NUMERICAL MUTATIONS (POLYPLOIDY)

Mutations which alter the chromosome structure, size or gene arrangement are chromosomal mutations. Chromosomal mutations are widely called as chromosomal aberrations. These are grouped into two broad classes based open whether they alter the structure or number of chromosomes.

Chromosome Mutations - gross changes in chromosomes.

Changes in the number of chromosomes.

1. Euploidy - variation in the number of sets of chromosomes.

a. Haploidy (Monoploidy) - one set of chromosomes (n) : ABC

b. Polyploidy-three or more sets of chromosomes.

c. Triploidy-3 sets of chromosomes (3n) : ABC, ABC, ABC.

d. Tetraploidy-4 sets of chromosomes (4n): ABC, ABC, ABC, ABC.

e. Pentaploidy-5 sets of chromosomes (5n) : ABC, ABC, ABC, ABC, ABC.

f. Hexaploidy (6n), **Septaploidy** (7n), **Octoploidy** (8n), etc

2. Aneuploidy - variation in the number of chromosomes of a set. (Reduction in the normal number of chromosomes.)

a. Monosomics - Loss of one chromosome (2n-1) : ABC, AB.

b. Double monosomics - Loss of 2 different chromosomes (2n-1-1): ABC, A.

b - loss of a pair of homologous chromosomes (2n-2) : AB, AB:

b. Increase in the number of chromosomes (polysomies).

Trisomies - presence of 1 extra chromosome (2n+ 1) : ABC, ABC, A.

Double trisomics - 2 different extra chromosomes (2n + 1 + 1) : ABC, ABC, AB.

Tetrasomics - an extra pair of homologous chromosomes (2n+2): ABC, ABC, AA.

pentasomics (2n+3), **Hexasomics** (2n+4), **Sepiasomics** (2n+5), etc. **Euploidy**

The term euploidy (Gr., eu-true or even; ploid-unit) designates genomes containing whole sets of chromosomes. The euploids are those organisms which contain balanced set or sets of chremosomes or genomes in any number, in their body cells. The euploidy is of following types: The number of chromosomes in a basic set is called the **monoploid number** (*x*). Organisms with multiples of the monoploid number of chromosomes are called euploid. Eukaryotes normally carry either one chromosome set (haploids) or two sets (diploids). Haploids and diploids, then, are both cases of normal euploidy. Euploid types that have more than two sets of chromosomes are called polyploid.

Polyploidy

Humans are diploid creatures, meaning for every chromosome in our body, there is another one to match it.

- Haploid creatures have one of each chromosome
- Diploid creatures have two of each chromosome
- Triploid creatures have three of each chromosome
- Polyploid creatures have three or more of each chromosome

They can be represented by n where n equals haploid, 2n equals diploid and so on. It is possible for a species, particularly plant species, to produce offspring that contains more chromosomes than its parent. This can be a result of non-disjunction, where normally a diploid parent would produce diploid offspring, but in the case of non-disjunction in one of the parents, produces a polyploid.

In the case of triploids, although the creation of particular triploids in species is possible, they cannot reproduce themselves because of the inability to pair homologous chromosomes at meiosis, therefore preventing the formation of gametes. Polyploidy is responsible for the creation of thousands of species in today's planet, and will continue to do so. It is also responsible for increasing genetic diversity and producing species showing an increase in size, vigour and an increased resistance to disease.

The polyploid types are named triploid ($3x$), tetraploid ($4x$), pentaploid ($5x$), hexaploid ($6x$), and so forth. Polyploids arise naturally as spontaneous chromosomal mutations. However, many species of plants and animals have clearly arisen through polyploidy, so evidently evolution can take advantage of polyploidy when it arises. It is worth noting that organisms with one chromosome set sometimes arise as variants of diploids; such variants are called monoploid ($1x$). In some species, monoploid stages are part of the regular life cycle, but other monoploids are spontaneous aberrations.

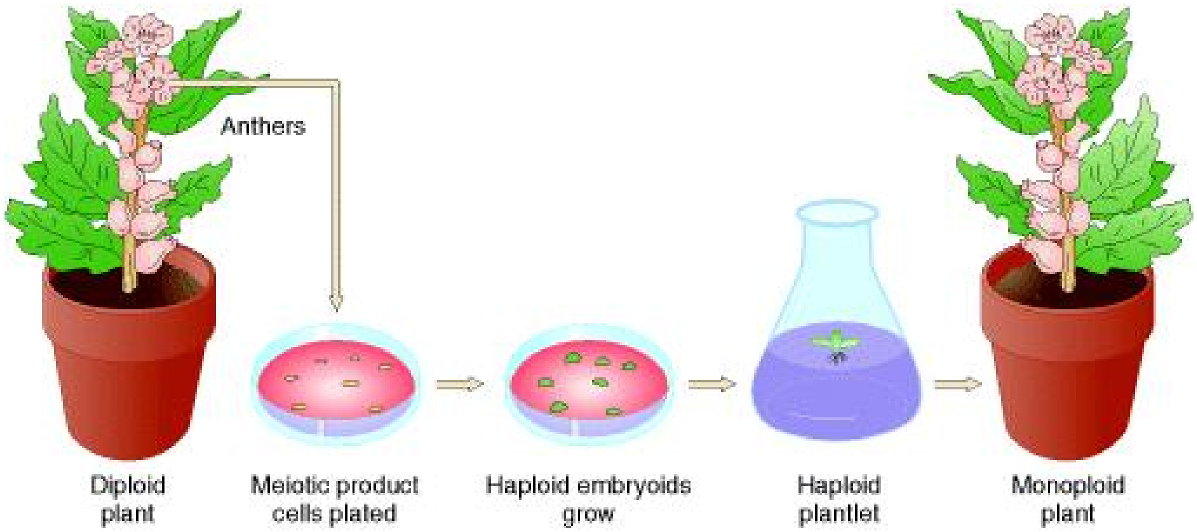
The haploid number (n), which we have already used extensively, refers strictly to the number of chromosomes in gametes. In most animals and many plants with which we are familiar, the haploid number and monoploid number are the same. Hence, n or x (or $2n$ or $2x$) can be used interchangeably. However, in certain plants, such as modern wheat, n and x are different. Wheat has 42 chromosomes, but careful study reveals that it is hexaploid, with six rather similar but not identical sets of seven chromosomes. Hence, $6x=42$ and $x=7$. However, the gametes of wheat contain 21 chromosomes, so $n=21$ and $2n=42$.

Monoploids

In monoploidy, the monoploid organisms have one genome (n) in their body cells. When monoploidy occurs in gametes (sperms and eggs) it is termed as haploidy. Most micro-organisms (e.g., bacteria. fungi and algae); gametophytic generation of plants (e.g., bryophytes and other plants); sporophytic generation of some higher angiospermic plants (e.g., Sorghum, Triticum, Hordeum, Datura, etc.) and certain hymenopteran male insects (e.g., wasps, bees, etc.) have one genome in their body cells, hence are monoploids. Monoploids are usually smaller and less vigorous than their diploid prototypes. Characteristically, monoploid plants are sterile. The reason of sterility is that the chromosomes have no regular pairing partners (homologous chromosomes) during meiosis, and meiotic products are deficient in one or more chromosomes. For instance, a haploid in maize (2n=20) will have 10 chromosomes and the number of chromosomes in a gamete can range from 0-10. Consequently, considerable sterility will be found in a monoploid maize.

Male bees, wasps, and ants are monoploid. In the normal life cycles of these insects, males develop parthenogenetically—that is, they develop from unfertilized eggs. However, in most species, monoploid individuals are abnormal, arising in natural populations as rare aberrations. The germ cells of a monoploid cannot proceed through meiosis normally, because the chromosomes have no pairing partners. Thus, monoploids are characteristically sterile.

Monoploids play an important role in modern approaches to plant breeding. Diploidy is an inherent nuisance when breeders want to induce and select new gene mutations that are favorable and to find new combinations of favorable alleles at different loci. New recessive mutations must be made homozygous before they can be expressed, and favorable allelic combinations in heterozygotes are broken up by meiosis. Monoploids provide a way around some of these problems. In some plant species, monoploids can be artificially derived from the products of meiosis in a plant's anthers. A cell destined to become a pollen grain can instead be induced by cold treatment to grow into an embryoid, a small dividing mass of cells. The embryoid can be grown on agar to form a monoploid plantlet, which can then be



potted in soil and allowed to mature.

Figure a. Generating monoploid plants by tissue culture. Pollen grains (haploid) are treated so that they will grow and are placed on agar plates containing certain plant hormones. Under these conditions, haploid embryoids will grow into monoploid plantlets. After having been moved to a medium containing different plant hormones, these plantlets will grow into mature monoploid plants with roots, stems, leaves, and flowers.

Plant monoploids can be exploited in several ways. In one, they are first examined for favorable traits or allelic combinations, which may arise from heterozygosity already present in the parent or induced in the parent by mutagens. The monoploid can then be subjected to chromosome doubling to achieve a completely homozygous diploid with a normal meiosis, capable of providing seed. It is achieved by the application of a compound called colchicine to meristematic tissue. **Colchicine**—an alkaloid drug extracted from the autumn crocus - inhibits the formation of the mitotic spindle, so cells with two chromosome sets are produced. These cells may proliferate to form a sector of diploid tissue that can be identified cytologically.

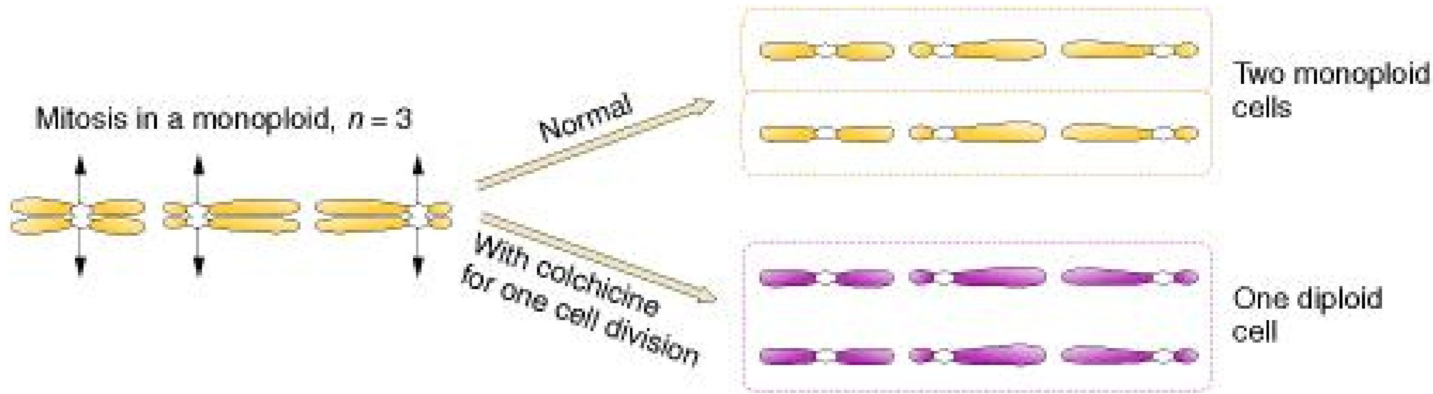


Figure b. The use of colchicine to generate a diploid from a monoploid. Colchicine added to mitotic cells during metaphase and anaphase disrupts spindle-fiber formation, preventing the migration of chromatids after the centromere is split. A single cell is created that contains pairs of identical chromosomes that are homozygous at all loci.

Another way in which the monoploid may be used is to treat its cells basically like a population of haploid organisms in a mutagenesis-and-selection procedure. A population of cells is isolated, their walls are removed by enzymatic treatment, and they are treated with mutagen. They are then plated on a medium that selects for some desirable phenotype. This approach has been used to select for resistance to toxic compounds produced by one of the plant's parasites and to select for resistance to herbicides being used by farmers to kill weeds. Resistant plantlets eventually grow into haploid plants, which can then be doubled (with the use of colchicine) into a pure-breeding, diploid, resistant type.

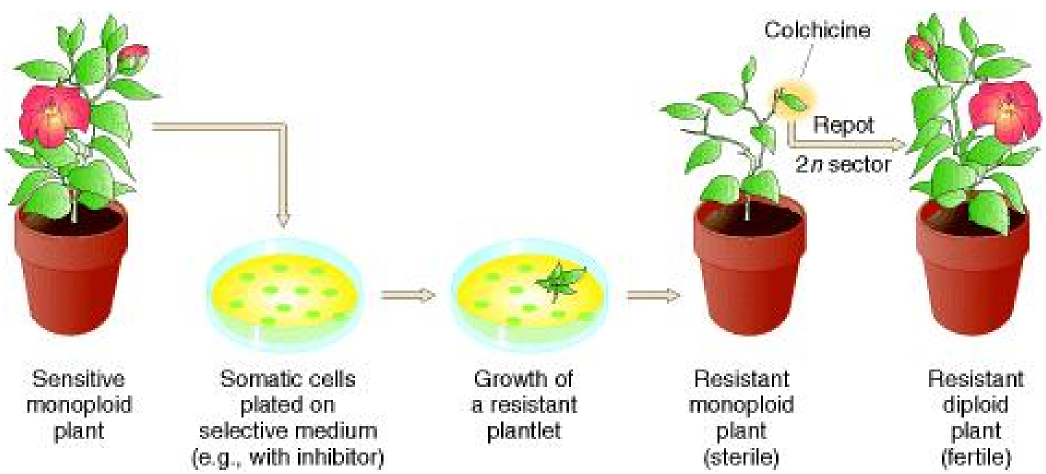


Figure c. Using microbial techniques in plant engineering. The cell walls of haploid cells are removed enzymatically. The cells are then exposed to a mutagen and plated on an agar medium containing a selective agent, such as a toxic compound produced by a plant parasite. Only those cells containing a resistance mutation that allows them to live within the presence of this toxin will grow. After treatment with the appropriate plant hormones, these cells will grow into mature monoploid plants and, with proper colchicine treatment, can be converted into homozygous diploid plants.

These powerful techniques can circumvent the normally slow process of meiosis-based plant breeding. The techniques have been successfully applied to several important crop plants, such as soybeans and tobacco.

The anther technique for producing monoploids does not work in all organisms or in all genotypes of an organism. Another useful technique has been developed in barley, an important crop plant. Diploid barley, *Hordeum vulgare,* can be fertilized by pollen from a diploid wild relative called *Hordeum bulbosum.* This fertilization results in zygotes with one chromosome set from each parental species. In the ensuing somatic cell divisions, however, the chromosomes of *H. bulbosum* are eliminated from the zygote, whereas all the chromosomes of *H. vulgare* are retained, resulting in a haploid embryo. (The haploidization appears to be caused by a genetic incompatibility between the chromosomes of the different species.) The chromosomes of the resulting haploids can be doubled with colchicine. This approach has led to the rapid production and widespread planting of several new barley varieties, and it is being used successfully in other species too.

Diploidy

The diploidy is characterized by two genomes (2n) in each somatic cell of the diploid organisms. Most animals and plants are diploids. The diploidy is related with fertility, balanced growth, great vigorosity, adapatability and survivality of the diploid organisms

Polyploids

The organisms with more than two genomes are called polyploids. Among plants and animals, the polyploidy occurs in a multiple series of 3, 4, 5, 6, 7, 8, etc., of the basic chromosome or genome number and thus is causing triploidy, tetraploidy, pentaploidy, hexaploidy, heptaploidy, octaploidy, respectively. Ploidy levels higher than tetraploid are not commonly encountered in natural populations, but our most important crops and ornamental flowers are polyploid, e.g., wheat (hexaploid, 6n), strawberries (octaploid, 8n), many commercial fruit and ornamental plants, liver cells of man, etc. Other examples of polyploidy among plants and animals are following:

A: Examples of polyploidy in plants

The polyploidy is most common among angiosperms and some of economically important polyploid angiospermic plants are peanuts (Arachis),oats (Avena), coffee (Coffea), strawberry (Fragaria), cotton (Gossypium), barely (Hordeum), sweet potato (Ipomoea), apple (Malus), alfa-alfa (Medicago), banana (Musa), tobacco (Nicolina), plum (Prunus), sugar cane (Saccharum), potato (Solanum), sorghum (Sorghum), clover (Trifolium), and wheat (Triticum).

A continuous polyploid series has been reported in rose plant. Aeuploid series of basic number of 7 (monoploid) including diploids (2n= 14), triploids (21), tetraploids (28), pentaploids (35), hexapolid (42), and octaploid (56) has been reported in different species of Rosa. Likewise, the genus Chrysanthemum has basic chromosome number 9 and has a euploidic series of diploid (2n = 18), tetraploids (4n=36), hexaploids (6n=54), octaploids (8n=72) and decaploids (10n=90) in its different species.

The genus Solanum has basic chromosome number 12 and has a euploidic series of diploids (2n=24), triploids (3n=36), tetraploids (4n=48), pentaploids (5n=60), hexaploids (6n=72), octaploids (8n=96), and decaploids (10n = 120) in its different species.

Origin of Polyploidy

Different degrees of ploidy are originated by different means. However, two basic irregular processes have been discovered by which polyploids may evolve from diploid plants and become established in nature:

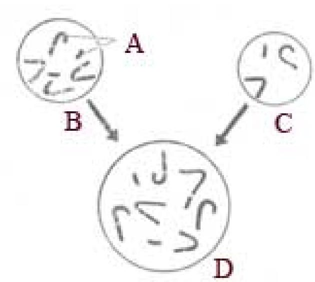
(1) Somatic doubling-cells sometimes undergo irregularities at mitosis and give rise to meristematic cells that perpetuate these irregularities in new generations of plants.

(2) Reproductive cells may have an irregular reduction or equation division in which the sets of chromosomes fail to separate completely to the poles at anaphase. Both sets thus become incorporated in the same nucleus resulting in the doubling of chromosome number in the gamete (see Gardner, 1912). Thus, a triploid originates

by the fusion of a haploid gamete (n) with a diploid gamete (2n), the later of which may be originated by irregularities during meiosis.

Likewise, a tetraploid may be originated by the somatic doubling of the chromosome number or by union of unreduced diploid gametes.

The somatic doubling of genome is accomplished either spontaneously or it can be induced in high frequency by exposure to chemicals such as colchicine, etc., or heat or cold. Other levels of polyploidy are also originated by same methods.



A. Chromosomes B. Diploid gamete C. Haploid gamete D. Triploid zygote

Induction of Polyploidy

The polyploidy can be induced in common diploid organisms by following methods:

1. Cell generation - In certain bryophytes such as mosses, the polyploidy has been induced by cutting their diploid sporophytes and keeping the sporophytes in moist conditions. The cells of the cut ends regenerated threads which were true protonema and produced diploid gametophytic generation instead of monoploidic generation.

2. Physical agents - Following kinds of physical conditions induce polyploidy in plants:

(i) Temperature shocks - Extreme temperature changes some. times result in a higher frequency of polyploid cells.

(ii) Centrifugation - The centrifugation of seedlings of plants causes polyploidy in their cells. In Nicotiana, polyploidy has been induced by this method.

(iii) X-rays - The radioactive substances such as radium and X-rays have been found to induce polyploidy in normal diploid plant cells.

3. Chemical agents - Some chemicals such as colchicine, chloral hydrate, acenaphthene, veratrine, sulfanil amide, ethyl, mercury chloride. hexachlorocyclohexane have been reported to induce polyploidy in plants. These chemical substances when used to dividing diploid cells, they disturb the mitotic spindle and cause non-segregation of already duplicated chromosomes and thus, convert the diploid cells into tetraploid cells. The tetraploid cells, likewise, are converted into different levels of polyploidy

Kinds of Polyploidy

In the realm of polyploids, we must distinguish between **autopolyploids,** which are composed of multiple sets from within one species, and **allopolyploids,** which are composed of sets from different species. Allopolyploids form only between closely related species; however, the different chromosome sets are **homeologous** (only partly homologous)—not fully homologous, as they are in autopolyploids.

Allopolyploidy- The prefix "allo" indicates that nonnhomologous sets of chromosomes are involved.

P_1: **Species X** **X** **Species Y**

(AA) ↓ **(BB)**

(Diploid) **(Diploid)**

F_1: **AB**

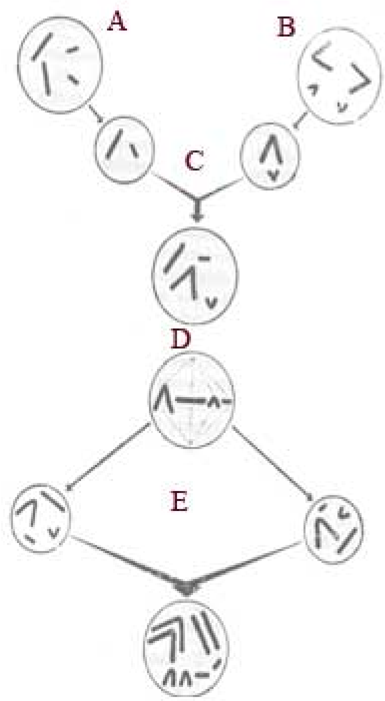
Diploid sterile hybrid

↓ **Colchicine**

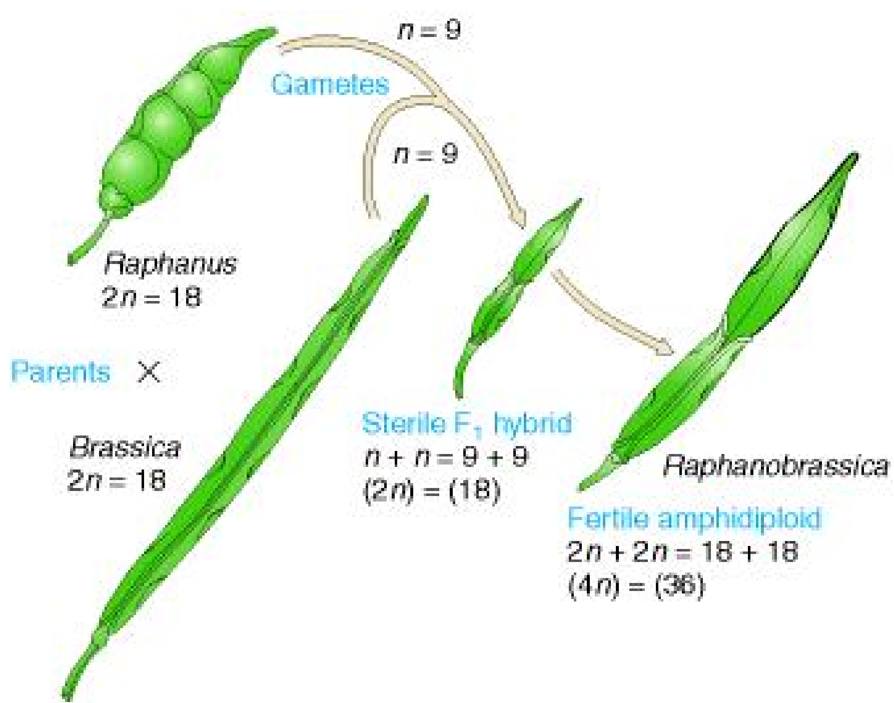
AABB

Fertile amphidiploid tetraploid

The union of unreduced or diploid or polyploid gametes from different diploid or polyploid species could produce in one step, an amphipolyploid or allopolyploid; which appears and behaves like a new species. Let A represent a set of genome in species X, and let B represent another genome in a species Y. The F 1 hybrids of these species than would have one A genome and another B genome. The F1 diploid but sterile hybrids can be converted into fertile allotetraploids by treating them by colchicine.



The Russian cytologist, G. D. Karpechenko (1928) first ore. , synthesized a allotetraploid genus called Rhaphanobrassica from the artificial crosses between vegetables belonging to different genera, the radish (Raphanus satirum, 20 = 18) and the cabbage (Brassica oleracea, 2n= 18). The F1 hybrids were diploid and having the root of cabbage and leaves of radish. They were highly sterile because of failure of each set of chromosomes to provide sufficient genetic homology to affect pairing. Among these sterile F1 hybrids, however, he found certain fertile allotetraploids which contained 36 chromosomes and were named as Rhaphanobrassica.



Triticum spelta, is a hexaploid wheat which was artificially synthesized in 1946 by E. S. McFadden and E. R. Sears and also by H. Kihara. They crossed an emmer wheat, Triticum dicoccoides (tetraploid: 2n= 28) with goat grass, Aegilops squarrosa (diploid; 2n= 14) and doubled the chromosome number in the F1 hybrids This artificially synthesized hexaploid wheat was found to be similar to the primitive wheat T. spelta. When the synthesized hexaploid wheat was crossed with naturally occurring T. spelta, the F1 hybrid was completely fertile, this suggested that hexaploid wheat must have originated in the past due to natural hybridization between tetraploid wheal and goat grass followed by subsequent chromosome doubling.

P_1	**Trilicum dicoccoides**	**X**	**Aegilops squarrosa**
	(Tetraploid emmer wheat)	↓	**(Diploid goat grass)**
	AABB		**DD**
	(2n=28; 14 bivalents)		**(2n=14; 7 bivalents)**
F_1:		**ABD**	

(Triploid hybrid)

(2n=21; 21 univalents)

↓

AA BB DD

Synthesized hexaploid wheat

(Triticum spelta)

(2n=42; 21 bivalents)

Another interesting case of allotetraploidy has been observed·--tn the production of a rust resistant allotetraploid wheat plant. Common wheat plant, Triticum vulgare is susceptible to leaf rust, a seriouse disease caused by the fungus

Puccinia triticina. A wild grass of the Mediterranean region, Aegilops umbellulata is completely resistant to this disease. Sears (1956) have transferred the genes of rust resistance of A. umbellulata into T. vulgare genome by following method:

He crossed the plants of A. umbellulata with T. dicoccoides and got sterile hybrid which by treatment with colchicine was transformed into a rust-resistant, fertile allotetraploid having 21 pairs of chromosomes. The allotetraploid was crossed to T. vulgare and a fertile, rust-resistant hybrid was produced.

Gossypium hirsutum, the New world cotton plant, is another interesting example of allopoly ploidy. Old world cotton, Gossypium herbaceum, has 13 pairs of chromosomes, while American or "upland cotton" also contains 13 pairs of chromosomes. J. O. Beasley crossed the old world and American cottons and doubled the chromosome number in the F) hybrids. The allopolyploids thus produced resembled the cultivated New world cotton and when crossed with it gave fertile F1 hybrids These results, thus, suggested that tetraploid Gossypium hirsutum originated from two diploid species, namely G. herbaceum (20=26) and G. raimondii (2n=26).

Gossypium herbaceum	**X**	**Gossypium raimondii**
(Old world cotton; 2n=26; 13 bivalents	↓	**(American or upland cotton, 2n=26; 13 bivalents**
	F1 hybrid	
	(2n=26, 26 univalents)	
	↓	**Colchicine**
	New wold cotton	
	(Gossypium hirsurum)	
	(2n=52; 26 bivalents)	

Today, allopolyploids are routinely synthesized in plant breeding. Instead of waiting for spontaneous doubling to occur in the sterile hybrid, the plant breeder adds colchicine 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 amphidiploid has ever been widely used. This amphidiploid is *Triticale,* an amphidiploid between wheat (*Triticum,* $2n=6x=42$ and rye (*Secale,* $2n=2x=14$ *Triticale* combines the high yields of wheat with the ruggedness of rye. The below figure shows the procedure for synthesizing *Triticale.*

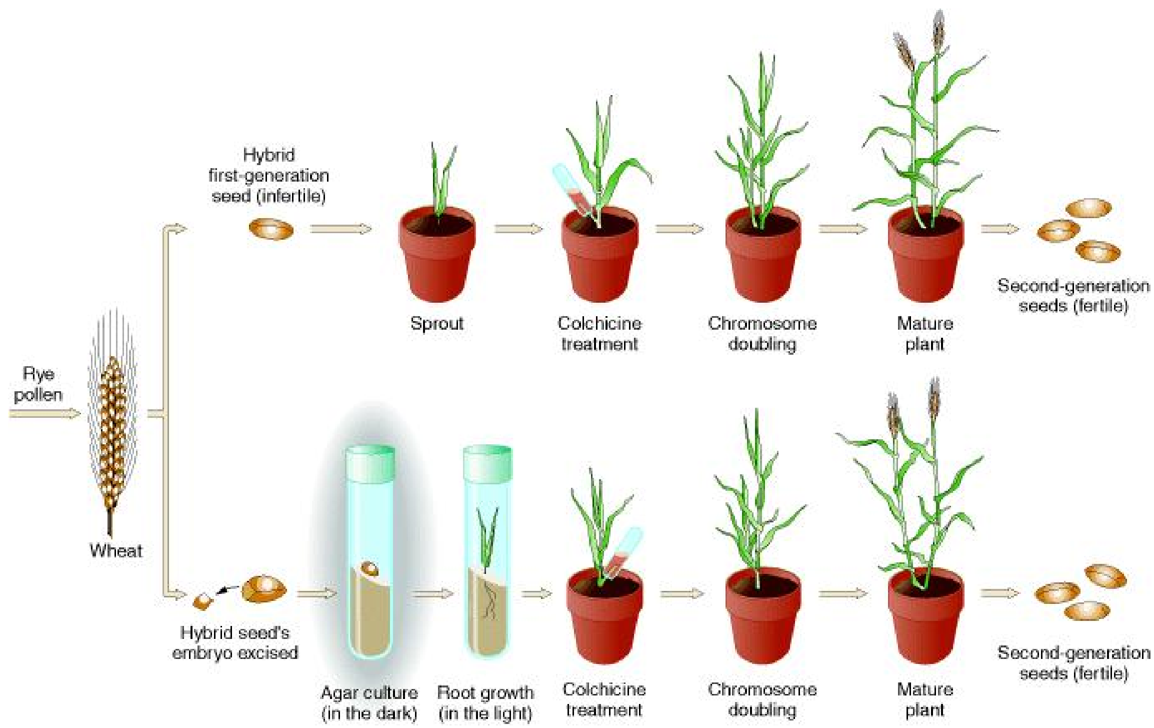


Figure 18-10. Techniques for the production of the amphidiploid *Triticale.* If the hybrid seed does not germinate, then tissue culture *(lower path)* may be used to obtain a hybrid plant.

In nature, allopolyploidy seems to have been a major force in speciation of plants. There are many different examples. One particularly satisfying one is shown by the genus *Brassica,* as illustrated in Figure 18-11. Here three different parent species have hybridized in all possible pair combinations to form new amphidiploid species.

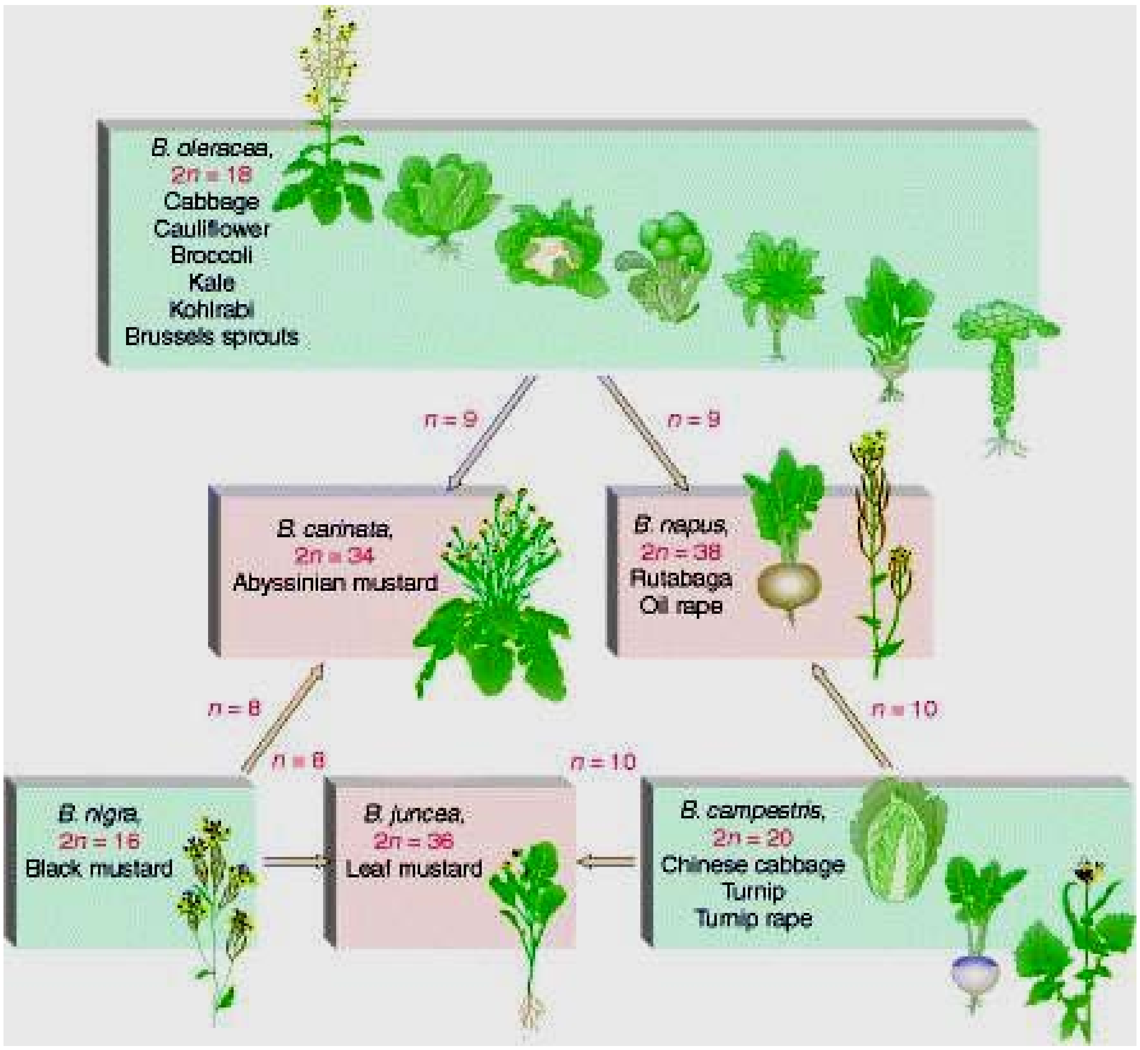


Figure 18-11. A species triangle, showing how amphidiploidy has been important in the production of new species of *Brassica.*

A particularly interesting natural allopolyploid is bread wheat, *Triticum aestivum* ($2n=6x=42$). By studying various wild relatives, geneticists have reconstructed a probable evolutionary history of bread wheat (Figure 18-12).

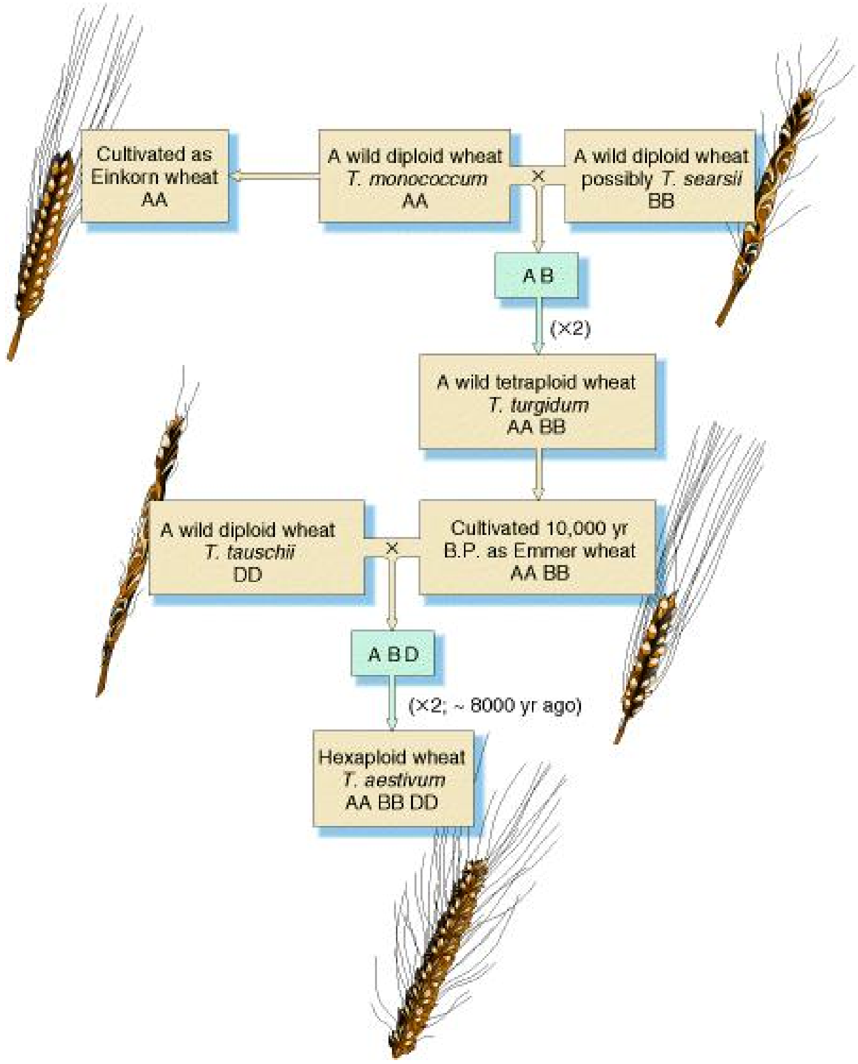


Figure 18-12. Diagram of the proposed evolution of modern hexaploid wheat, in which amphidiploids are produced at two points. A, B, and D are different chromosome sets

Autopolyploids

The prefix "auto" indicates that the ploidy involves only homologous chromosome sets. Somatic doubling of a diploid produces four sets of genomes of a tetraploid and likewise, somatic doubling of a tetraploid produces eight sets of genomes of a octaploid. Union of unreduced diploid or tetraploid gametes from the same species would accomplish the same result.For example, if a diploid species has two similar sets of chromosomes or genomes (AA), an autotriploid will have three similar genomes (AAA), and an autotetriploid, will have four such genomes

(AAAA). Since an autotriploid remains sterile and cannot produce seeds, therefore, it has great commercial value in producing seedless varities of economical plants.

For example, in Japan, H. Kihara produced seedless watermelons, which were autotriploids. Common "doob' grass of U. P. and Bihar is an autotriploid. Other common seedless autopolyploids are grapes, sugarbeet, Banana, etc. In O. lamarckiana, the giant mutant described by de Vries was later on discovered to be an autotetraploid. Further, whenever autopolyploids, originate in nature, these would be eliminated due to natural selection.

The chromosome sets or genomes are identical. The genome formula (capital letters represent a group of chromosomes that is generally referred to as the basic genome or chromosome set) is AAA (autotriploidy), or BBBB (autotetraploidy),etc. Autopolyploids are also called polysomicpolyploids.

Origin of Autopolyploids

Autopolyploids spontaneously occur in the nature in a low frequency and can be induced artificially using various ways, such as heat and chemical treatments, decapitation, and selection from twin seedlings. The effective method to obtain autopolyploids is using colchicine.
Colchicine is a spindle fiber poison or suppressant. It inhibits the spindle mechanism at mitosis, resulting in multiples of normal chromosome number.

Triploids

Triploids are usually autopolyploids. They arise spontaneously in nature or are constructed by geneticists from the cross of a $4x$ (tetraploid) and a $2x$ (diploid). The $2x$ and the x gametes unite to form a $3x$ triploid. Autotriploids are genetically equal to trisomics for each chromosome. The three chromosomes will pair as a trivalent or a bivalent plus a univalent. Chromosome separation from such pairing is irregular. Daughter nuclei will receive either one or two copies from each chromosome. Consequently, most of the gametes resulting from autotriploid individuals do not have balanced chromosome complements and are not viable. If progeny survives from autotriploids it is mostly an aneuploid. Autotriploids can be produced by crossing diploids with their corresponding autotetraploids. The high sterility of autotriploids has been explored in plant breeding. Triploid bananas (2n = 33) are vigorous but seedless and therefore preferred for food consumption.

Triploid watermelons have only undeveloped seeds. Triploid is also applied in seedless Citrus cultivars.

Triploids are characteristically sterile. The problem, like that of monoploids, lies in pairing at meiosis. Synapsis, or true pairing, can take place only between two chromosomes, but one chromosome can pair with one partner along part of its length and with another along the remainder, which gives rise to an association of three chromosomes. Paired chromosomes of the type found in diploids are called **bivalents.** Associations of three chromosomes are called **trivalents,** and unpaired chromosomes are called **univalents.** Hence in triploids there are two pairing possibilities, resulting either in a trivalent or in a bivalent plus a univalent. Paired centromeres segregate to opposite poles, but unpaired centromeres pass to either pole randomly as below:

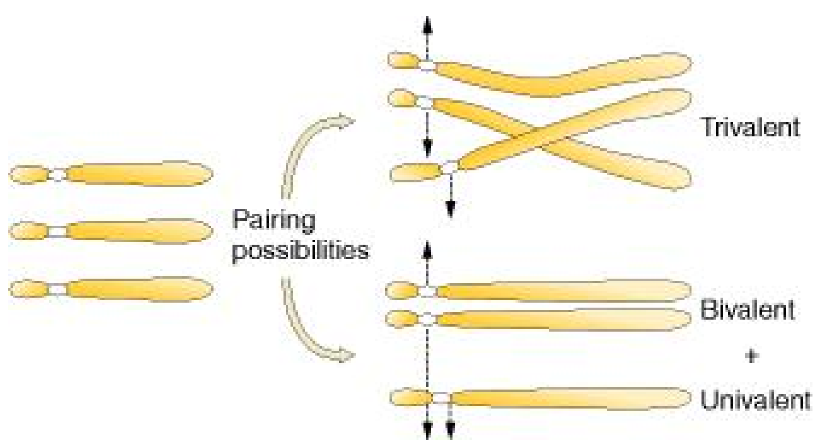


Figure d. Two possibilities for the pairing of three homologous chromosomes before the first meiotic division in a triploid. Notice that the outcome will be the same in both cases: one resulting cell will receive two chromosomes and the other will receive just one. The probability that the latter cell can become a functional haploid gamete is very small, however, because, to do so, it would also have to receive only one of the three homologous chromosomes of every other set in the organism. Note that each chromosome is really a pair of chromatids.

Autotetraploids

Autotetraploids arise naturally by the spontaneous accidental doubling of a 2*x* genome to a 4*x* genome, and autotetraploidy can be induced artificially through the use of colchicine. Autotetraploid plants are advantageous as commercial crops because, in plants, the larger number of chromosome sets often leads to increased size. Cell size, fruit size, flower size, stomata size, and so forth, can be larger in the polyploidy.

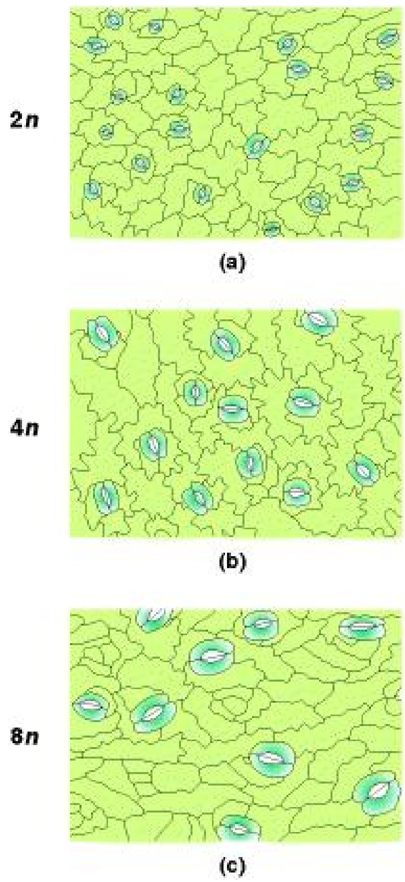


Figure e. Epidermal leaf cells of tobacco plants, showing an increase in cell size, particularly evident in stoma size, with an increase in autopolyploidy. (a) Diploid, (b) tetraploid, (c) octoploid. (From W. Williams, *Genetic Principles and Plant Breeding.* Blackwell Scientific Publications, Ltd.)

Here we see another effect that must be explained by gene numbers. Presumably the amount of gene product (protein or RNA) is proportional to the number of genes in the cell, and this number is higher in the cells of polyploids compared with diploids.

Polyploid plants are often larger and have larger organs than their diploid relatives. Because 4 is an even number, autotetraploids can have a regular meiosis, although this is by no means always the case. The crucial factor is how the four homologous chromosomes, one from each of the four sets, pair and segregate. There are several possibilities, as shown in figure below.

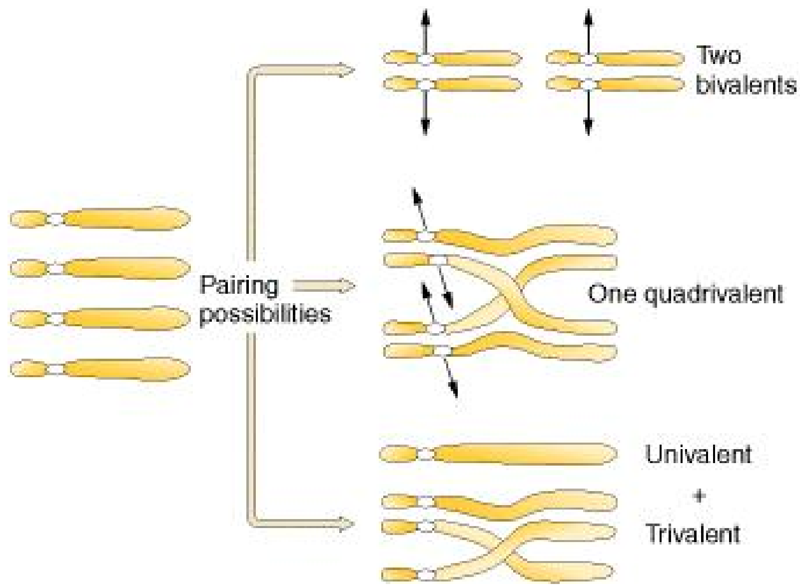


Figure f. Meiotic pairing possibilities in tetraploids. (Each chromosome is really two chromatids.) The four homologous chromosomes may pair as two bivalents or as a quadrivalent. Both possibilities can yield functional gametes. However, the four chromosomes may also pair in a univalent-trivalent combination, yielding nonfunctional gametes. A specific tetraploid can show one or more of these pairings.

Aneuploidy

Aneuploidy is the second major category of chromosome mutations in which chromosome number is abnormal. An **aneuploid** is an individual organism whose chromosome number differs from the wild type by part of a chromosome set. Generally, the aneuploid chromosome set differs from wild type by only one or a small number of chromosomes. Aneuploids can have a chromosome number either greater or smaller than that of the wild type. Aneuploid nomenclature is based on the number of copies of the specific chromosome in the aneuploid state. For example, the aneuploid condition $2n-1$ is called **monosomic** (meaning "one chromosome") because only one copy of some specific chromosome is present instead of the usual two found in its diploid progenitor. The aneuploid 2*n*+1 is called **trisomic**, 2n−2 is **nullisomic**, and n+1 is **disomic.**

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 homoeologous chromosomes apparently compensate for a missing pair of homologs. In fact, all the possible 21 bread wheat nullisomics have been produced. Their appearances differ from the normal hexaploids; furthermore, most of the nullisomics grow less vigorously.

The diploid organisms which have lost a pair of homologous chromosomes are called nullosomics with the genomic formula, 2n-2. The nullosomics exhibit reduced vigor, fertility and survivality, but, polyploidic nullosomics such as nullosomic hexaploid what (6n-2) survive to maturity because of the genetic redundancy in polyploids.

Monosomics (2n−1)

Monosomic chromosome complements are generally deleterious for two main reasons. First, the missing chromosome perturbs the overall gene balance in the chromosome set. 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.

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.

Monosomics show the deleterious effects of genome inbalance, as well as unexpected expression of recessive alleles carried on the monosomic chromosome.

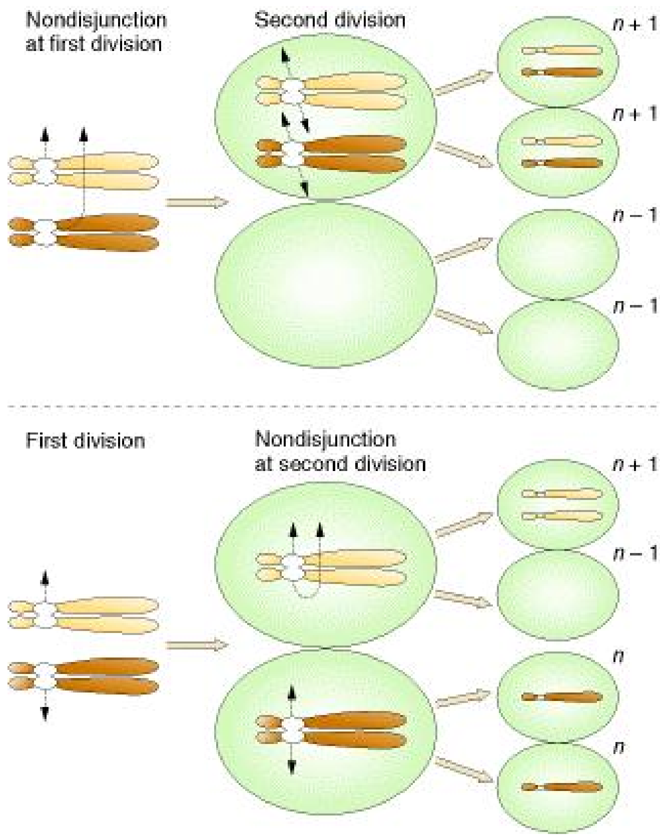


Figure AN 1. The origin of aneuploid gametes by nondisjunction at the first or second meiotic division.

Trisomics

A diploid cell with an extra chromosome. Basically a diploid with an extra chromosome of one type, producing a chromosome number of the form 2n + I. The diploid organisms which have one extra chromosome are called trisomies. They have the chromosomal formula 2n+1. In a trisomic, one of the pairs of chromosomes has an extra member, therefore, forms a trivalent structure during meiosis.During anaphase of meiosis, two chromosomes go to one pole and one chromosome to another pole and thus, two types of gametes n + 1 and n are resulted. The trisomy has variable effects on the phenotype of the organism and in man trisomy of autosome 21 cause mongolism.

In plants, first case of trisomy was investigated in Datura stromonium (Jimson weed) by

Blakeslee and Belting in 1924. D. stramonium normally has 12 pairs of chromosomes in the somatic cells, but in a individual they discovered 25 chromosomes (2n + 1). The size, shape and spine characteristic of seed capsule of this trisomic plant had difference with seed capsule of the wild type. species. Theoretically, because the complement was composed of 12 chromosome pairs differing in the genes they carried, 12 distinguishable trisomies were possible in Jimson weed. Through experimental breeding, Blakeslee and his associates succecceded in producing all 12 possible trisomies. These were grown in Blakeslee's garden and each was found to have a distinguishable phenotype that was attributed to an extra set of the genes contained in one of the 12 chromosomes.

Tetrasomy

When one chromosome of an otherwise diploid organism is present in quadruplicate, the tetrasomy is resulted. The tetrasomics have the .chromosomal formula 20+2. During meiosis a quadrivalent is formed by extra chromosomes and segregation of chromosomes occurs like autotetraploids.

Double Trisomy

In a diploid organism when two different chromosomes are represented in triplicate, the, double, trisomy is resulted. A double trisomic has the chromosomal formula 2n+1+1.

The following indicates in details the types of chromosome mutation where whole genes are moved: