

Session-14

Aneuploids can have a chromosome number either greater or smaller than that of the wild type. Hypo-ploidy may be due to loss of a single chromosome – monosomy ($2n - 1$), or due to loss of one pair of chromosomes – nullisomy ($2n - 2$). Similarly, hyper-ploidy may involve addition of either a single chromosome-trisomy ($2n + 1$) or a pair of chromosomes ($2n + 2$) – tetrasomy (Fig. 11.1).

Nondisjunction in mitosis or meiosis is the cause of most aneuploids. Disjunction is the normal separation of homologous chromosomes or chromatids to opposite poles at nuclear division. Nondisjunction is a failure of this disjoining process, and two chromosomes (or chromatids) go to one pole and none to the other. Nondisjunction occurs spontaneously; it is another example of a chance failure of a basic cellular process.

In meiotic nondisjunction, the chromosomes may fail to disjoin at either the first or second division. Either way, $n + 1$ and $n - 1$ gametes are produced. If an $n - 1$ gamete is fertilized by an n gamete, a monosomic ($2n - 1$) zygote is produced. The fusion of an $n + 1$ and an n gamete yields a trisomic $2n + 1$.

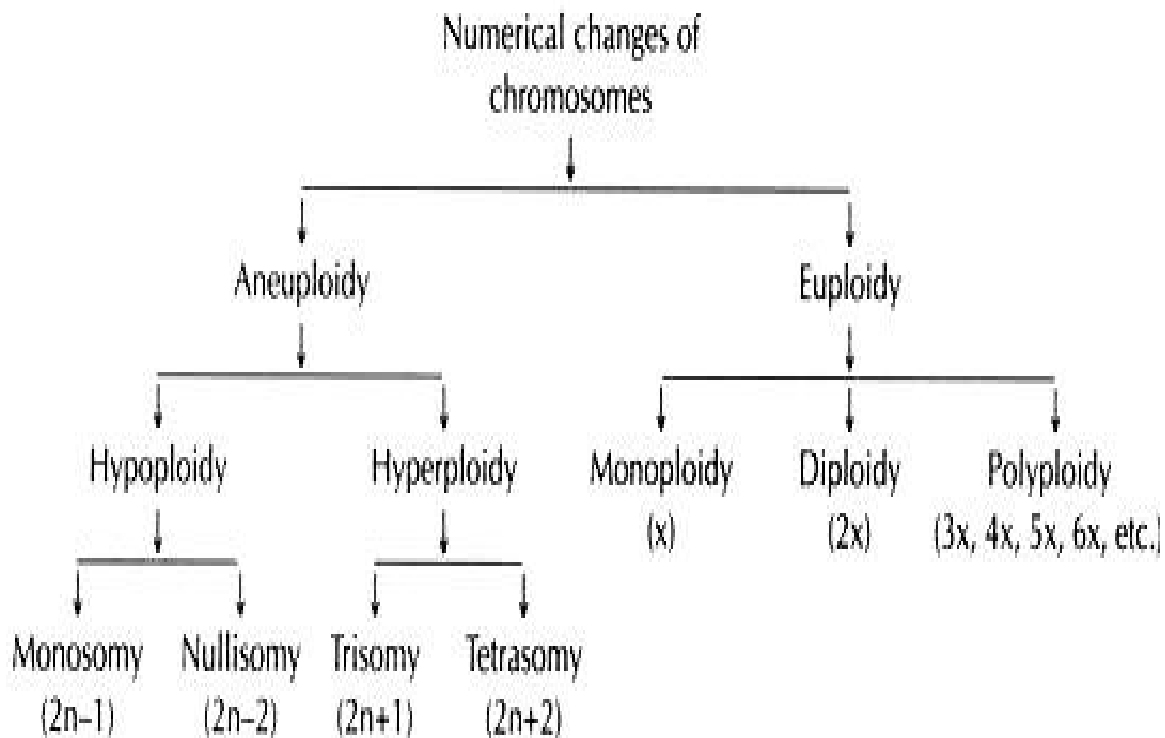


Fig. 11.1: Different kinds of numerical changes of chromosomes (x = basic chromosome number, $2n$ = somatic chromosome number)

Table 11.1: Common types of changes in chromosome number

Type	Change in chromosome number	Symbol
Heteroploid	Change from the $2n$ state	
A. Aneuploid	One or a few chromosomes extra or missing from $2n$	$2n \pm \text{a few}$
Nullisomic	One chromosome pair missing	$2n - 2$
Monosomic	One chromosome missing	$2n - 1$
Double monosomic	Two nonhomologous chromosomes missing	$2n - 1 - 1$
Trisomic	One extra chromosome	$2n + 1$
Double trisomic	Two extra nonhomologous chromosomes	$2n + 1 + 1$
Tetrasomic	One extra chromosome pair	$2n + 2$
B. Euploid	Number of genomes different from two	
Monoploid	Only one genome present	x
Haploid	Gametic chromosome number of the concerned species present	n
Polyploid		
1. Autopolyploid	More than two copies of the same genome present	
Autotriploid	Three copies of the same genome	$3x$
Autotetraploid	Four copies of the same genome	$4x$
Autopentaploid	Five copies of the same genome	$5x$
Autohexaploid	Six copies of the same genome	$6x$
Autooctaploid	Eight copies of the same genome	$8x$
2. Allopolyploid	Two or more distinct genomes; each genome has two copies	
Allotetraploid	Two distinct genomes; each has two copies	$(2x_1 + 2x_2)$
Allohexaploid	Three distinct genomes; each has two copies	$(2x_1 + 2x_2 + 2x_3)$
Allooctaploid	Four distinct genomes; each has two copies	$(2x_1 + 2x_2 + 2x_3 + 2x_4)$

Balanced Tertiary Trisomics (BTT)

Balanced Tertiary Trisomics (BTT) described by Ramage in 1965 provide an alternative method for the continuous availability of male sterile line to be used as female parent for commercial hybrid seed production in Barley. A Balanced Tertiary Trisomic (BTT) is a heterozygous tertiary trisomic has genotype aaA with dominant male fertile allele A or M that is located on the extra translocated chromosome while the recessive allele 'a', also called male sterile allele 'ms' on the two normal chromosomes). BTT produces three types of functional female gametes and only one type of male gamete. This is because male gametes having extrachromosome are inviable and hence cannot be transmitted through pollen, but only by egg.

Selfing of BTT produces three kinds of progeny:

- 1) 70% homozygous recessive (aa or $msms$) diploids that are used as female parents in hybrid production.
- 2) 30% heterozygous (aaA or $msmsMs$) balanced tertiary trisomic. These are vigorous, male fertile and produce abundant pollen for effective wind or insect pollination.
- 3) 1.0% homozygous recessive primary trisomic. These are in less in proportion.

Using BTT, the first hybrid variety named "Hembar" was produced by Ramage and Weibe in 1969. Hembar was produced by crossing a commercial inbred cultivar 'Arivat' onto GENETICRECESSIVE MALE STERILE DIPLOID PLANTS. Because of problems of cross-fertilization, susceptibility of hybrid to ergot infection and the complexity of the system, BTT have not proved practical for implementing hybrid barley on a commercial scale.

Balanced Tertiary Trisomics (BTT) Parent



$\frac{3}{4}$ viable Female gametes &

$\frac{1}{4}$ viable Male gametes

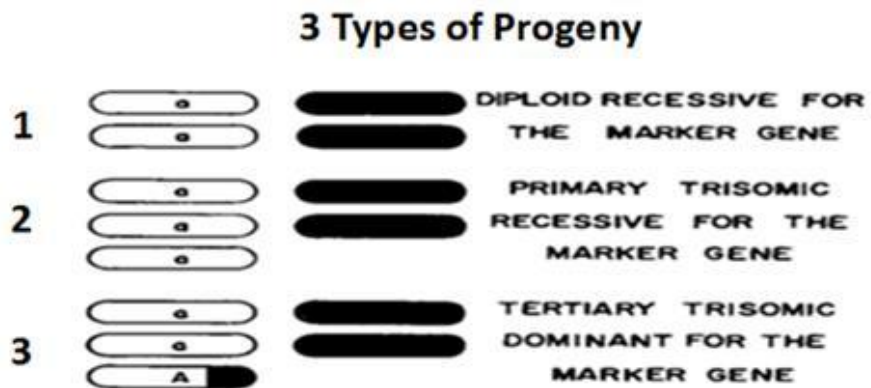
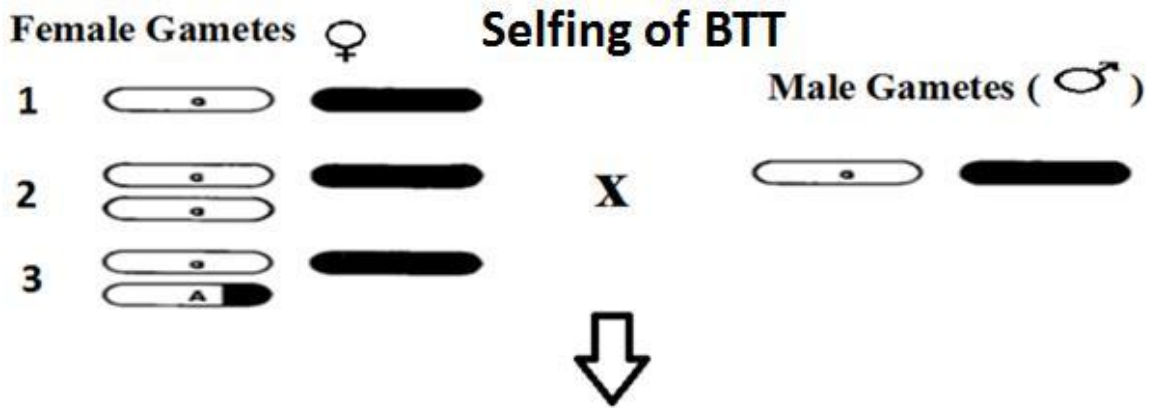
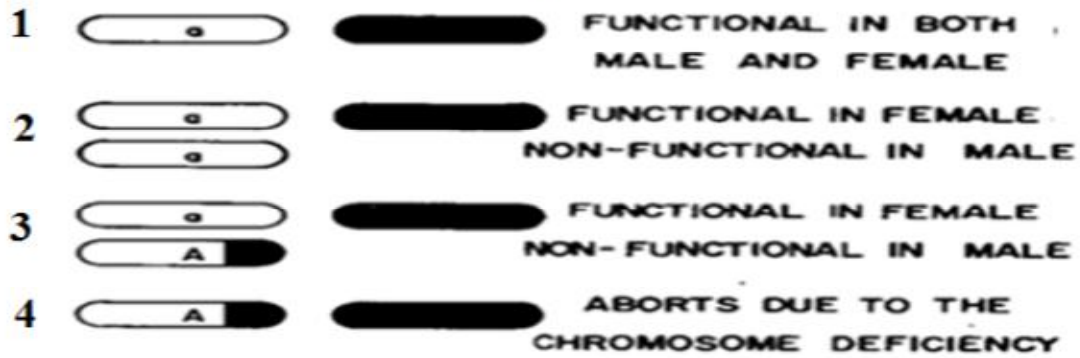


Figure 1. Breeding behavior of selfed balanced tertiary trisomics. Left, marker gene on centromere segment

Trisomy

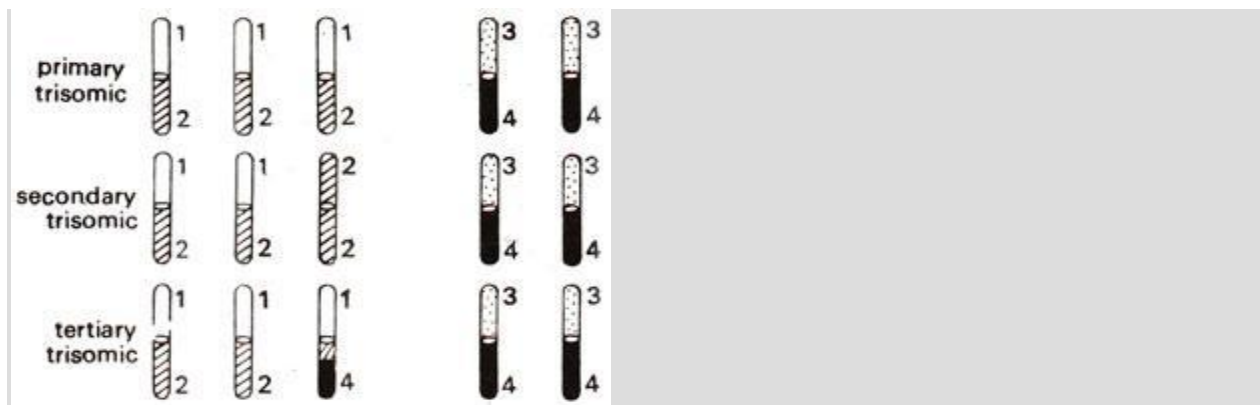


Fig. 20.2. Three kinds of trisomics.

One of the most extensively studied trisomic series is that produced and studied by **T. Tsuchiya** (who died in May, 1992) in barley. Trisomics are also known in *Homo sapiens* (human beings). Trisomy for certain chromosomes causes definite morphological abnormalities in human beings.

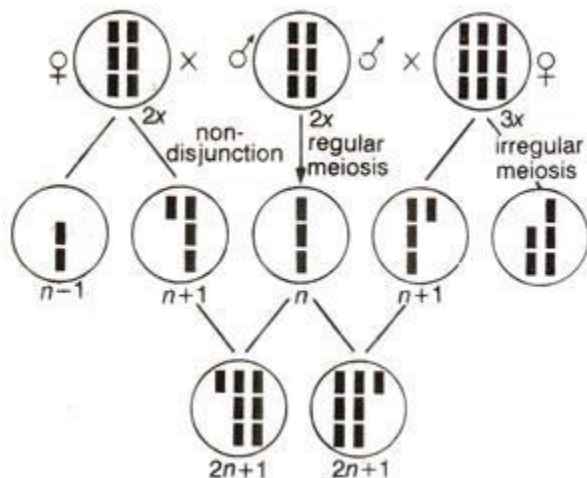


Fig. 20.4. Trisomic conditions associated with changes in fruit morphology in *Datura stramonium*.

Mongolism (Down's syndrome) is one such **feature, which** is common in children and is characterized by mental retardation, a short body, swollen tongue and eyelid folds resembling those of Mongolian races (for details, see Human Genetics). Other cases of trisomy are also known in a number of different plant and animal species.

Production of trisomics. Trisomics may originate spontaneously due to production of $n + 1$ type of gametes (Fig. 20.4) due to rare non-disjunction of a bivalent. However more often trisomics are produced artificially either by selfing triploids (triploids are produced by crossing diploids

and autotetraploids) or by crossing these Triploids as females with diploids as male ($3x \times 2x$). In either case trisomics are obtained in large number and can be identified through phenotypic effects of individual chromosomes.

Cytology of trisomics.

A trisomic has an extra chromosome which is homologous to one of the chromosomes of the complement. Therefore, it forms a trivalent. This trivalent may take a variety of shapes in primary and secondary trisomics as shown in Figure 20.5. In a tertiary trisomic a characteristic pentavalent is observed.

Trisomic analysis.

Trisomics are also used for locating genes on specific chromosomes. If a particular gene is located on the chromosome involved in trisomy, segregation in the progeny of this trisomic will not follow a Mendelian pattern, but the ratio will deviate from normal $3 : 1$ F_2 and $1 : 1$ test cross ratios. The expected ratios in trisomics can be worked out, and are given in Table 20.1. In Table 20.1, ratios based on chromosome segregation as well as those based on chromatid segregation are given. **Chromosome** segregation will hold good, when the gene is located very close to centromere permitting no crossing over between the gene and the centromere, so that both sister chromatids will be similar. In **chromatid segregation**, gene is located away from centromere permitting crossing over between gene and centromere.

Table 20.1. Phenotypic ratios obtained from different genotypes of trisomic ($2x + 1$) or triploid ($3x$) individuals (assuming 50% ♀ transmission and no ♂ transmission of $n + 1$ type of gametes).

Genotype	Phenotypic ratios			
	Chromosome segregation		Chromatid segregation	
	selfing	testcross	selfing	testcross
(1) AAA (triplex)	all : 0	all : 0	all : 0	all : 0
(2) AAa (duplex)	17 : 1	5 : 1	14 : 1	4 : 1
(3) Aaa (simplex)	2 : 1	1 : 1	29 : 16	14 : 16
(4) aaa (nulliplex)	0 : all	0 : all	0 : all	0 : all

More in this section

Trisomics are those organisms, which have an extra chromosome ($2n + 1$). Since the extra chromosome may belong to any one of the different chromosomes of a haploid complement, the number of possible trisomics in an organism will be equal to its haploid chromosome number. For instance, we know that haploid chromosome number in barley is $n = 7$, consequently, seven trisomics are possible. Trisomics, where extra chromosome is identical to two homologues, are called **primary trisomics**. Besides these, there are secondary and tertiary trisomics. While a **secondary trisomic** means that extra chromosome should be an isochromosome (both arms genetically similar), a **tertiary trisomic** would mean that extra chromosome should be a

translocated one (Fig. 20.2). Trisomics were obtained for the first time in *Datura stramonium* (jimson weed) by A.F. Blakeslee and his co-workers. Since haploid chromosome number in this species is $n = 12$, 12 primary trisomics, 24 secondary trisomics and a large number of tertiary trisomics are possible. Most of the trisomics were identified by size, shape and other morphological features of the fruit (Fig. 20.3).

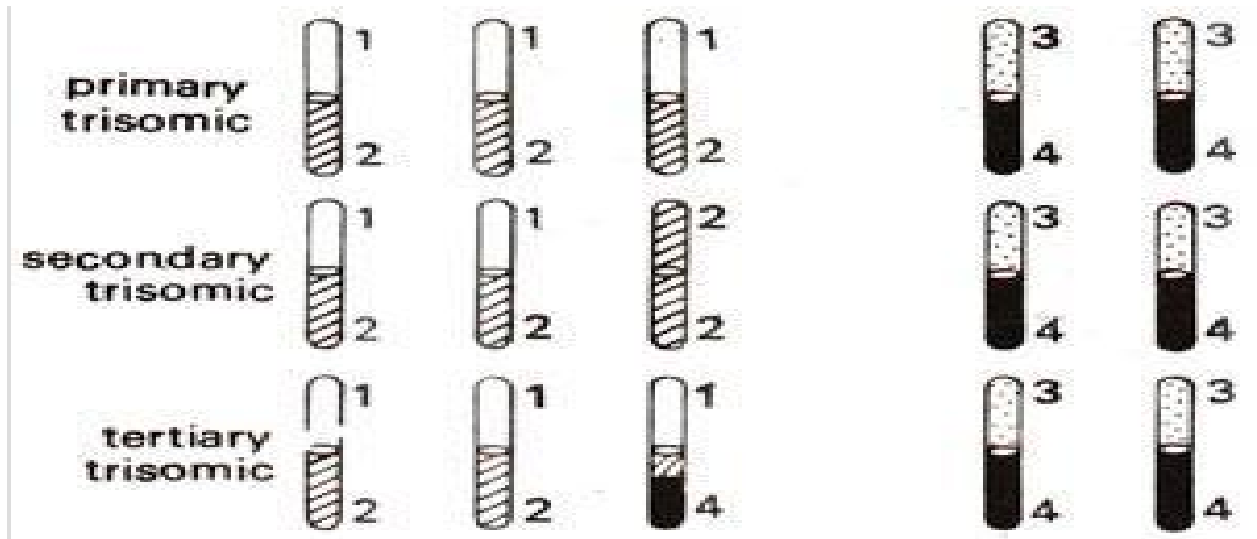


Fig. 20.2. Three kinds of trisomics.

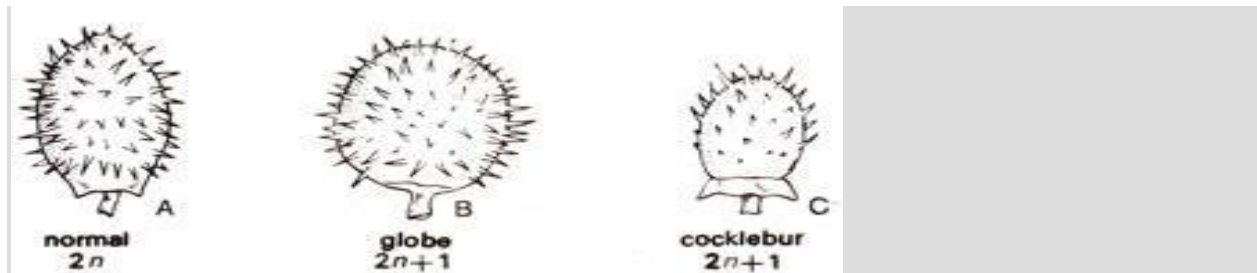


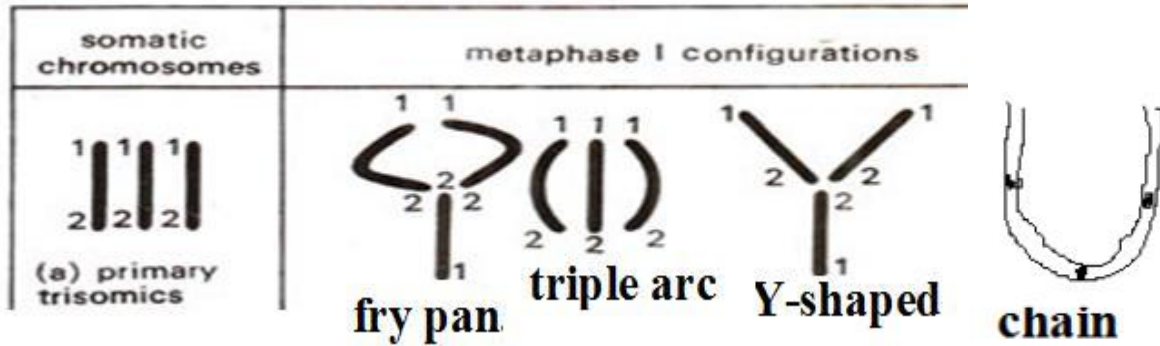
Fig. 20.3. Trisomic conditions associated with changes in fruit morphology in *Datura stramonium*.

Meiotic behavior of trisomics

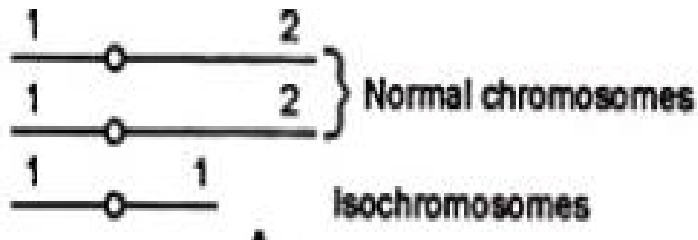
The most characteristic feature of primary trisomics is the formation of a trivalent at metaphase-I of meiosis. Secondary trisomics at metaphase-I forms mostly ring trivalents while tertiary trisomics form dumbbell shaped pentavalents.

Primary Trisomics: the trivalents in primary trisomics assumes different configurations such as chain, fry pan, triple arc and Y-shaped. The most common configuration in primary trisomic is

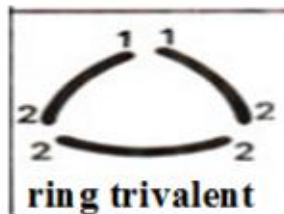
chain trivalent. Based on the extent of pairing of chromosome and number of chiasmata formed at pachytene stage. For longer chromosomes that can form two chiasmata, frypan trivalents are frequently formed and for smaller chromosomes which cannot form two chiasmata, the most frequent trivalent configuration is chain type.



Secondary Trisomics: When the extra chromosome is an iso-chromosome, the aneuploid is called secondary trisomic; its formula is “2n + iso”. One chromosome arm is represented four times in the secondary trisomic. In secondary trisomics, chromosome configuration based on chromosome pairing may be a trivalent (1^{III}) or “bivalent + univalent” ($1^{II} + 1^I$) and 3^I



The most frequent trivalent configuration in secondary trisomics is **ring trivalent** (1^{III})



Sometimes both the normal homologues pair to form a bivalent, while both arms of the iso-chromosome pair together to form a U-shaped univalent ($1^{II} + 1^I$).



Tertiary Trisomic: A dumbbell shaped pentavalent configuration is formed at metaphase-I



Uses of Trisomics

1. Secondary trisomics can be used to locate genes on specific chromosomes and finding out distance of these genes from centromere, a technique called **chromosome mapping**.
2. They may be used to know the method of action of certain genes.
3. Primary trisomics are used in assigning linkage groups to specific chromosomes.
3. Tertiary Trisomics are used to produce female parent for hybrid seed production.

Trisomics in polyploids: trisomics have been produced in crops such as wheat (*Triticum aestivum* $2n = 6x = 42$), oats (*Avena sativa* $2n = 6x = 42$), tobacco (*Nicotiana tabacum* $2n = 4x = 48$) and cotton (*Gossypium hirsutum* $2n = 4x = 52$). These trisomics in polyploids were resulted due to non-disjunction of homologous chromosomes. Sears produced complete set of trisomics only in 'chinese spring' variety of wheat by pollinating a haploid plant with the pollen from a normal wheat. These trisomics resembled in morphology to hexaploid wheat. However more often trisomics are produced artificially either by selfing triploids or by crossing these Triploids as females with diploids as male ($3x \times 2x$). In either case trisomics are obtained in large number. In wheat trisomics are used to obtain tetrasomics and the 'compensating nullisomic-tetrasomic' lines which proved useful for establishing homeologous relationships. In cotton trisomics, amphidiploids are first produced using diploid parents. The amphidiploids are then backcrossed to tetraploid cotton to isolate trisomics with $2n = 4x + 1 = 53$ chromosomes. These trisomics are better called as alien addition lines or interspecific trisomics and resembled the characteristics of diploid species.

Tetrasomics in plants: Tetrasomics are individuals having an extra pair of homologous chromosomes. In diploids these will be represented as $2x + 2$ and in tetraploid as $4x + 2$ and in hexaploid as $6x + 2$. Tetrasomics have a particular chromosome represented in four doses ($2n + 2$). The types of possible tetrasomics are equal to the haploid chromosome number of an organism.

All 21 possible tetrasomics are available in wheat. Tetrasomics have been originated in polyploid plants such as wheat, oat and cotton by **selfing of trisomics**. In wheat tetrasomics are used to obtain 'compensating nullisomic-tetrasomic' lines which proved useful for establishing homeologous relationships. However tetrasomics are not useful for chromosome mapping. **Meiotic Behaviour of tetrasomics:** During meiosis, the four homologues of the tetrasomic set tend to form a quadrivalent.

Wheat is unique in that it can tolerate a high degree of aneuploidy (abnormal chromosome number) because of its polyploid buffering capacity. Some of the most important and useful genetics stocks ever developed in wheat are the nullisomic-tetrasomic (NT) lines and the ditelosomic (dt) lines. NT lines are lacking one pair of chromosomes and the absence of the pair is compensated for by an extra pair of homoeologous chromosomes. Ditelosomic lines are lacking one pair of chromosome arms. For example, the line dt1AS has the complete normal chromosome constitution except that the long arms of the pair of 1A chromosomes are missing. *Homeologous chromosomes* (Partially homologous *chromosomes*) include chromosomes of different 'genomes' which pair occasionally in many interspecific hybrids or allopolyploids. R.L. Knight defines homoeologous chromosomes as 'chromosomes that are homologous in parts of their length but are not homologous along their entire length as seen in homologous chromosomes.