provide answer to the following question in 20 minutes

What is a triploid gamete?

4.6. Occurrence of Polyploidy in Plants as Compared to Animals
Polyploids (and monoploids) are much more common in plants than in animals. 30-35% of the angiosperms are polyploids. In Gramineae (grass family) it occurs with a frequency of 75%.
Reasons why Polyploidy is less common in Animals than in plants
(i) Disturbance of Sex Determination Mechanism
The XY sex determination mechanisms is upset in animals when chromosome sets are duplicated.
(ii) Histological Barrier
Animals are complex and polyploidy interferes with developmental pattern during tissue differentiation. For instance occurrence of plyploidy in man leads to spontaneous abortion.
(iii) Cross Fertilization Barrier
Interspectific cross fertilization is rare in animals but common in plants. Moreover, hybrids are developmentally defective.
(iv) Hybrid Sterility
Even if viable hybrids are formed they are unable to reproduce sexually, and cannot last long for the rare occurrence of chromosome doubling which gives fertile allotetraploids.
Note: Polyploid cells occur in particular organs in some mammals e.g. some liver cells are polyploids.
Is polyploidy more common in plants or animals?
One of the most striking genetic differences between plants and animals is the prevalence of polyploidy in the former group and its rarity in the latter: although 30%-50% of all angiosperm species are polyploid, few polyploid series are found among animals

Self-Assessment Exercise 5

Provide answer to the following question in 20 minutes

What is the most common polyploidy?

4.7. Advantages of Polyploidy

Increased chromosome number leads to larger nucleus. The amount of cytoplasm therefore increases to preserve the nucleus: cytoplasm ratio. This leads to increase in size of plant and its parts.

Polyploidy allow for greater genetic diversity than in its diploid progenitors since more than two alleles will be present at a locus while the diploid will have only 2 alleles per locus.

Greater enzyme multiplicity and activity resulting from gene diversion may allow the polyploidy to be more physiologically and ecologically more successful than the diploid counterparts.

Buffer Effect: In polyploids, extra chromosomes function as genetic buffers. Therefore, aneuploidy usually go unnoticed. For instance, a nullisomic diploid often does not survive; however, a nullisomic polyploidy may survive but exhibit reduced vigour and reduced fertility. The buffer effect is caused by chromosome compensation.

What are the advantages of polyploidy?

There are three obvious advantages of becoming polyploid: heterosis, gene redundancy (a result of gene duplication) and asexual reproduction. Heterosis causes polyploids to be more vigorous than their diploid progenitors, whereas gene redundancy shields polyploids from the deleterious effect of mutations.

Self-Assessment Exercise 6

Provide answer to the following question in 20 minutes

i. Why is it that banana cannot be propagated by seed?

ii. What are the disadvantages of polyploidy?

4.8. Summary

Triploids are the first group of polyploids. They have 3x chromosome complement. Although triploids have improved qualities when compared to diploids, they are sterile because unbalanced gametes and seedlessness result from meiotic irregularities of triploids.

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4.10. Possible Answers to Self-Assessment Exercises

SAE 1
Recent studies have shown that allopolyploidy accelerates genome evolution in wheat in two ways: (1) allopolyploidization triggers rapid genome changes (revolutionary changes) through the instantaneous generation of a variety of cardinal genetic and epigenetic alterations, and (2) the allopolyploid condition

SAE 2
Triploid production by the conventional techniques involves chromosome doubling, followed by crossing the tetraploids with their diploids. This approach is not only laborious, but in many cases, may not be possible because of the high sterility of tetraploids.

SAE 3
46 chromosomes
A cell containing two copies of each chromosome is referred to as a 'diploid cell'; human somatic cells are diploid: they contain 46 chromosomes, 22 pairs of autosomes and a pair of sex chromosomes.

SAE 4
Triploidy is the presence of an additional set of chromosomes in the cell for a total of 69 chromosomes rather than the normal 46 chromosomes per cell. The extra set of chromosomes originates either from the father or the mother during fertilization.

SAE 5
i. Polyploids are common in insects, fish and amphibians, and hybrids such as mule also occur in mammals. Estimates indicate that ~10% of animal and ~25% of plant species hybridize with at least one other species. Two models for the origin of allopolyploids have been proposed.

ii. Aneuploidy
Aneuploidy is more common. Polyploidy occurs in humans in the form of triploidy, with 69 chromosomes (sometimes called 69, XXX), and tetraploidy with 92 chromosomes (sometimes called 92, XXXX). Triploidy, usually due to polyspermy, occurs in about 2–3% of all human pregnancies and ~15% of miscarriages.

SAE 6
i. Banana is triploid. Since the ploidy number is odd, there is no way to ensure normal bivalents at meiosis. Thus segregation of chromosomes is irregular. The gametes will be mainly aneuploids and non-functional. So banana is seedless. The only option left for propagation of banana is through vegetative means.

ii. Among the disadvantages that could lead to less vigor and a reduced adaptive capacity in polyploids are the increased number of chromosomes, and the greater complexity of their pairing and segregation interactions that can cause abnormalities (including aneuploidy) during meiosis and mitosis

Unit 5: Tetraploidy and Higher Polyploidy

CONTENTS

 5.1. Introduction

Tetraploids are the second group of polyploids after triploids. As the name suggests, tetraploids have 4x chromosome complement. This means that each chromosome type occurs in quadruplicate. Unlike triploids that are usually autotriploids, autotetraploids and allotetraploids occur frequently in plants.

 5.2. Learning Outcome

At the end of the class, student must be familiar and have understood how tetraploidy occurs well in plants, chromosome complement and genome formulas of different types of autotetraploids, and the advantage tetraploids have over triploids in terms of fertility.

 5.3. **Autotetraploids**

Autotetraploids have 4x chromosome complement with genome formula AAAA. **If two diploid gametes fuse**, an autotetraploid will be created whose nucleus contains four copies of each chromosome (Figure 10). Autopolyploids are often viable because each chromosome still has a homologous partner and can therefore form a bivalent during meiosis.

5.3.1. Production of Autotetraploids

Production involves 2 mechanisms:

(a) Genetic non-reduction and

(b) Somatic doubling

Chromosome doubling can be spontaneous or induced. Induction is through colchicines treatment, a poisonous alkaloid drug that binds to tubulin, the major protein component of the spindle thereby preventing formation of the spindle apparatus. In cells without spindle apparatus, the sister chromatids do not separate after the centromere splits, so the chromosome no doubles. Since there is no spindle, metaphase chromosomes (colchicines metaphase or c- metaphase) remain scattered in the cytoplasm. Continuous colchicines treatment may cause reduplication. For instance, onion cells (2n = 16) bathed with colchicines for 4 days may contain 1,000 chromosome per nucleus.

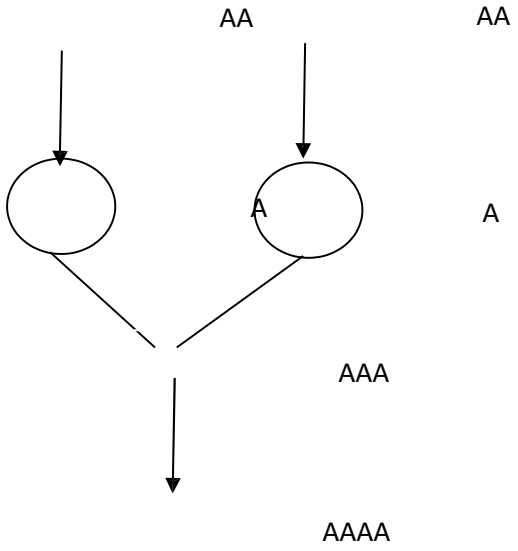

Figure 10. Crosses between two diploid gametes

What are autotetraploids?
an individual or strain whose chromosome complement consists of four copies of a single genome due to doubling of an ancestral chromosome complement.

5.3.2. Fertility in Autotetraploids
Because 4 is an even number, autotetraploids can have a higher meiosis. The crucial factor is how the four homologous chromosomes pair and segregate. There are several possibilities which include two bivalents, one quadrivalent and a univalent-trivalent association. In tetraploids, the two-bivalent and the quadrivalent pairing modes tend to be most regular, even here there is no guarantee for a 2:2 segregation.

If all chromosome sets segregate 2:2 as they do in some species, then the gametes will be functional and genetic analysis can be made.
Are Autopolyploids fertile?

Autopolyploidy can double the chromosome number, producing a fertile hybrid with two of each chromosome. If these new fertile hybrids cannot reproduce with the parental types, but can reproduce with each other, a new species has been formed.

Self-Assessment Exercise 1

Provide short answers to the following questions in 10 minutes

i. Why Autopolyploids are usually sterile?

ii. What are the limitations of Autopolyploids?

5.4. Genetics of a Fertile Autotetraploids

5.4.1. Determination of Genotypes

In an autotetraploid, a gene is represented 4 times at a locus. Thus, we can have the following allelic constitutions with reference to the dominant allele 'A' at locus A/a.

AAAA - quadriplex AAAa - triplex AAaa - duplex Aaaa - simplex aaaa - nulliplex

Gamete production is a duplex – Aaaa.

Note: We have further concern whether the locus in question is tightly linked or not to the centromere since the two situations give different results.

Case 1: Locus is tightly linked to the centromere:

	A	A	a	a
A	A	AA	Aa	Aa
A		A	Aa	a
a			a	aa
a				a

Balanced Gametes i.e. diploid gametes = 4Aa: 1AA: 1aa = 6

Thus if there are 4 different alleles at the locus, we shall have 6 different alleles

$$^4C_2 = \frac{4!}{(4-2)!2!} = \frac{4.3.2!}{2!\ 2!}$$

= 6 by combinatorial analysis

Therefore, the probability of a nulliplex progeny (a/a/a/a) is 1/6 x 1/6 = 1/36
Ratio = 1a/a/a/a : 35 A/-/-/-

OR

	4Aa	1AA	1aa	
4Aa	16AAaa	4AAAa	4Aaaa	
1AA	4AAAa	1AAAA	1AAaa	
1aa	4Aaaa	1AAaa	1aaaa	
Total	24	6	6	= 36

1/36 aaaa

Case 2: Locus is not linked to the centromere.
In this case crossing-over must be considered. This forces us to think in terms of chromatids.

The packaging of genes two at a time into games is very much like grabbing two balls at random from a bag of eight balls: 4 of one kind, 4 of another. The probability of picking two b genes = probability of bb gamete.
= 4/8 (the first one) x 3/7 (the 2nd one) = 3/14

... b/b/b/b = (3/14)2 = 9/196

How is Autotetraploid formed?
The most common one is through fusion of unreduced gametes. Autotetraploidy can also occur via cross-fertilization between an unreduced gamete and a diploid gamete from a triploid intermediate, also known as a "triploid bridge".

Self-Assessment Exercise 2

Provide short answers to the following questions in 10 minutes

Why are tetraploids fertile?

5.5. Allotetraploids
5.5.1. Classical Allotetraploid – *Raphinobrassica*
The "classic allotetraploid" was synthesized by G. Karpechenko in 1928. He wanted to make a fertile hybrid that would have the leaves of the cabbage (*Brassica*) and the roots of the radish (*Raphanus*). Each of these species has 18 chromosomes, and they are related closely enough to allow intercrossing. A viable hybrid progeny individual was produced from

seed. However, this hybrid was functionally sterile because the nine chromosomes from the cabbage percent were different enough from the radish chromosomes that pairs did not synapse and disjoin normally (Figure 11):

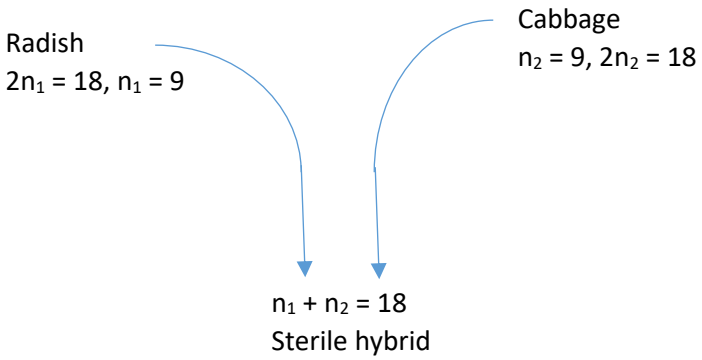

Figure 11. Allotetraploid as achieved by crosses between Radish and Cabbage

However, one day a few seeds were in fact produced by this (almost) sterile hybrid. On planting, these seeds produced fertile individuals with 36 chromosomes. All these individuals were allopolyploids. They had apparently been derived from spontaneous, accidental chromosome doubling to *2n1 + 2n2* in the sterile hybrid, presumably in tissue that eventually became germinal and underwent meiosis. Thus, in *2n1 + 2n2* tissue, there is a pairing partner for each chromosome and balanced gametes of the type *n1 + n2* are produced. These gametes fuse to give *2n1 +2n2* allopolyploid progeny, which also are fertile. This kind of allopolyploid is sometimes called an amphidiploids, which means "doubled diploid" (Figure 10). Unfortunately for Karpechenko, his amphidiploids had the roots of a cabbage and the leaves of a radish.

When the allopolyploid was crossed with either parental species, sterile offspring resulted. The offspring of the cross with radish were *2n1 + n2*, constituted from an *n1 + n2* gamete from the allopolyploid and an n1 gamete from the radish. The *n2* chromosomes had no pairing partners, so sterility resulted. Consequently, Karpechenko had effectively created a new species, with no possibility of gene exchange with its parents. He called his new species *Raphanobrassica*.

What are allotetraploids?

Allotetraploids are hybrid cells or individual possessing four times the chromosomes in a haploid organism. Adjective or possessing four times the chromosomes in a haploid organism.

5.5.2. Production of Allotetraploid

It is produced by hybridization of 2 different species to yield an infertile hybrid in F1. F1 must be able to propagate vegetatively before chromosome doubling to produce a vigorous fertile hybrid.

5.5.2.1. Production of Allotetraploid Triticale by Hybridization

Today, allopolyploids are routinely synthesized in plant breeding. Instead of waiting for spontaneous doubling to occur in the sterile hybrid, the plant breeder adds colchicines to induce doubling. The goal of the breeder is to combine some of the useful features of both parental species into one type. This kind of endeavor is very unpredictable, as Karpechenko learned. In fact, only one synthetic amphidiploids has ever been widely used. This amphidiploids is *Triticale*, an amphiphidiploid between wheat (*Triticum*, 2n = 6x = 42) and rye (*Secale*, 2n = 2x = 14). *Triticale* combines the high yields of wheat with the ruggedness of rye. Figure 11 shows the procedure for synthesizing Triticale.

How are Allopolyploids produced?

Allopolyploids are formed by the hybridization of two closely related species, primarily by fertilization of two unreduced gametes or, to a lesser extent, by genome doubling after fertilization of two reduced gametes

Self-Assessment Exercise 3

Provide answer to the following question in 10 minutes

Arrange the plants with the genome formulas below according to their degree of fertility y starting with the most fertile. Give reasons to support your answer.

AAAAA. AABB. AAAA

5.6. Summary

There are two major types of tetraploids namely autotetraploids and allotetraploids. Tetraploids have considerably high fertility because the ploidy is even (i.e. 4x). Several mechanisms ensure that normal bivalents are formed as is the case with normal diploids. Thus, an odd number of chromosomes sets makes an organism sterile because there is not a partner for each chromosome at meiosis, whereas even number of sets can produce standard segregation ratios to cause fertility

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 5.8. Possible Answers to Self-Assessment Exercises

SAE 1
i. Because autopolyploid individuals have three or more chromosome sets, each chromosome has more than one homologous pairing partner. During meiosis, multivalents are produced leading to unbalanced gametes and zygotes, sterility, and other problems.

ii. Autopolyploids generally have no novel alleles and therefore little genetic advantage over their progenitor diploid species. The fertility of autotetraploids is frequently reduced because of multivalent chromosome association at meiosis I.

SAE 2
When two of these gametes (2n) combine, the resulting offspring are tetraploid (4n). This is common in plants as they produce both male and female gametes and are often capable of self-fertilisation. The resulting offspring a generally fertile as they have an even number of chromosomes.

SAE 3
The arrangement of the genome formula will be
AABB, AAAA, AAAAA
AABB is an allotetraploid with normal fertility because chromosome are in pairs, and there is formation of normal bivalents. AAAA (autotetraploid) is also fertile because the ploidy number is even; however, possibilities of multivalent and other abnormal pairings cannot be totally ruled out. AAAAA is a pentaploid with odd ploidy number.

Meiosis will be irregular leading to aneuploid gametes which will be non-functional.

Glossary
mol^{-1} – Unit mole
g mol^{-1} – gram par unit mole
Γ - gamma function
N_A – Avogadro number
μg/mL – microgram per millilitre
mL - millilitre
DNA – Deoxyribonucleic acid
n - number
F1- First filial generation

End of Module Questions
How many chromosomes does an Allotriploid have?
40 chromosomes
It has two sets of chromosome pairs, one from each of the ancestral species: a type of polyploid termed allotetraploid (AABB-type genome; 2n = 4× = 40 chromosomes; genome size of ~2.7 Gb).

What is the role of Allopolyploidy in evolution?
Recent studies have shown that allopolyploidy accelerates genome evolution in wheat in two ways: (1) allopolyploidization triggers rapid genome changes (revolutionary changes) through the instantaneous generation of a variety of cardinal genetic and epigenetic alterations, and (2) the allopolyploid condition

What is the difference between an Allotetraploid and a Autotetraploid?
An autotetraploid plant has four copies of the same chromosome set. In contrast, an allotetraploid or amphidiploid plant has two diploid sets of chromosomes derived from distinct species.

Why are autotriploid crop plant sterile?
One way to produce an autotriploid is by mating a diploid with haploid (n) gamete and a tetraploid with 2n gamete. The offspring would have an unbalanced gametes and therefore possibly sterile. Thus, autotriploids are mostly propagated asexually.

How can a sterile hybrid plant become a fertile tetraploid?
Polyploid hybrids commonly form in nature in plants, and hybrid plants can be fertile when they are crossed with other polyploid plants with even numbers of chromosomes. In order for plants to be fertile, they must be able to produce gametes with balanced numbers of chromosomes in their cells.

Can sterile hybrids reproduce?
In short, hybrid animals are infertile because they don't have viable sex cells, meaning they can't produce sperm or eggs. This is the case because the chromosomes from their different species parents don't match up.

Module 3

Module 3
Unit 1: Aneuploidy: An Overview
CONTENTS

 1.1 Introduction

Aneuploidy is a numerical variation that affects a particular chromosome and not the whole set as is the case with euploidy. Aneuploidy is the second major category of chromosome mutations in which chromosome number is abnormal. An aneuploidy is an individual organism whose chromosome number differs from the wild type by part of a chromosome set. Generally, the aneuploid chromosome set differs from wild type by only one or a small number of chromosomes. Aneuploids can have a chromosome number either greater or smaller than that of the wild type. Aneuploid nomenclature is based on the number of copies of the specific chromosome in the aneuploid state. For example, the aneuploid condition 2n-1 is called **monosomic** (meaning "one chromosome") because only one copy of some specific chromosome is present instead of the usual two found in its diploid progenitor. The aneuploid 2n +1 is called **trisomic**, 2n-2 is **nullisomic**, and n +1 is **disomic**.

 1.2. Learning Outcomes

At the end of the class, student must be familiar and will be able to distinguish aneuploids from euploids, understand the causes of aneuploidy, and the concept of genic balance and how it affects aneuploids and euploids.

1.3. Types of Aneuploids

Aneuploidy is the presence of chromosome number that is different from the simple multiple of the basic chromosome number. An organism which contains one or more incomplete chromosome sets is known as aneuploidy (Table 2). Aneuploidy can be either due to loss of one or more chromosomes (hypo-ploidy) or due to addition of one or more chromosomes to complete chromosome complement (hyper-ploidy). There are two main types of aneuploidy; there can be an extra copy of a chromosome (trisomy) or a missing copy of a chromosome (monosomy). Humans have 23 pairs of chromosomes, totaling 46 chromosomes. A person with an aneuploidy condition typically has either one more or one less chromosome than normal. Aneuploidy arises from errors in chromosome segregation, which can go wrong in several ways.

Table 2. Tabular representation of Aneuploids

		Normal (2n) 1 1 2 2 3 3 4 4		
nullisomy (2n-2) 1 1 2 2 4 4 5 5	monosomy (2n-1) 1 1 2 3 3 4 4	disomy (2n) 1 1 2 2 3 3 4 4	trisomy (2n + 1) 1 1 2 2 2 3 3 4 4	tetrasomy (2n + 2) 1 1 2 2 2 2 3 3 4 4
double nullisomy (2n-2-2) 1 1 3 3 5 5	double monosomy (2n-1-1) 1 1 2 3 3 4 5 5		double trisomy (2n + 1 +1) 1 1 2 2 2 3 3 4 4 4 5 5	double tetrasomy (2n+2+2) 1 1 2 2 2 2 3 3 4 4 4 4 5 5

What are the causes of aneuploidy?
Errors in chromosome segregation lead to aneuploidy, a state where the number of chromosomes in a cell or organism deviates from multiples of the haploid genome. Aneuploidy arising through chromosome mis-segregation during meiosis is a major cause of infertility and inherited birth defects.

Self-Assessment Exercise 1

Provide answers to the following questions in 20 minutes

i. What is risk of aneuploidy?

ii. Is aneuploidy normal?

1.4. Aneuploid Series

Aneuploid series refers to all possible aneuploids (Table 3). Thus, it is equal to the number of haploid chromosome number. Therefore, if an organisms has chromosome number 2n = 5, then there will be 5 kinds of aneuploids given a particular type of aneuoploidy. This is because each chromosome type can be affected by aneuploidy.

Table 3: Monosomic Series for a hypothetical Plant with 2n=5. There are 5 possible aneuploids

normal	Monosomy -1	Monosomy -	Monosomy – 3
1 1 2 2 3 3 4 4	1 2 2 3 3 4 4	2 1 1 2 3 3	1 1 2 2 3 4 4
Monosomy – 4 1 1 2 2 3 3	Monosomy-5 1 1 2 2 3 3 4 4		

A diploid plant was observed to have chromosome number 2x=14. How many types of monosomics do you expect to constitute its monosomic series?

2x = 14, therefore x + 7. Thus, there are 7 types of nullisomics in the series.

Self-Assessment Exercise 2

Provide answers to the following questions in 20 minutes

i. What does it mean to be an aneuploidy?

ii. Is aneuploidy normal?

1.5. Causes of Aneuploidy

i. Nindisjunction: Members of a pair of chromosomes or chromatids fail to disjoin (separate) properly so that some gametes are formed containing more or less of a particular chromosome. This may lead to hypoploidy and hyperploidy.

ii. Lagging Chromosomes: Lagging chromosomes or laggards are characterized by retarded movement during anaphase resulting in non-incorporation into gametes. This always leads to hypoploidy.

iii. Irregular Chromosome Distribution: Random chromosome distribution into gametes may cause more or less of particular chromosomes in the resulting individual.

iv. Multipolar Mitosis: Abnormal chromosome distribution.

Explain why aneuploidy usually has more deleterious effect than polyploidy?

Aneuploidy offsets the normal proportion of genes than euploidy and therefore causes more serious problem of genome imbalance.

Self-Assessment Exercise 3

Provide answers to the following questions in 20 minutes

i. How do you think multipolar mitosis might cause aneuploidy?

ii. What is the most common cause of aneuploidy?

1.6. Concept of Genic Balance

Genic balance refers to the proper amount of gene products resulting from appropriate rate of transcription and consequently the number of copies of that gene in a cell.

The theory of genic balance given by Calvin Bridges (1926) states that instead of XY chromosomes, sex is determined by the genic balance or ratio between X-chromosomes and autosome genomes. In Drosophila, Y chromosome is not involved in the process of sex determination.

1.6.1. Genic Balance and Development

Normal development depends on gene balance i.e. genes in appropriate amount. In euploidy multiples of the whole set does not affect this balance significantly because there is no change in the relative proportions of genes. However, in aneuploidy, numerical changes not involving the whole set alters the proportions of genes. Therefore, polyploidy does not usually cause a wide change in development as aneuploidy.

Note: Aneuploidy disturbs genic balance more significantly than euploidy.

What does genic balance mean?

The relation whereby a specific gene acts as a part of the entire gene complex in the production of a particular phenotypic character.

Self-Assessment Exercise 4

Provide answer to the following question in 10 minutes

i. Who proposed the genic balance theory Drosophila?

ii. What is the genic balance ratio in super male?

1.7. Summary

Aneuploids usually results in an unbalanced genome with an abnormal phenotype. Examples of aneuploids include nullisomic (2n-3), monosomic (2n-1), trisomics (2n+1). There are also double, triple etc. aneuploids. Aneuploid series refers to all possible aneuploids of a particular type.

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 1.9. Possible Answers to Self-Assessment Exercises

SAE 1
i. Aneuploidy is the most common chromosomal abnormality and also is the leading cause of early fetal loss and serious mental retardations.

ii. Aneuploidy is extraordinarily common in humans, occurring in an estimated 20–40% of all conceptions. It is the most common cause of miscarriages and congenital defects in our species and is a leading impediment to the treatment of infertility. Most aneuploidy results from maternal meiotic nondisjunctional errors.

SAE 2
i. The occurrence of one or more extra or missing chromosomes leading to an unbalanced chromosome complement, or any chromosome number that is not an exact multiple of the haploid number

ii. Aneuploidy is extraordinarily common in humans, occurring in an estimated 20–40% of all conceptions. It is the most common cause of miscarriages and congenital defects in our species and is a leading impediment to the treatment of infertility. Most aneuploidy results from maternal meiotic nondisjunctional errors.

SAE 3
i. Multipolar mitosis means there are more than two poles for chromosome distribution during meiosis. This will make normal distribution of chromosomes into gametes impossible, and aneuploid gametes will be produced.

ii. The most common cause of aneuploidy is nondisjunction, the failure of chromosomes to disjoin normally during meiosis. Nondisjunction can occur during meiosis I or II or during mitosis, although maternal meiosis I is the most common nondisjunction in aneuploidies (e.g., Down syndrome, trisomy 18).

SAE 4

i. Calvin Bridges
Hint: The genic balance theory of sex determination in Drosophila was given by Calvin Bridges in 1926. He states that instead of XY chromosomes, sex determined by the genic balance or the ratio between the X-chromosomes and autosome genomes.

ii. According to the theory of the genetic balance: Gene ratio (X/A) of 1.0 produces fertile females. Genic ratio (X/A) of 0.5 is a productive male. Genic ratio less than 0.5 produced infertile meta-males (super males).

Unit 2: Aneuploidy and its Genetic Consequences

CONTENTS

2.1. Introduction

It was learnt in the last unit of this module that genic imbalance is an important genetic consequence of aneuploidy. There are, however, other genetic consequences that need to be understood. In this unit, therefore, more detail consideration of aneuploidy and their genetic consequences will be taught.

2.2. Learning Outcomes

At the end of the class, student must be familiar and have understood other genetic consequences of aneuploidy apart from genic imbalance, and its effect on gamete production and inheritance.

2.3. Nullisomics (2n-2)

Although nullisomy is a lethal condition in diploids, an organism such as bread wheat, which behaves meiotically like a diploid although it is a hexaploid, can tolerate nullisomy. The four homoeologus chromosomes apparently compensate for a missing pair of homologs. In fact, all the possible 21 bread wheat nullisomics have been produced. Their appearances differ from the normal hexaploids; furthermore, most of the nullisomics grow less vigorously.

Nullisomy is usually lethal, but allotetraploids have a chance of surviving lethal effect of nullisomy. Why?

Other chromosomes compensate for the lost ones. This phenomenon is called chromosome compensation.

Self-Assessment Exercise 1

Provide answer to the following question in 5 minutes

Why is aneuploidy of more deleterious consequence than euploidy?

2.4. Monosomics (2n-1)

Monosomic chromosome complements are generally deleterious for two main reasons. First, the missing chromosome perturbs the overall gene balance in the chromosome set. (We encountered this effect earlier). Second, having a chromosome missing allows any deleterious recessive allele on the single chromosome to be hemizygous and thus to be directly expressed phenotypically. Notice that these are the same effects produced by deletions.

What does the term monosomic mean? The term monosomic is used to describe a cell that has only a single copy of a given chromosome. If a monosomic plant is produced from a diploid one, all the chromosomes are present in two copies except one, which is present in a single copy.

Self-Assessment Exercise 2

Provide answer to the following question in 5 minutes

What causes monosomy?

2.5. Trisomics (2n +1)

The trisomic condition also is one of chromosomal imbalance and can result in abnormality or death. However, there are many examples of viable trisomics. Furthermore, trisomics can be fertile. When cells from some trisomic organisms are observed under the microscope at the time of meiotic chromosome pairing, the trisomic chromosomes are seen to form a trivalent, an associated group of three, whereas the other chromosomes form regular bivalents.

What is trisomic?

Trisomics are aneuploids, carrying an extra chromosome to the normal somatic complement. The nature of this additional chromosome determines its types; normal in primary, isochromosome in secondary, and translocated in tertiary or interchange types.

Self-Assessment Exercise 3

Provide answer to the following question in 5 minutes

What is trisomic in plants??

2.6. Genetic Analysis of Trisomics for Allelic Constitution

The method is similar to that of triploids.

Which is trisomic condition formula?

The number of chromosomes in the cell where trisomy occurs is represented as, for example, 2n+1 if one chromosome shows trisomy, 2n+1+1 if two show trisomy, etc. "Full trisomy", also called "primary trisomy", means that an entire extra chromosome has been copied.

Self-Assessment Exercise 4

Provide answer to the following question in 5 minutes

What is trisomic in genetics?

2.7. Summary

- Nullisomy (2n – 2) is lethal in diploids, amphidiploids (allopolyploids) survive as a result of chromosome compensation.
- Monosomic (2n-1) show the deleterious effect of genome imbalance. They are hemizygous for deleterious recessive alleles on the monosomic chromosome.
- Trisomics (2n+1) show the deleterious effect of genome imbalance. However, unlike in monosomy, deleterious recessive alleles are masked by the dominant alleles on the trisomic chromosome.

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 2.9. Possible Answers to Self-Assessment Exercises

SAE 1
Aneuploids suffer genic imbalance since the change in chromosome number does not involve the whole set unlike the case with euploids. Genic imbalance results from the fact that the genes are not in proportion.

SAE 2
Monosomy. The complete absence of an X chromosome generally occurs because of an error in the father's sperm or in the mother's egg. This results in every cell in the body having only one X chromosome.

SAE 3
Trisomic plants can be distinguished from diploids byenlarged bracts (Sutton, 1939), an increased number of flowers per inflorescence and slightly undulated margins of stipules and leaflets (Berdnikov et al., 1993), and the mean number of seeds per pod (3.5) is reduced compared to ordinary diploids

SAE 4
Trisomy ('three bodies') means the affected person has three copies of one of the chromosomes instead of two. This means they have 47 chromosomes instead of 46. Down syndrome, Edward syndrome and Patau syndrome are the most common forms of trisomy.

Unit 3: Variations in Chromosome Structure

CONTENTS

3.1. Introduction

Variation in chromosome structure involve changes in parts of chromosomes rather than changes in the number of chromosomes or sets of chromosomes in a genome. There are four types of such mutations: *deletions* and *duplications* (both of which involve a change in the amounts of DNA on a chromosome), *inversions* (which involve a change in the arrangement of a chromosomal segment), and translocations (which involve a change in the location of a chromosomal segment). Duplication, inversion, and translocation mutations can change back (revert) to the wild-type state by a reversal of the process by which they were formed. However, deletion mutations cannot revert because a whole segment of chromosome is missing, not simply changed in position or copy number. All four classes of chromosomal structure mutations are initiated by one or more breaks in the chromosome. If a break occurs within a gene, then a gene mutation has been produced, the consequence of which depends on the function of the gene and the time of its expression. Wherever the break occurs, the breakage process leaves broken ends without the usual specialized sequences found at the ends of chromosomes (the telomeres) that prevent degradation by exonucleases and "stickiness". As a result, the end of a chromosome that has broken is "sticky", meaning that it may adhere to other broken chromosome ends or even to the normal ends of other chromosomes. This stickiness properly can help us understand the formation of the types of chromosomal structure mutations.

In discussions of chromosome rearrangements. It is convenient to use letters to represent different chromosome regions. These letters therefore represent large segments of DNA, each containing many genes.

The simple loss of a chromosomal segment is called a **deletion** or **deficiency.**

In the following diagram, region B has been deleted:

A B C D E F A C D E F

The presence of two copies of a chromosomal region is called a **duplication**:

A B B C D E F

A segment of a chromosome can rotate 180 degrees and rejoin the chromosome, resulting in a chromosomal mutation called an **inversion**:

A E D C B F

Finally, two nonhomologous chromosomes can exchange parts to produce a chromosomal mutation called a **translocation**:

A B C D J K G H I E F

3.2. Learning Outcomes

Student must have understood the process of deletion and duplication, and cytological and genetic consequences of deletion and duplication.

3.3. Deletion

The process of spontaneously occurring deletion must include two chromosome breaks to cut out the intervening segment. If the two ends join and one of them bears the centromere, a shortened chromosome results, which is said to carry a deletion. The deleted fragment is acentric; consequently it is immobile and will be lost. An effective mutagen for inducing chromosomal rearrangements of all kinds is ionizing radiation. This kind of radiation, of which X rays and rays are examples, is highly energetic and causes chromosome breaks. The way in which the breaks rejoin determines the kind of rearrangement produced. Two types of deletion are possible. Two breaks can produce an **interstitial deletion**. In principle, a single break can cause a **terminal deletion**; but, because of the need for the special chromosome tips (telomeres), it is likely that apparently terminal deletions include two breaks, one close to the telomere.

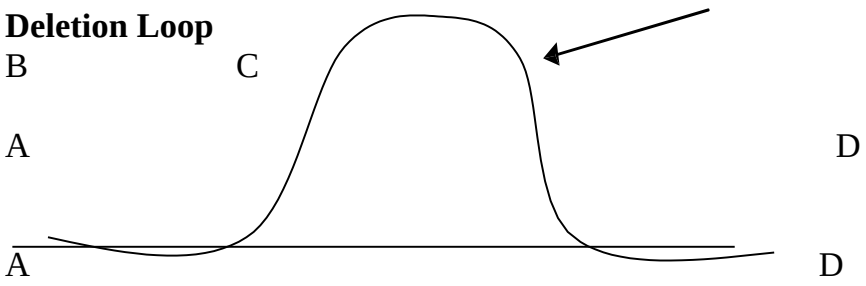
The effects of deletions depend on their5 size. A small deletion within a gene, called an **intragenic deletion,** inactivates the gene and has the same effect as other null mutations of that gene. If the homozygous null phenotype is viable (as, for example, in human albinism), then the homozygous deletion also will be viable. Intragenic deletions can be

distinguished from single nucleotide changes because they are nonrevertible.

3.3.1. Consequences of Deletion

Deletion becomes very serious if it is multigenic. Multigenic deletions are those that remove two to several thousand genes. If multigenic deletion is made homozygous (that is, if both homologs have same deletion), then the combination is almost always lethal. This outcome suggests that most regions of the chromosomes are essential for normal viability and that complete elimination of any segment from the genome is deleterious. Even individuals heterozygous for a multigenic deletion – those with one normal homolog and one that carries the deletion – may not survive. There are several possible reasons for this failure to survive. First, a genome has been "fine-tuned" during evolution to require a specific balance of genes, and the deletion upsets this balance. We shall encounter this balance notion several times in this chapter and the next, because several different types of chromosome mutations upset the ratio, or balance, of genes in a genome. Second, in many organisms there are recessive lethal and other deleterious mutations throughout the genome. If "covered" by wild-type alleles on the other homolog, these recessives are not expressed. However, a deletion can "uncover" recessives, allowing their expression at the phenotypic level.

Some heterozygous deletions are viable. In these cases the deletion can sometimes be identified by cyctogenetic analysis (Figure 12). During meiosis, homologous chromosomes attempts to maximize pairing such that corresponding segment on the normal homolog forms a deletion loop.



Note : Chromosomal region BC is deleted

Figure 12. Cytogenetic analysis showing deletion region (loop)

What is deletion and its types?

Types of deletion include the following: Terminal Deletion - a deletion that occurs towards the end of a chromosome. Intercalary Deletion - a deletion that occurs from the interior of a chromosome.

Is deletion mutation good or bad?
Because an insertion or deletion results in a frame-shift that changes the reading of subsequent codons and, therefore, alters the entire amino acid sequence that follows the mutation, insertions and deletions are usually more harmful than a substitution in which only a single amino acid is altered.

Self-Assessment Exercise 1

Provide answers to the following questions in 20 minutes

What happens if you have chromosome deletion?

3.4. Duplication
The process of chromosome mutation sometimes produce an extra copy of some chromosome region. In considering a haploid organisms, which has one chromosome set, we can easily see why such a product is called duplication because the region is now present in duplicate. The duplicate regions can be located adjacent to each other or one of the duplicate regions can be in its normal location and the other in a novel location on a different part of the same chromosome or even on another chromosome. In a diploid organism, the chromosome set containing the duplication is generally present together with a standard chromosome set. The cells of such an organism will thus have three copies of the chromosome region in question, but nevertheless such duplication heterozygotes are generally referred to as duplications because they carry the product of one duplication event.
Cytologically, duplication heterozygotes also show interesting pairing structures at meiosis. The precise structure that forms depends on the type duplication. We should concern ourselves with adjacent duplications which can be:
Tandem: A B CD → A B C B C D or Reverse: A B C D → A B C C B D

3.4.1. Evolutionary Significance of Duplication
Duplication is a very important process in gene evolution. The extra region of a duplication is free to undergo gene mutation because the necessary basic functions of the region will be provided by the other copy. Mutation in the extra region provides an opportunity for divergence in the function of the duplicated genes, which could be advantageous in genome evolution. Indeed, from situations in which different gene products with related functions can be compared, such as the globins, there is good evidence that these products arose as duplicates of one another.

What is duplication in genetics?

Duplication, as related to genomics, refers to a type of mutation in which one or more copies of a DNA segment (which can be as small as a few bases or as large as a major chromosomal region) is produced. Duplications occur in all organisms.

Self-Assessment Exercise 2

Provide answers to the following questions in 20 minutes

What is duplication and its types?

3.5. Summary

Variation in chromosome structure are exemplified by those mutations in which changes from the normal state occur in parts of individual chromosomes rather than number of chromosome. The four major types of structural mutations are:

- deletion, in which a chromosome segment is lost.
- duplication, in which more copies of a chromosome segment are present than in the normal state.
- Inversion, in which the orientation of a chromosome segment is opposite that of the wild type and
- translocation, in which a chromosome segment has moved to a new location in the genome. The consequences of these structural mutations depend on the specific mutation involved.

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 3.7. Possible Answers to Self-Assessment Exercises

SAE 1
Chromosomal deletion syndromes result from loss of parts of chromosomes. They may cause severe congenital anomalies and significant intellectual and physical disability.

SAE 2
Gene duplication can occur by several mechanisms, including whole-genome duplication (WGD) and single gene duplication. Single gene duplication includes four types, tandem (TD), proximal (PD), retro-transposed (RD), DNA-transposed (DD) and dispersed duplication (DSD)

Unit 4: Inversion

CONTENTS

 4.1. Introduction

If two breaks occur in one chromosome, sometimes the region between the breaks rotates 180 degrees before rejoining with the two end fragments. Such an event creates a chromosomal mutation called an **inversion**. Unlike deletions and duplications, inversions do not change the overall amount of the genetic material, so inversions are generally viable and show no particular abnormalities at the phenotypic level. In some cases, one of the chromosome breaks is within a gene of essential function, and then that breakpoint acts as a lethal gene mutation linked to the inversion. In such a case, the inversion could not be bred to homozygousity. However, many inversions can be made homozygous; furthermore, inversions can be detected in haploid organisms.

 4.2. Learning Outcomes

Students must be familiar and understand the process and types of inversion, and the genetic and cytological consequences of inversion.

 4.3. Types and Consequences of Inversion

Most analyses of inversions use heterozygous inversions – diploids in which one chromosome has the standard sequence and one carries the inversion. Microscopic observation of meioses in inversion heterozygotes reveals the location of the inverted segment because one chromosome twists once at the ends of the inversion to pair with the other, untwisted chromosome; in this way the paired homologs form an **inversion loop** (Figure 13).

The location of the centromere relative to the inverted segment determines the genetic behavior of the chromosome. If the centromere is outside the inversion, then the inversion is said to be **paracentric**, whereas inversions spanning the centromere are **pericentric**.

How do inversion behave genetically? Crossing-over within the inversion loop of a paracentric inversion connects homologous centromeres in a **dicentric bridge** while also producing an **acentric fragment -** a fragment

without a centromere. Then, as the chromosomes separate in anaphaseI, the centromeres remain linked by the bridge, which orients the centromeres so that the noncrossover chromatids lie farthest apart. The acentric fragment cannot align itself or move and is, consequently, lost. Tension eventually breaks the bridge, forming two chromosomes with terminal deletions (Figure 13). The gametes containing such deleted chromosomes may be inviable but, even if viable, the zygotes that they eventually form are unviable. Hence, a crossover event, which normally generates the recombinant class of meiotic products, instead produces lethal products. The overall result is a lower recombinant frequency. In fact, for genes within the inversion, the RF is zero. For genes flanking the inversion, the RF is reduced in proportion to the relative size of the inversion.

Inversions affect recombination in another way too. Inversion heterozygotes often have mechanical pairing problems in the region of the inversion; these pairing problems reduce the frequency of crossing-over and hence the recombinant frequency in the region.

The net genetic effect of a pericentric inversion is the same as that of a paracentric one – crossover products are not recovered – but for different reasons. In a pericentric inversion, because the centromeres are contained within the inverted region, the chromosomes that have crossed over disjoin in the normal fashion, without the creation of a bridge. However, the crossover produces chromatids that contain a duplication and a deficiency for different parts of the chromosome. In this case, if a nucleus carrying a crossover chromosome is fertilized, the zygote dies because of its genetic imbalance. Again, the result is the selective recovery of noncrossover chromosomes in viable progeny. Thus inversions are referred to as crossover suppressors because crossover of inversion are not recovered.

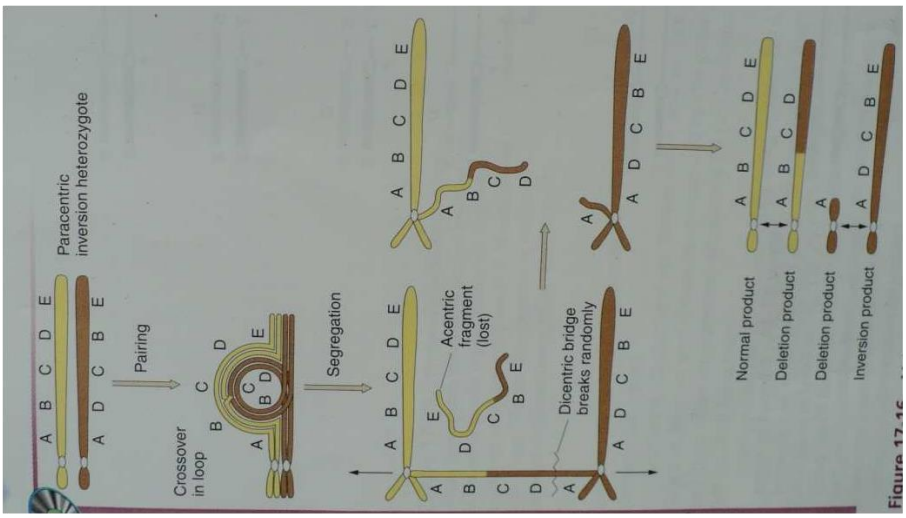

Figure 13: Synapsis in Paracentric Inversion Heterozygote

Why is inversion sometimes regarded as crossover suppressor?

Inversion is sometimes regarded as crossover suppressor because recombinant products are due to crossing over during meiosis, and these are not recovered because the offspring possessing them are non-viable.

Self-Assessment Exercise 1

Provide answers to the questions in 20 minutes

Identify the following chromosomal aberration found in cytological examination of the chromosomes of a plant species in a particular population:

(i) ABC.DE to ACB.DE

(ii) ABC.DE to ABD.CE

4.4. Summary

Inversion occurs when two breaks in a chromosome is followed by a 1800 rotation and rejoining of the inverted segment. The main diagnostic features of inversions are inversion loops, reduction of recombinant frequency, and reduced fertility from unbalanced or deleted meiotic products characteristic of inversion heterozygotes.

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 4.6. Possible Answers to Self-Assessment Exercises

SAE 1

(i) paracentric inversion

(ii) pericentric inversion

Unit 5: Translocation

CONTENTS

 5.1. Introduction

A translocation is a chromosomal mutation in which there is a change in position of chromosome segments and the gene sequences they contain. There is no gain or loss of genetic material involved in a translocation.

 5.2. Learning Outcomes

Student must have understood the process of translocation, and the genetic and cytological consequences of translocation.

 5.3. Translocation

Two simple kinds of translocations occur. One kind involves a change in position of a chromosome segment within the same chromosome: this is called an *intrachromosomal* (within a chromosome) translocation. The other kind involves the transfer of a chromosome segment from one chromosome into a nonhomologous chromosome; this is called an interchromosomal (between chromosomes) translocation. If this latter translocation involves the transfer of a segment in one direction form one chromosome to another, it is a nonreciprocal translocation; if it involves the exchange of segments between the two chromosomes it is a reciprocal translocation.

Differentiate between translocation heterozygote and translocation homozygote

Translocation heterozygote is an individual that carry a translocation on one of the homologous pair of chromosome.

Self-Assessment Exercise 1

Provide Answer to the following question in 10 minutes

Which of the following can easily be identified cytologically and why?

i. translocation heterozygote

ii. translocation homozygote

5.4. Consequence of Translocation

In organisms homozygous for the translocations, the genetic consequence is an alteration in the linkage relationships of genes. For example, in the nonreciprocal intrachromosomal translocation shown in figure, the BC segment has moved to the other chromosome arm and has become inserted between the F and G segments (Figure 14). As a result, genes in the F and G segments are now farther apart than they are in the normal strain, and genes in the A and D segments are now more closely linked. Similarly, in reciprocal translocations new linkage relationships are produced.

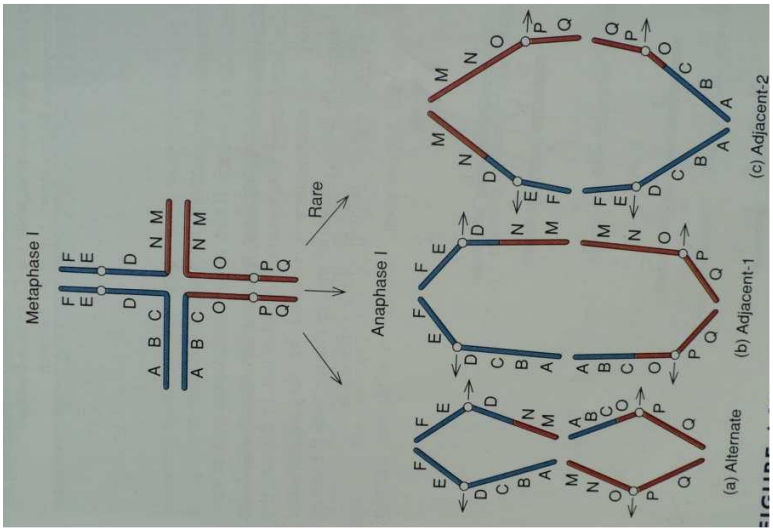

Figure 14: Synapsis in Translocation Heterozygote

How can you identify the other translocation cytologically?

Translocation homozygote can be identified by karyotyping where the translocation pair of homolog will be noticed to change morphologically.

Self-Assessment Exercise 2

Provide Answer to the following question in 10 minutes

Can translocation be harmful?

5.5. Summary

Translocation is when a part of a chromosome joins part of another chromosome. Translocation heterozygotes are usually identified cytologically because they form characteristic cross-like shape during synapsis in meiosis. Translocation affect fertility of organisms carrying them.

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5.7. Possible Answers to Self-Assessment Exercises

SAE 1

Translocation heterozygote can easily be identified because synapsis of the chromosome carrying translocation and its normal homolog produces a characteristic cross-like shape.

SAE 2

Translocations can either have beneficial (heterosis) or deleterious effects on reproductive fitness (outbreeding depression), that can change over time, and both effects can occur simultaneously

Glossary
DNA – Deoxyribonucleic acid
n - number
RF – Recombination frequency

End of Module Questions
1. Make a diagrammatic illustration of the different types of aneuploidy

Answer:

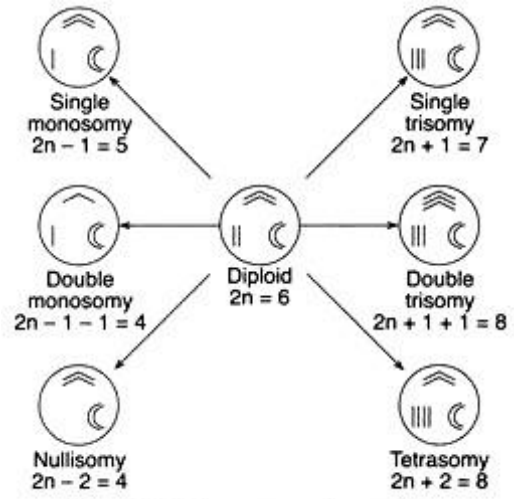

Fig. 11.2 : Different types of aneuploids

2. Make a tabular presentation of the common types of changes in chromosome number
Answer:

Type		Change in chromosome number	Symbol
	Heteroploid	Change from the 2n state	
A.	Aneuploid	One or a few chromosomes extra or missing from 2n	2n ± a few
	Nullisomic	On chromosome pair missing	2n-2
	Monosomic	One chromosome missing	2n-1
	Double monosomic	Two non homologous chromosome missing	2n-1-1
	Trisomic	One extra chromosome	2n=1
	Double trisomic	Two extra non homologous chromosome	2n+1+1
	Tetrasomic	One extra chromosome pair	2n+2
B.	Euploid	Number of genomes different from two	
	Monoploid	Only one genome present	x

	Haploid	Gametic chromosome number of the concerned species present	n
1.	Polyploid		
	Autopolyploid	More than two copies of the same genome present	
	Autotriploid	Three copies of the same genome	3x
	Autotetraploid	Four copies of the same genome	4x
	Autopentaploid	Five copies of the same genome	5x
	Autohexaploid	Six copies of the same genome	6x
	Autooctaploid	Eight copies of the same genome	8x
2	Allopolyploid	Two or more distinct genomes; each has two copies	
	Allotetraploid	Two distinct genomes; each has two copies	$(2x_1+2x_2)$
	Allohexaploid	Three distinct genomes; each has two copies	$(2x_1+2x_2+2x_3)$
	Allooctploid	Four distinct genomes; each has two copies	$(2x_1+2x_2+2x_3+2x_4)$