Course: Fundamentals of Genetics
Class: - Ist Year, IInd Semester
Lecture No. XXI
Title of topic: - Chromosomal Aberrations (Numerical)
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Variation in Chromosome number

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

Aneuploidy

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

Types of Aneuploids

Types Genomic constitution

Monosomic	2n-1
Double monosomic	2n-1-1
Nullisomic	2n-2
Trisomic	2n+1
Double trisomic	2n+1+1
Tetrasomic	2n+2
Pentasomic	2n+3

1. Monosomics

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

2. Nullisomics

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

3. Polysomics

An individual having either single or one pair of extra chromosome in the diploid complement is known as polysomics. Polysomics are called as hyperploids.

Polysomics are of two types (i) trisomics and (ii) tetrasomics.

(i) Trisomics

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

(ii) Tetrasomics

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

Use of aneuploidy

i. Aneploids are extremely useful in several genetic studies.

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

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

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

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

Aneuploids in Human beings

i. Down's syndrome

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

ii. Kline felters syndrome (44+XXY)

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

iii. Turners syndrome

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

The origin of aneuploids

i. Spontaneous

ii. Meiotic irregularities

iii. Triploid individuals

iv. Translocation heterozygote

Use of aneuploids in crop improvement

a. Aneuploids are useful tools for locating the genes on a specific chromosome. Monosomics and nullisomics are used for this prupose.

b. Monosomics are also used in interspecific gene transfer *ie* monosomics are used in transferring chromosomes with a desirable genes from one species to another.

c. Aneuploids are used for developing alien addition and alien substitution lines in various crops.

d. Primary trisomics are useful in identification of chromosomes involved in translocations.

Polyploidy

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

Types	**Genomic formula**
Monoploids	n
Diploid	2n

Triploid	3n
Tetraploid	4n
Pentaploid	5n
Hexaploid	6n

Monoploid

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

Differences between monoploids and haploids:

Monoploids	**Haploids**
1. Represent gametic chromosome number of a diploid species	1. Represent gametic chromosome number of any species
2. Denoted by 'x'	2. Denoted by 'n'
3. Monoploids are always haploids	3. Haploids cannot always be monoploids
4. Contain single set of genome	4. May contain one or more copies of genome.

Diploids

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

Polyploids

Polyploids refer to any organism in which the number of chromosome sets exceeds two *i.e.*, an organism with more than two set of chromosomes or genome.

They have larger cells than diploids. These larger cell sizes contribute to larger plant size and higher yield. Polyploids have generally larger, thicker and darker green leaves bigger flowrf, fruits than the diploids. In each genus, there is an optimum level of polyploidy beyond which growth may be depressed with increasing number of chromosomes. (eg) triploid (3n).

There are two types of Polyploids.

1. Autopolyploid

In autoployploids the multiple sets of chromosomes are identical (eg). Genome is identical with each other.

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

Auto triploid - 3x

Auto tetraploid- 4x

Auto hexaploid- 6x

(eg). Banana 2n=3 x=33, Groundnut 2n=4x=40; Sweet potato 2n=6x=90 and

Potato 2n=4x=48

Autotriploid

The triploid organisms have three sets of chromosomes. A triploid may originate by the union of a monoploid gamete (n) with a diploid gamete (2n). Since an autotriploid remains sterile and cannot produce seeds, it has great commercial value in producing seedless varieties of economic plantgs. Eg. Seedless water melon.

Autotetraploid:

The organisms with four genomes (4n) in the nuclei of their somatic cells are called tetraploids. They arise due to somatic doubling of chromosome number. Doubling is

accomplished by either spontaneously or it can be induced by chemicals such as colchicines.

2. Allopolyploid

A species or types of plant derived from doubling the chromosomes in the F1 hybrid of two species, is called an amphidiploids. In amphidiploids the two species are knonw.

Eg. Gossyplum hirsutum - 2n=4x=52
G.barbadense - 2n=4x=52
Nicotiana tabacum - 2n=4x=48
Triticum aestivum - 2n=6x=42
Saccharum officinarum - 2n=8x=80

Morphological and Cytological features of polyploids are

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

Role of polyploids and their evaluation

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

Role of polyplodis in evolution of crops

Wheat:

Triticum monococcum X Unknown diploid species
2n = 14(diploid) 2n = 14(diploid)
AA BB

↓ Spontaneous chromosome doubling of F_1

Triticum turgidum X *Triticum tauschii*
2n = 28 (allotetraploid) 2n = 14(diploid)
AABB DD

↓ Spontaneous chromosome doubling of F_1

Triticum aestivum
2n = 42 (allohexaploid)
AABBDD

Cotton:

Gossypium herbaceum X *Gossypium raimondii*
Var. *Africanum*
(Old world cotton) (American cotton)
2n = 26 (diploid) 2n = 26 (diploid)
13 small bivalents 13 large bivalents
AA DD

↓

F_1 hybrids (highly sterile)

2n = 26
26 (13 small + 13 large) univalents (diploid)
AD
↓ diploidization
(Treated with colchicine)

Gossypium hirsutum,
(New world cotton)
2n = 52 (tetraploid)
26 (13 small + 13 large) bivalents
AADD

Triticale: Triticale, a man made cereal, is first produced by Muntzing. Triticale is a new crop species synthesized by crossing wheat and rye (*Secale cereale)*.

a) Some triticales are hexaploids and are developed from a cross between tetraploid wheat (*Triticum turgidum)* and rye.

Wheat	X	Rye
Triticum turgidum	X	*Secale cereale*
2n = 28 (Tetraploid)		2n = 14 (diploid)
AABB		RR

↓

F_1 Hybrid
2n = 21 (allotriploid)
ABR (sterile)
↓ doubling of chromosomes

Triticale hexaploide
2n = 42 (allohexaploid)
AA BB RR

Effects of polyploidy:

1. Genetical effects: The polyploidy often results in sterility. For example, an extra set of chromosomes in case of triploids is distributed in various combinations resulting in genetically imbalanced gametes.

2. Phenotypic effects: Most usual phenotypic effect of polyploidy is gigantism in morphology of plants. Eg: The tetraploid plants may have large sized pollen grains, cells of leaves, stomata, xylem etc. than a normal diploid plant. They are also more vigorous. As a result, large sized fruits, seeds and flowers are obtained from economically important plants.

3. Physiological effects: The ascorbic acid content has been reported to be higher in tetraploid cabbage and tomato than in corresponding normal diploid species. Corn flour of tetraploid maize has been found to contain 40% more vitamin A than that of normal diploid species.