NATIONAL OPEN UNIVERSITY OF NIGERIA

SCHOOL OF SCIENCE AND TECHNOLOGY

COURSE CODE: BIO 402

COURSE TITLE: Cytogenetics of Plants

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MODULE 1

Unit 1: Definition and History of Cytogenetics

Content

1.0 Introduction

1.1 Definition of Cytogenetics

Cytogenetics is 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.

1.2 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). Jan ssen – 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**. H is 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 ferlization 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
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.

The first paper (1902) contained the earliest detailed demonstration that the somatic chromosomes of the lubber grasshopper, *Brachystola magna,* occur in definite distinshably 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.

4.0 Conclusion

Several contributors play significant roles in the development of cytogenetics as a discipline.

5.0 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.

6.0 Tutor Marked Assessment

Que: Define cytogenetics and outline the contributions of the following scientists to the development of cytogenetics as a discipline
(i) Walter Flemming (ii) Thomas Morgan (iii) Emil Heitz

Ans: 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.

Contributions

(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.

7.0 References/Further Reading

Bridges, C. 1931. Nondisjunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 2: Chromosome Theory of Inheritance

Content

7.0 References/Further Reading

1.0 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.

2.0 Objective

To understand the experiments of Theodore Boveri and other scientists in the development of chromosome theory of inheritance.

3.0 Main Content

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.

3.2 Edmund Beecher Wilson's Principle of Chromosome Theory of Inheritance

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

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.

4.0 Conclusion

The major conclusion derived from chromosome theory of inheritance is that genes are arranged in a linear fashion on chromosomes.

5.0 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.

6.0 Tutor Marked Assessment

Que: (i) What do you consider as the main idea of the chromosome theory of inheritance?

(ii) What was Thomas Morgan's contribution to the chromosome theory of inheritance?

Ans: (i) The main idea of the theory is that genes are arranged linearly on the chromosomes.
(ii) 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.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 3: Chromosome Packaging

Content

3.2 Second Level Packaging
3.3 Third Level Packaging
3.4 Fourth Level
4.0 Conclusion
5.0 Summary
6.0 Tutor Marked Assessment
7.0 References/Further Reading

1.0 Introduction

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.

2.0 Objectives

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

3.0 Main Contents

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 (~100A^0 diameter) on a string (~20A^0 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.

3.2 Second Level Packaging

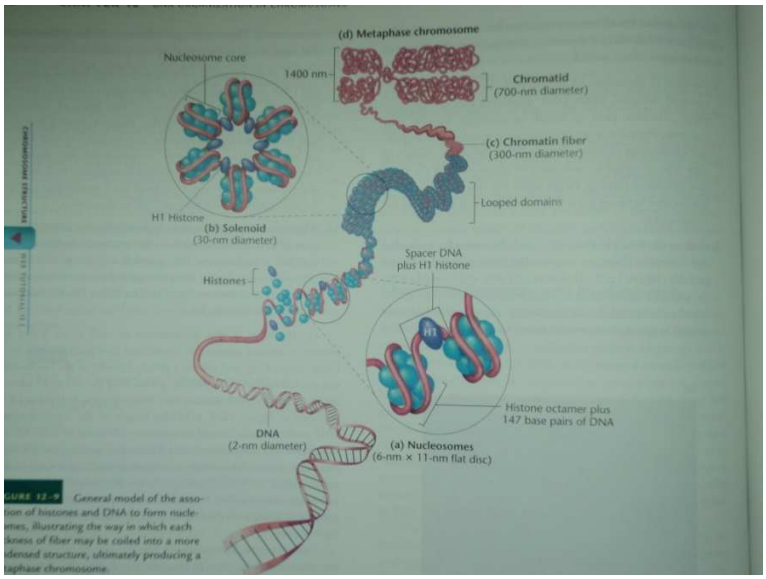
Further packaging of several (6?) 100A0-nucleosomes gives a solenoid of about 300A^0. This further reduces the chromosomes length by 1/6.

3.3 Third Level Packaging

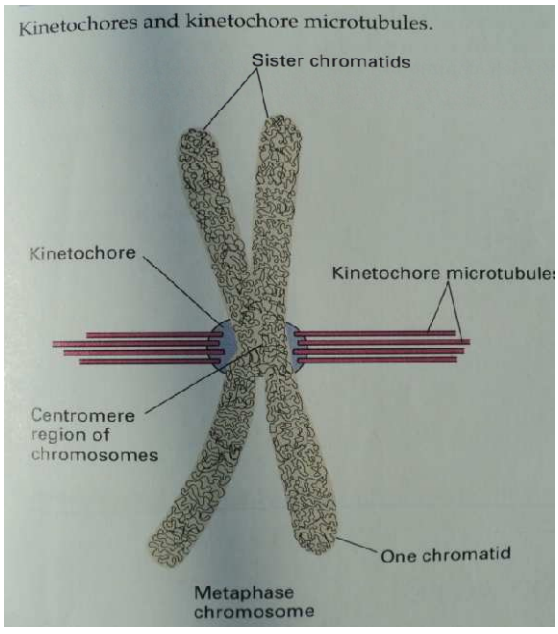
In transition to mitosis, the 300A^0 structure forms a series of looped domains of about 3,000A^0 in diameter.

3.4 Fourth Level

The condensed fibers are further coiled into chromosome arms (of about 7000A^0) 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.



Successive Levels of Chromosome Packaging



A Metaphase Chromosomes

4.0 Conclusion

The enormous eukaryotic DNA should be well packaged in order to be contained in the nucleus. Thus, the highly condensed chromosome seen in metaphase is as a result of coiling and recoiling of chromatin which is made up of DNA and histones.

5.0 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.

6.0 Tutor Marked Assessment

Que: (i) How is DNA organized Into chromatin?
(ii) What is the importance of chromosome packaging?

Ans: (i) DNA is negatively charged; it is therefore found associated with positively charged histones and other non-histone proteins to form the chromatin.
(ii) Chromosome packaging is important for proper chromosome movement and distribution during cell division.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 4: Chromosome Morphology

Content

1.0 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.

2.0 Objectives

To be able to describe chromosome morphology in terms of centromere location, chromosome sizes etc.

3.0 Main Content

3.1 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.

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 t he central CCG triplet within region III completely inactivates centromere function.

3.2 Chromosome Classification based on the number of centromeres

3.2.1 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 behavior. They are formed as a result of structural aberrations. Such chromosomes are more appropriately referred to as **acentric fragments**.

3.2.2 Monocentric Chromosomes

Chromosomes having one centromere per chromosome are known as monocentrics. This is the usual case. Thus, normal chromosomes are usually monocentric.

3.2.3. 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.

3.3 Chromosome Classification Based on Centromere Location

3.3.1 Centromere Located in the Middle of the Chromosome

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

3.3.2 Centromere Located near the Middle of the Chromosome

This is refer to as sub-median centromere, and the chromosome is called a submetacentric chromosome.

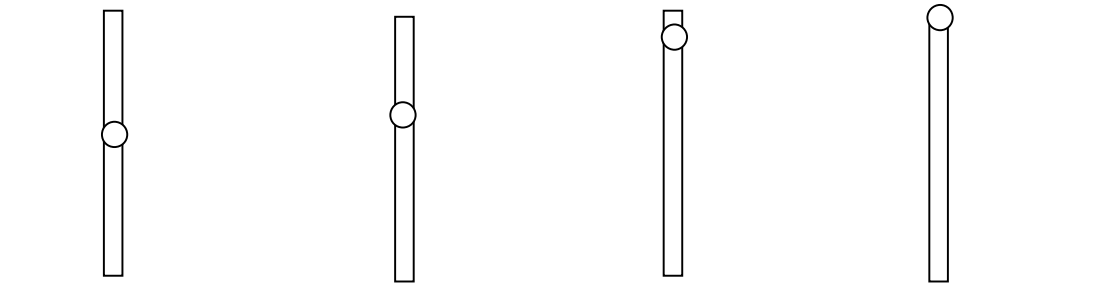
3.3.3 Centromere Located near the end of the Chromosome

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

3.3.4 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. Thus, they are rare in nature.



Metacentric chromosome | Sub-metacentric chromosome | Sub-elocentric chromosome | Telocentric chromosome

Figure showing different types of chromosomes

3.4 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

4.0 Conclusion

Chromosomes have characterized shaped based on different criteria. This is of immense important in agriculture and plant disease diagnoses.

5.0 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.

6.0 Tutor Marked Assessment

Que: Complet the Table

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

Ans:

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

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 5: Chromosome Classification Based On Size and Other Attributes

Content

1.0 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.

2.0 Objectives

(i) To know the types of chromosome based on size, presence of secondary constriction
(ii) To know the functions of satellite chromosomes
(iii) To know what heterochromatin and euchromatin are.

3.0 Main Content

3.1 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 idiograms to demonstrate such characteristics. The total chromosomal complement of a cell is refer to as the karyotype. The complement can be photographed during mitosis and rearranged in pairs to produce a picture refer to as karyotype. An ideogram is a digramatic 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.

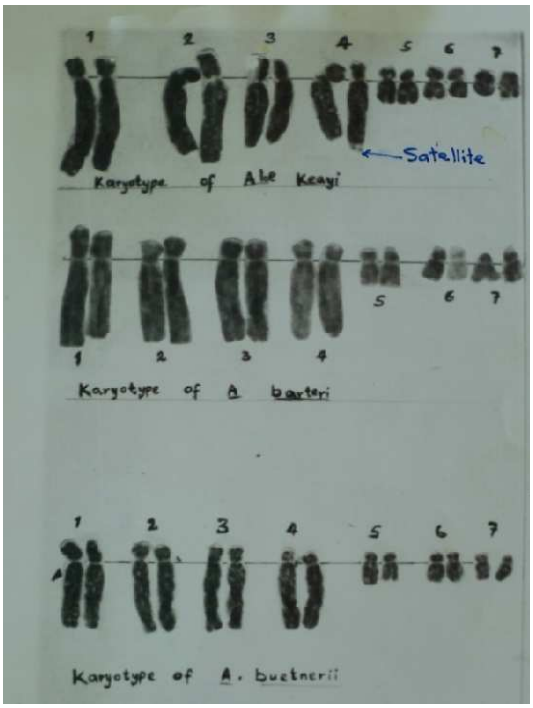


Figure Showing Karyotype

3.2 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 t hat appears removed is called satellite. Thus NOC can also be called satellite chromosome. Large satellites can possess a separate constriction and are called tandem satellite.

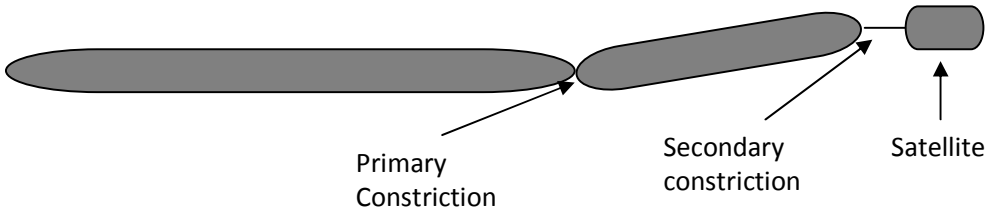


Figure showing satellite Chromosome

3.2.1 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.

3.3 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.

3.4 Telomere

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

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

4.0 Conclusion

Chromosomes can be classified and identified on the basis of size. Presence of secondary is another important attributes of chromosomes.

5.0 Summary

Chromosomes can be classified as long, medium and small based on their sizes. Apart from the cenromere (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.

6.0 Tutor Marked Assessment

Que: (1) Distinguish between heterochromatin and euchromatin.
(2) What are telomeres? What are their cytogenetic importance?

Ans: (1) Heterochromatin stains darkly, and they are genetically inactive while euchromatin stains lightly and are genetically active.

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

Importance of telomeres:
(1) They prevent one chromosome joining the other.
(2) They help proper replication of chromosome ends in order to prevent shortening of chromosomes due to replication.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

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: Variations in Chromosome Number: An Overview

Content

1.0 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.

2.0 Objective

To understand the nature and consequence of variation in chromosome numbers.

3.0 Main Content

3.1 Aneuploidy and Euploidy

In aneuploidy, an organism has gained or lost one or m ore 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 8.1 provides an organizational framework for you to follow.

Table 1.1: Terminology for Variation in Chromosome Numbers

Term	Explanation
Aneuploidy	2n $\pm$ x; chromosomes
Monosomy	2n – 1
Disomy	2n
Trisomy	2n + 1
Tetrasomy, pentasomy, etc	2n +2, 2n + 3, etc
Euploidy	Multiples of n
Diploidy	2n
Polyploidy	3n, 4n, 5n
Triploidy	3n
Tetraploidy, pentaploidy etc	4n, 5n, etc
Autopolyploidy	Multiples of the same genome
Allopolyploidy (Amphidiploidy)	Multiples of closely related genomes

3.2 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.

3.3 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.

3.4 Genome Formula

This is away 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.

4.0 Conclusion

Mutation can occur at the chromosomal level in the form of loss or gain of chromosome. Such mutation affects the chromosome complement of the organism.

5.0 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.

6.0 Tutor Marked Assessment

Ques: You are given the following chromosome complement for a plant with chromosome number 2n = 4:

aa
bb
cc
dd

Give the chromosome complement and the chromosome number of the following aneuploids:

(i) A trisomic for chromosome
(ii) a double nullisomic for chromosomes b and d.
(iii) A monosomic for chromosome a.

Ans: A trisomic for chromosome c

aa
bb
ccc
dd 2n = 9

A double mullisomic for chromosome b and c
aa
cc
A monosomic for chromosome a
a
bb
cc
dd

7.0 References/Further Reading

Swanson C.P.; Mertz T.; Young W.J. 1981. Cytogenetics: The Chromosome in Division, Inheritance and Evolution. Englewood Cliffs, N.J. Prentice-Hall.

Weaver RF; Hedrick PH. 1997. Genetics 3rd EEd. W.C.B. Publishers.

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Unit 2: Monoploidy

Content

1.0 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.

2.0 Objective

(i) To know what monoploids are
(ii) To understand how monoploids arise
(iii) To know the difference between monoploidy and haploidy

3.0 Main Content

3.1 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

3.2 Production of Monoploids

3.2.1 Interspecific and Intergeneric Hybridization

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

(b) Irradiation and Chemical Treatment

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

3.2.2 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.

3.2.3 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.

3.2.4 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.

3.3 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 th at chromosoma had been duplicated at the S phase.

3.4 Fertility in Monoploids

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.

4.0 Conclusion

Some organisms occur naturally as monoploids although artificial monoploids can be created through various techniques.

5.0 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.

6.0 Tutor Marked Assessment

Que: (i) Differentiate between the terms monoploid and haploid numbers

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

Ans: (i) 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.

(ii) 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 = $(^1/_2)^{x-1}$.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 3: Diploidy and Introduction to Polyploidy

Content

1.0 Introduction

Diploids especially and monoploids are cases of normal euplodiy. 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), petaploids (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.

2.0 Objective

(i) To appreciate that diploids and sometimes monoploids are cases of normal euploidy.

(ii) To appreciate the difference between diploidy and polyploidy in termsof their level of ploidy.

3.0 Main Content

3.1 Diploidy

This is when an organism have 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.

3.2 Meiotic Behavior and Fertility in Diploids

Normal bivalents are formed at meiosis and therefore balanced gametes are produced.

4.0 Conclusion

Most plant species and of course animal species occur as diploids and sometimes monoploids. Thus diploids and monoploids are regarded as cases of

normal euploidy. Organisms with higher ploidy level (i.e. polyploids) are, therefore, cases of abnormal euploidy.

5.0 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.

6.0 Tutor Marked Assessment

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

Ans: 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.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 4: Triploidy

Content

1.0 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.

2.0 Objectives

(i) To know that triploids are the first set of polyploids.
(ii) To know the differences between autopolyploids and allopolyploids.
(iii) To know that triploids have improved qualities.
(iv) To know why triploids are sterile.

3.0 Main Content

3.1 Autopolyploids and Allopolyploids

In the real of polyploids, we must distinguish between autopolyploids and allopolyploids. Autopolyploids are composed of multiple sets from within oen 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.

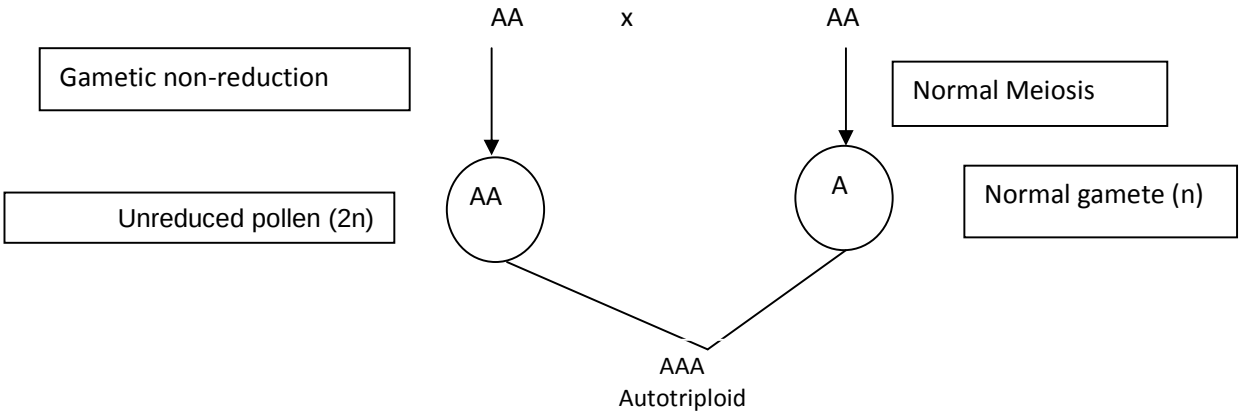
Important: Triploids are usually autotriploids.

When we consider allopolyploidy, the degree of unrelatedness matters. Genome allopolyploids contain clearly different basic genomes derived frok 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 AAA_1.

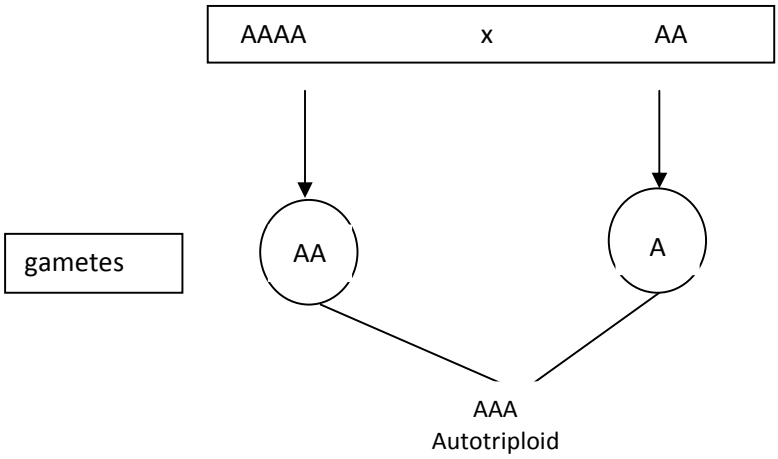
3.2 Production of Triploids

3.2.1 Genetic Non-Reduction

This is occurs when a normal gamete is fertilized by an unreduced pollen.



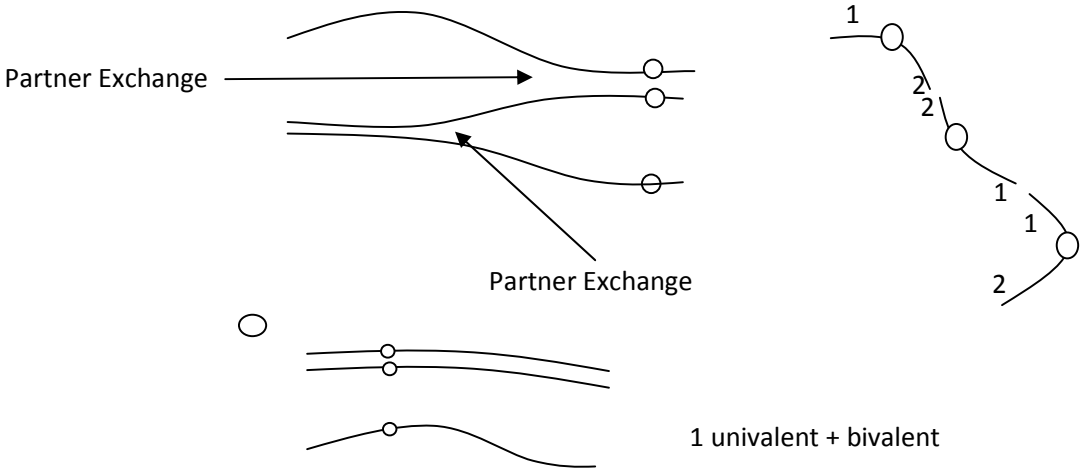
3.2.2 Crossing between tetraploids and diploids



3.3 Fertility in Triploids

3.3.1 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 below:



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.

3.3.2 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 $(^1/_2)^{x-1}$.

3.4 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

(1) Disturbance of Sex Determination Mechanism
The XY sex determination mechanisms is upset in animals when chromosome sets are duplicated.

(2) 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.

(3) Cross Fertilization Barrier
Interspectific cross fertilization is rare in animals but common in plants. Moreover, hybrids are developmentally defective.

(4) 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.

3.5 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 cou9nterparts.
- 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.

4.0 Conclusion

Although triploids have improved qualities over their diploid counterparts, they are, however, sterile in view of their meiotic irregularities.

5.0 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.

6.0 Tutor Marked Assessment

Que: Why is it that banana cannot be propagated by seed.

Ans: 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.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 5: Tetraploidy and Higher Polyploidy

Content

4.0 Conclusion
5.0 Summary
6.0 Tutor Marked Assessment
7.0 References/Further Reading

1.0 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.

2.0 Objectives

(i) To know that tetraploidy occurs well in plants.
(ii) To know the chromosome complement and genome formulas of different types of autotetraploids.
(iii) To understand that tetraploids have significantly higher fertility than triploids.

3.0 Main Content

3.1 Autotetraploids

Autotetraploids have 4x chromosome complement with genome formula AAAA.

3.1.1 Production of Autotetraploids

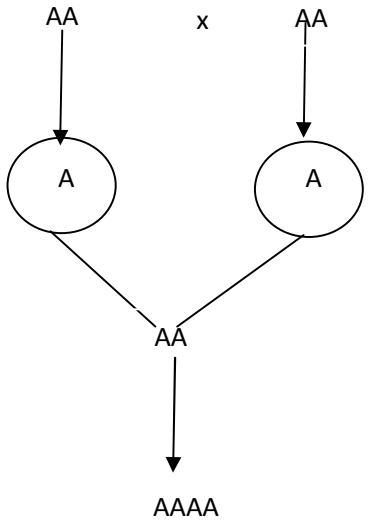
Production involves 2 mechanisms:

(a) Genetic non-reduction and

(b) Somatic doubling

(a) Genetic Non-reduction (Refer to production of autotriploids)

(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.

3.1.2 Fertility in Autotetraploid

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 tetraploidds, 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.

3.2 Genetics of a Fertile Autotetraploid

3.2.1 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 liked 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.

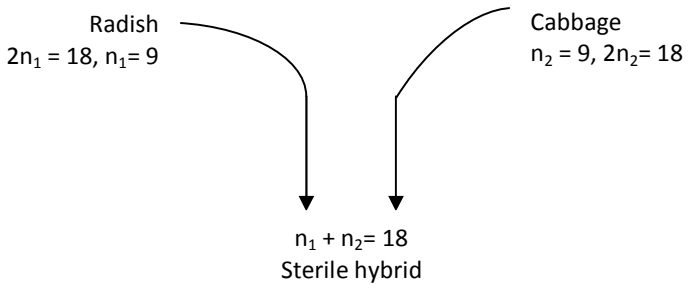
The packaging of genes two at a time into games is very much like grabbing two balls at random from a bag of eight balls: 4 of one kind, 4 of another. The probability of picking two b genes = probability of bb gamete.
= 4/8 (the first one) x 3/7 (the 2nd one) = 3/14

∴ b/b/b/b = $(^3/_{14})^2 = {}^9/_{196}$

3.3 Allotetraploids

3.3.1 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:



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 $2n_1 + 2n_2$ in the sterile hybrid, presumably in tissue that eventually became germinal and underwent meiosis. Thus, in $2n_1 + 2n_2$ tissue, there is a pairing partner for each chromosome and balanced gametes of the type $n_1 + n_2$ are produced. These gametes fuse to give $2n_1 + 2n_2$ allopolyploid progeny, which also are fertile. This kind of allopolyploid is sometimes called an amphidiploids, which means "doubled diploid" (Fig.). (Unfortunately for Ka rpechenko, 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 $2n_1 + n_2$, constituted from an $n_1 + n_2$ gamete from the allopolyploid and an n1 gamete from the radish. The n_2 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.*

3.3.2 Production of Allotetraploid

It is produced by hybridization of 2 different species to yield an infertile hybrid in F_1. F_1 must be able to propagate vegetatively before chromosome doubling to produce a vigorous fertile hybrid.

3.3.2.1 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 shows the procedure for synthesizing Triticale.

4.0 Conclusion

Autotetraploids and allotetraploids can occur spontaneous or artificially in plant breeding. Tetraploids are usually produ ced in plant breeding because of their commercial and other economic advantages over their diploid counterparts.

5.0 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 chromosome 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 .

6.0 Tutor Marked Assessment

Que: 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

Ans: 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.

7.0 Reference/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

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

Unit 1: Aneuploidy: An Overview

Content

1.0 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 o fchromosomes. 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**.

2.0 Objectives

(i) To be able to distinguish clearly aneuploidy and euploidy.
(ii) To know the causes of aneuploidy
(iii) To understand the concept of genic balance and how it affects aneuploids and euploids.

3.0 Main Content

3.1 Types of Aneuploidy

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

Class exercise

More Practice: Describe a triple trisomic .

3.2 Aneuploid Series

Aneuploid series refers to all possible aneuploids. 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.

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

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

3.3 Causes of Aneuploidy

1) 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.

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

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

4) Multipolar Mitosis: Abnormal chromosome distribution.

3.4 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.

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.

4.0 Conclusion

Aneuploidy is a numerical variation that affects a particular chromosome and not the whole set. Aneuploids are produced by nondisjunction or some other types of chromosome misdivision at either meiosis or mitosis. Aneuploidy causes alteration of delicate genetic balance that enables normal development.

5.0 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.

6.0 Tutor-Marked Assessment

Que: (i) How do you think multipolar mitosis might cause aneuploidy?

(ii) Explain why aneuploidy usually has more deleterious effect than polyploidy?

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

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

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

(Note: there are 7 types of any aneuploid series)

7.0 Reference/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 2: Aneuploidy and its Genetic Consequences

Content

1.0 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.

2.0 Objectives

(i) To understand other genetic consequences of aneuploidy apart from genic imbalance.

(ii) To know the effect of aneuploidy on gamete production and inheritance.

3.0 Main Content

3.1 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; they are illustrated in Figure . Their appearances differ from the normal hexaploids; furthermore, most of the nullisomics grow less vigorously.

3.2 Monosomics (2n-1)

Monosomic chromosome complements are generally deleterious for two main reasons. First, the missing chromosome perturbs the ovrall 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.

3.3 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, trisomimcs 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.

3.4 Genetic Analysis of Trisomics for Allelic Constitution

The method is similar to that of triploids.

4.0 Conclusion

The adverse effect of genic imbalance characteristic of aneuploidy not only affect the organism but also gametes. Aneuploid gametes have lower fertilizing efficiency and therefore causes reduced fertility.

5.0 Summary

Nullisomy (2n – 2)

Nullisomy is lethal in diploids, amphidiploids (allopolyploids) survive as a result of chromosome compensation.

Monosomy (2n-1)

(1) Monosomic show the deleterious effect of genome imbalance

(2) They are hemizygous for deleterious recessive alleles on the monosomic chromosome.

Trisomy (2n+1)

(1) Trisomics show the deleterious effect of genome imbalance

(2) However, unlike in monosomy, deleterious recessive alleles are masked by the dominant alleles on the trisomic chromosome.

6.0 Tutor Marked Assessment

Que: (i) Why is aneuploidy of more deleterious conseq uence than euploidy?

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

Ans: (i) 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.

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

7.0 Reference/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 3: Variations in Chromosome Structure

Content

4.0 Conclusion
5.0 Summary
6.0 Tutor Marked Assessment
7.0 References/Further Reading

1.0 Introduction

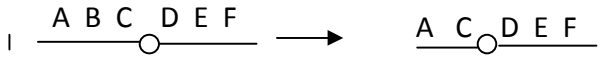
Variation in chromosome structure involve changes in parts of chromosomes rather than changes in the number of chromosomes or sets of chromosomes in a genome. There are four types of such mutations: *deletions* and *duplications* (both of which involve a change in the amounts of DNA on a chromosome), *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.

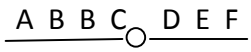
1.1 Types of Change

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.

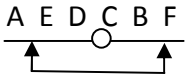
The simple loss of a chromosomal segment is called a **deletion** or **deficiency.** In the following diagram, region B has been deleted:



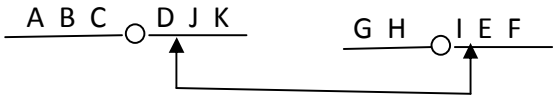
The presence of two copies of a chromosomal region is called a **duplication**:



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



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



2.0 Objectives

(i) To understand the process of deletion and duplication.

(ii) To know the cytological and genetic consequences of deletion and duplication.

3.0 Main Content

3.1 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 contromere, 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 rearrangmenets 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.

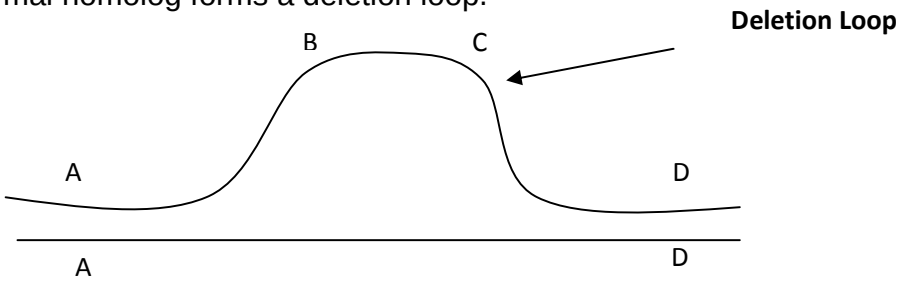
The effects of deletions depend on their5 size. A small deletion within a gene, called an **intragenic deletion,** inactivates the gene and has the same effect as other null mutations of that gene. If the homozygous null phenotype is viable (as, for example, in human albinism), then the homozygous deletion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

3.1.1 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 cyclogenetic analysis. 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

3.2 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

3.2.1 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.

4.0 Conclusion

The lethality of heterozygous deletions can be explained by genome imbalance and by unmasking of recessive lethal alleles. Deletions are recognized cytologically by deletion loop. Duplications supply additional genetic material capable of evolving new functions.

5.0 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:

(1) deletion, in which a chromosome segment is lost.

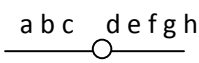
(2) duplication, in which more copies of a chromosome segment are present than in the normal state.

(3) Inversion, in which the orientation of a chromosome segment is opposite that of the wild type and

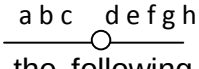
(4) translocation, in which a chromosome segment has moved to a new location in the genome. The consequences of these structural mutations depend on the specific mutation involved.

6.0 Tutor Marked Assessment

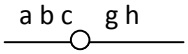
Que: A normal chromosome has the following gene sequence (0 = centromere).

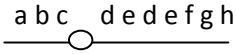


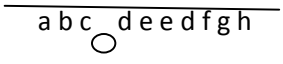
-irradiation of the organism produced the following types of chromosomes

Que: A normal chromosome has the following gene sequence 

(0 = centromere). X-irradiation of the organism produced the following types of chromosomes:

(i) 

(ii) 

(iii) 

(a) Name the type of chromosomal aberration that produced each type of chromosome.

(b) Diagram synapsis with the wild type chromosome in a structural heterozygote for the chromosome in (i) above

Ans: (a) (i) Deletion (or deficiency)
(ii) Tandem duplication
(iii) Reverse duplication

(b) See diagram for synapsis in Section 3.1.1. ensure that the deletion loop of the wild type homolog is at the deleted region 'def'

7.0 Reference/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 4: Inversion

Content

1.0 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.

2.0 Objectives

(i) To understand the process and types of inversion
(ii) To know the genetic and cytological consequences of inversion.

3.0 Main Content

3.1 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.).

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 spaning the centromere are **pericentric**:

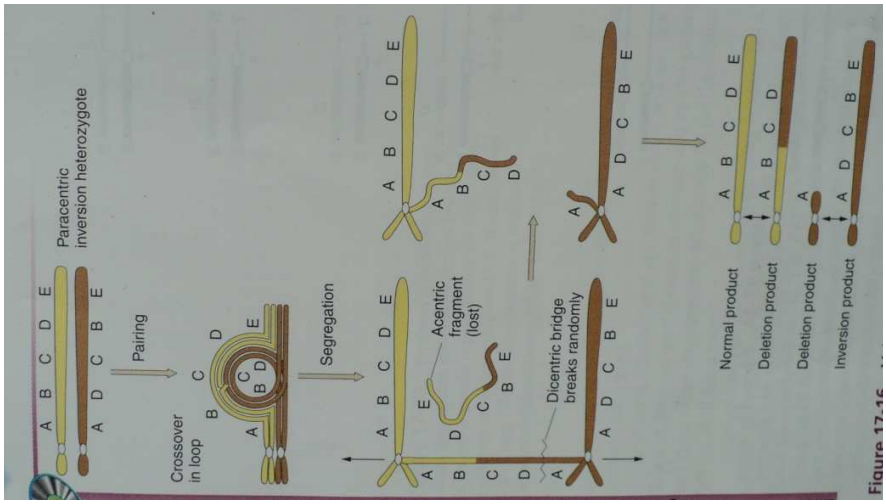
How do inversion behave genetically? Crossing-over within the inversion loop of a paracentric inversion connects homologous centromeres in a **dicentric bridge** while also producing an **acentric fragment** - a fragment without a centromere. Then, as the chromosomes separate in anaphaseI, the centromeres 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.). The gametes containing such deleted chromosomes may be inviable but, even if viable, the zygotes that they eventually form are inviable. 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 disjoion in the normal fashion, without the creaton 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 imbaolnace. Again, the result

is the selective recovery of noncrossover chromosomes in viable progeny. Thus, inversions are referred to as crossover suppressors because crossover products of inversion are not recovered.

Synapsis in Paracentric Inversion Heterozygote



4.0 Conclusion

The mechanisms reduce the number of recombinant product among the progeny of inversion heterozygote: elimination of the products of crossovers in the inversion loop and inhibition of pairing in the region of the inversion.

5.0 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.

6.0 Tutor Marked Assessment

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

(a) (i) ABC.DE to ACB.DE (ii) ABC.DE to ABD.CE

(b) Why is inversion sometimes regarded as crossover suppressor?

Ans: (a) (i) parcentric inversion (ii) pericentric inversion

(b) 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.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.

Unit 5: Translocation

Content

1.0 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.

2.0 Objectives

(1) To understand the process of translocation.

(2) To know the genetic and cytological consequences of translocation.

3.0 Main Content

3.1 Types of Translocation

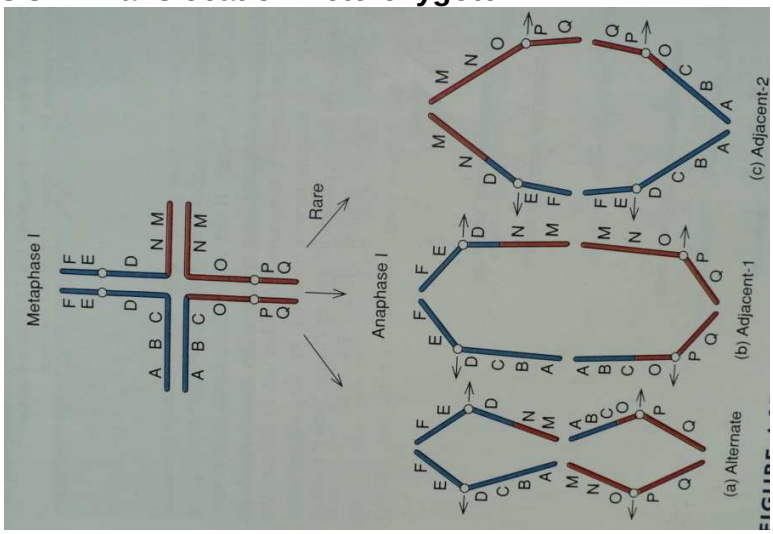
Two simple kinds of translocations occur. One kind involves a change in position of a chromosome segment within the same chromosome: this is called an *intrachromosomal* (within a chromosome) translocation. The other kind involves the transfer of a chromosome segment from one chromosome into a nonhomologous 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.

3.2 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. 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.

Synapsis in Translocation Heterozygote



4.0 Conclusion

Reciprocal translocations are diagnosed genetically by semisteriity and by the apparent linkage of genes known to be on separate chromosome.

5.0 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.

6.0 Tutor Marked Assessment

Que: (a) Differentiate between traslocation heterozygote and translocation homozygote.

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

(c) How can you identify the other translocation cytologically?

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

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

(c) translocation homozygote can be identified by caryotyping where the translocation pair of homolog will be noticed to change morphologically.

7.0 References/Further Reading

Bridges, C. 1931. Nondijunction as proof of the chromosome theory of heredity. *Genetics*. **1**: 1-52; 107-163.

Mcintosh JR; McDonald KL. 1989. The Mitotic Spindle. *Scientific American.* **261**: 48-56.