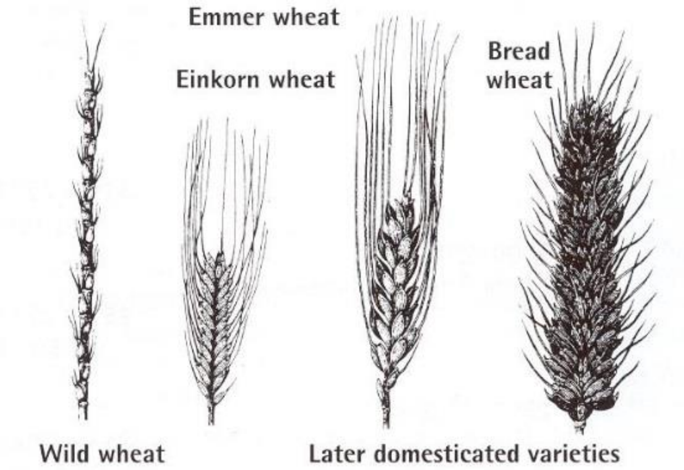
GENETICS - BOT-A-CC-4-10-TH, 4TH SEMESTER HONOURS

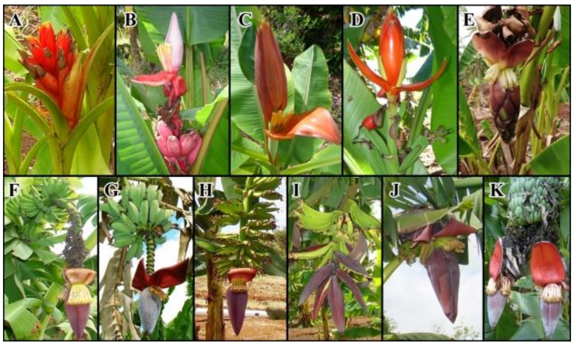
ANEUPLOIDY AND POLYPLOIDY

The cultivation of wheat originated some 10,000 years ago in the Middle East. Today, wheat is the principal food crop for more than a billion people. It is grown in diverse environments, from Norway to Argentina. More than 17,000 varieties have been developed, each adapted to a different locality. Modern cultivated wheat, ***Triticum aestivum***, is a hybrid of at least three different species. Its progenitors were low-yielding grasses that grew in Syria, Iran, Iraq, and Turkey. Some of these grasses appear to have been cultivated by the ancient peoples of this region. What made the triple-hybrid wheats so superior to their ancestors?

The phenotypes of many organisms are affected by changes in the number of chromosomes in their cells; sometimes even changes in part of a chromosome can be significant. These numerical changes are usually described as variations in the ploidy of the organism. Organisms with complete, or normal, sets of chromosomes are said to be **euploid**. Organisms that carry extra sets of chromosomes are said to be **polyploid** and the level of polyploidy is described by referring to a basic chromosome number, usually denoted x. Organisms in which a particular chromosome, or chromosome segment, is under- or overrepresented are said to be **aneuploidy**. These organisms therefore suffer from a specific genetic imbalance.

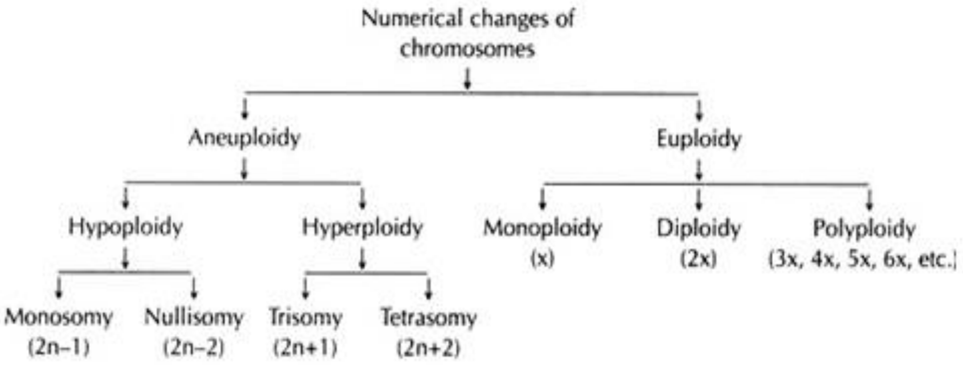
The distinction between aneuploidy and polyploidy is that aneuploidy refers to a numerical change in part of the genome, usually just a single chromosome, whereas polyploidy refers to a numerical change in a whole set of chromosomes. Aneuploidy implies a genetic imbalance, but polyploidy does not.





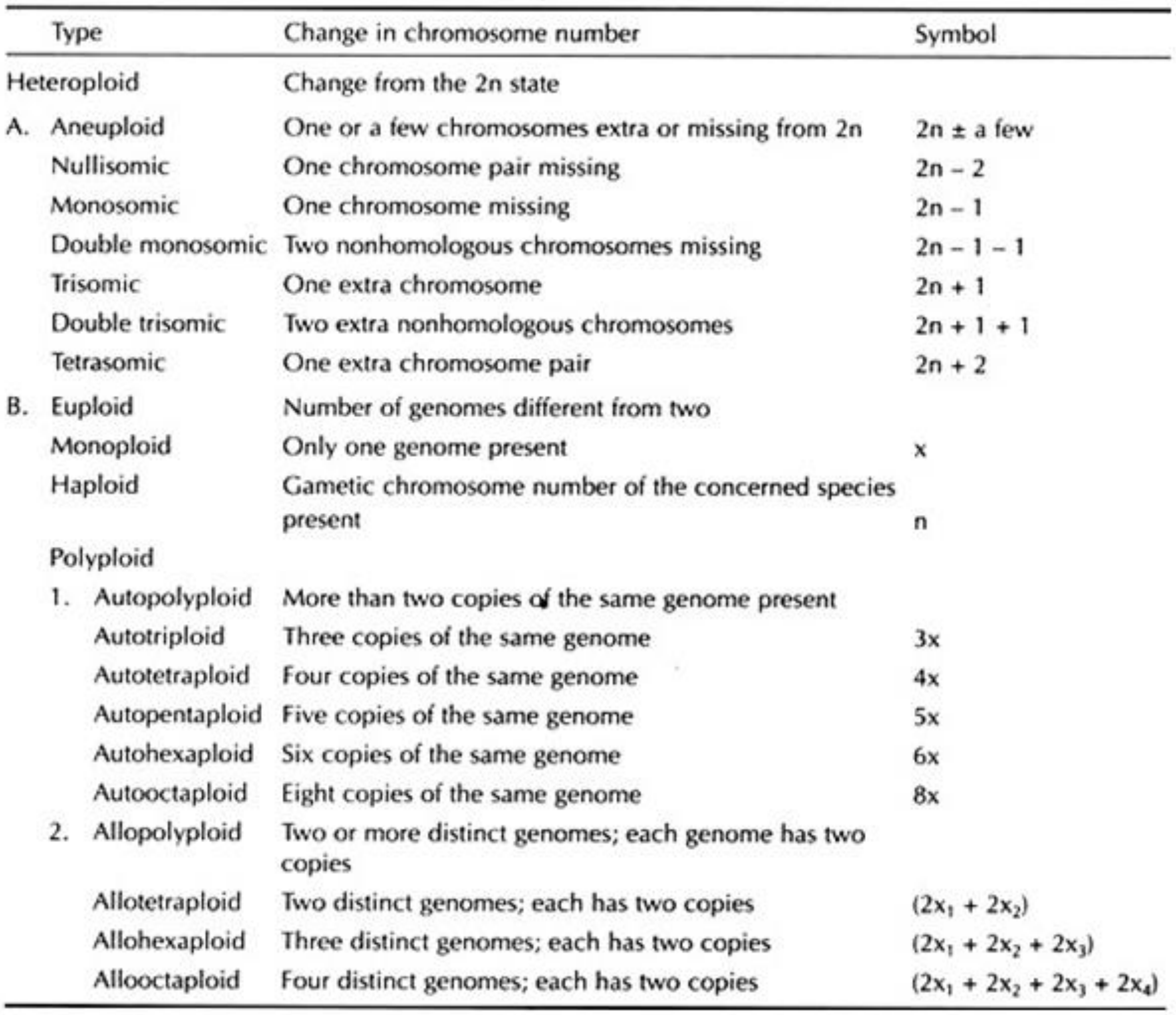
Aneuploidy

Aneuploidy is the presence of chromosome number that is different from the simple multiple of the basic chromosome number. An organism which contains one or more incomplete chromosome sets is known as aneuploid. Aneuploidy can be either due to loss of one or more chromosomes (**hypo-ploidy**) or due to addition of one or more chromosomes to complete chromosome complement (**hyper-ploidy**).



Different kinds of numerical changes of chromosomes (x = basic chromosome number, 2n = somatic chromosome number)

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 ± 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	$(2x_1 + 2x_2 + 2x_3 + 2x_4)$

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) is to be called tetrasomy.

Origin of Aneuploids:

Aneuploids originate from aneuploid gametes. Sometimes abnormal mitosis may also produce aneuploid sectors. Following are the main modes of the origin of aneuploids.

(1) During mitosis or meiosis, abnormalities, e.g., lagging chromosomes result in the formation of nuclei/cells with hypoploid chromosome numbers.

(2) Hypo- and hyper-ploid nuclei may be formed due to chromosome/chromatid nondisjunction during mitotic or meiotic cell division. Such gametes would, on union with normal (n) gametes, give rise to aneuploid progeny.

(3) In the polyploids, especially those with an odd number of chromosome sets, e.g., triploid, pentaploid etc., univalents are often observed during meiosis. The anaphase distribution of the univalents is irregular as a result of which, aneuploid gametes are produced.

(4) Aneuploid cells may be produced due to multipolar mitoses where distribution of chromosomes in the daughter cells is irregular. Such aneuploid cells may possess varying numbers of chromosomes and may occur in the same tissue forming chromosome mosaicism. Such type of aneuploidy is called as multiform-aneuploidy.

(5) Aneuploid gametes may be produced due to nondisjunction during postmeiotic divisions, such as during the formation of microgametophytes and megagametophytes.

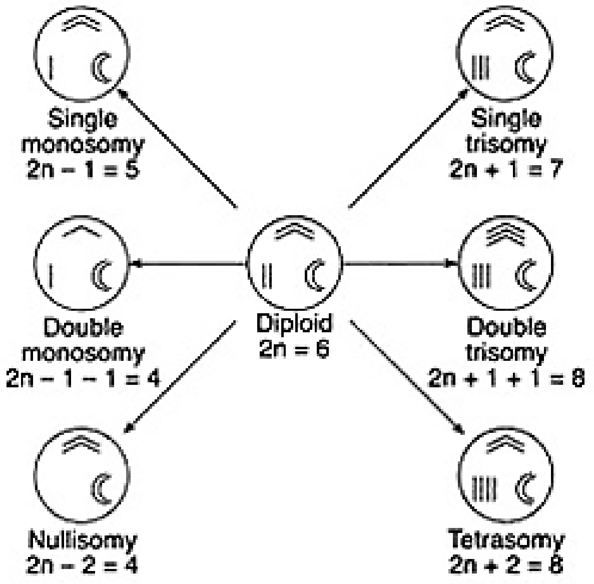
Forms of Aneuploidy:

Monosomy:

Monosomy is the phenomenon where an individual lacks one or a few non-homologous chromosome(s) of a diploid complement.

Types of Monosomy:

Single monosomies lack one com-plete chromosome (2n–1), these create major imbalance and may not be tolerated in diploids. Monosomies, however, are viable in polyploid species, in which the loss of one chromosome has a less marked effect. For instance, in ordinary tobacco, ***Nicotiana tabacum***, which is a tetraploid species with 2n = 48 chromosomes, a series of different monosomic types with 47 chromosomes is known.



The number of possible monosomies in an organism is equal to the haploid chromosome number. Double monosomies (2n – 1 – 1) or triple monosomies (2n – 1 – 1 – 1) could also be produced in polyploids.

Origin of Monosomy:

The origin of the monosomies may be from the production of n – 1 types of gametes due to rare nondisjunction of a bivalent.

Meiotie Behaviour:

Monosomies show irre-gular meiosis (univalents in addition to bivalents). Moreover, in progeny of a monosomic, a mixture of disomic (2n), monosomies (2n – 1) and nuilisomics (2n – 2) is obtained.

Use:

Monosomic condition for a particular chromosome is associated with a characteristic morphology. Looking at the morphology of the monosomies, and of their progeny, genes could be located on a specific chromosome. In wheat, monosomies have been utilized with great suc-cess for the localization of different genes in specific chromosomes by Sears.

Nullisomy:

The plants, in which a chromosome pair is missing, are called nullisomics. The chromo-some formula would be (2n – 2) and not (2n – 1 – 1), which would mean a double monosomic. The number of possible nullisomics in an orga-nism will be equal to the haploid chromosome number.

Origin of Nullisomy:

The origin of nullisomics is gene-rally by the selfing of the monosomies.

Use of Nullisomy:

Nullisomics can be effectively used in locating different genes. In wheat, nullisomics have been obtained with 40 chromosomes instead of 42 chromosomes.

Datura ferox

D. ferox × D. stramonium f. *tatula*

D. stramonium f. *tatula*

D. ferox × D. stramonium f. *stramonium*

D. stramonium f. *stramonium*

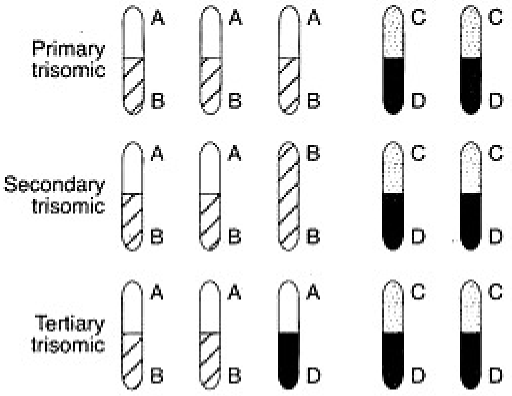
Trisomy:

Trisomies are those organisms, which have an extra chromosome (2n + 1). The number of possible trisomies in an organism is equal to the haploid chromosome number.

Types of Trisomy:

Trisomies are of different types —primary trisomies where extra chromosome is identical to two homologues; secondary trisomies where the extra chromosome is an iso-chromosome with two genetically identical arms; tertiary trisomies are the products of translocation (Fig. 11.3). **Double trisomies (2n + 1 + 1)** are also available in nature.

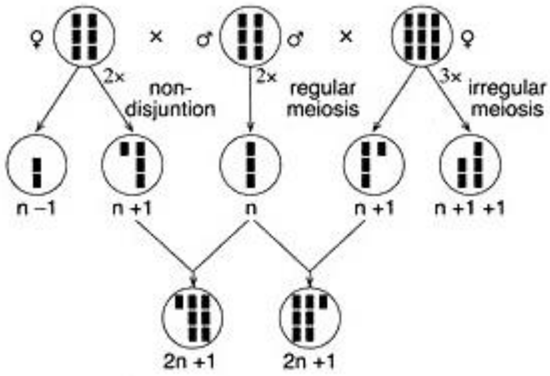
Three Kinds of Trisomics



Three kinds of trisomics

Origin of Trisomy:

The origin of the trisomies may be from the production of n + 1 types of gametes due to rare non-disjunction of a bivalent in a diploid or may also be produced by triploids through irregular meiosis.



Production of trisomics due to formation of n+1 type of gametes in diploid (2x) and triploid (3x) individuals

Meiotic Behaviour:

Trisomies show irregular meiosis. Since the trisomies have an extra chromosome which is homologous to one of the chromosomes of the complement, they form a trivalent.

Blakeslee and Belling obtained trisomic individuals in Datura stramonium hav-ing 25 instead of 24 chromosomes.

Gradually, all the 12 kinds of trisomies, which are theoreti-cally possible, were obtained; each one of the 12 qualitatively different chromosomes in the genome appeared as an extra chromosome.

Use: The trisomies are of significance in locating genes on specific chromosomes. The trisomies have somewhat poorer vigour and less fertility than the normal diploid form.

Tetrasomy:

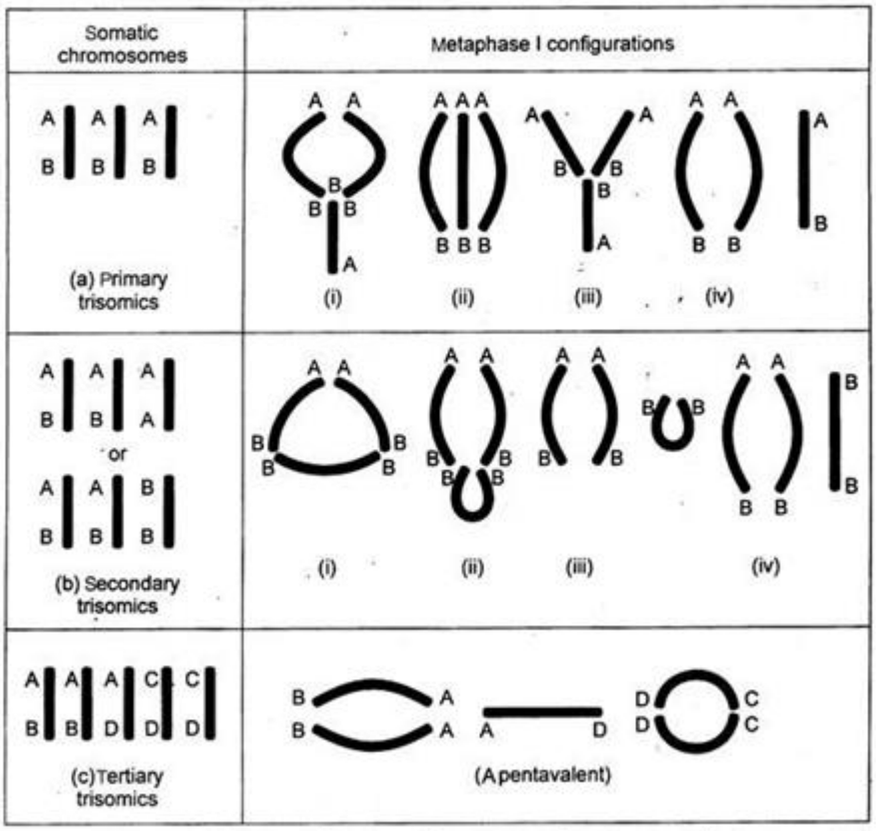
Tetrasomics have a particular chromosome represented in four doses (2n+2). The types of possible tetrasomics are equal to the haploid chro-mosome number of an organism. All 21 possible tetrasomics are available in wheat.

Origin of Tetrasomy:

Tetrasomics may be originated by selfing of trisomies.

Meiotic Behaviour:

During meiosis, the four homologues of the tetrasomic set tend to form a quadrivalent.



Different types of trisomics and their meiotic configurations at metaphase I

Importance of Aneuploidy in Plants:

Aneuploids have played a role in evolution and have importance in plant breeding in addition to genetic analysis.

(a) Detecting linkage group: The aneu-ploids have played an important role in locating a linkage group and a gene in a particular chromosome. Particularly nullisomics, mono-somies and trisomies have been used to deter-mine linkage groups in tobacco, wheat, etc.

The study of aneuploids have shown homoeology between A, B and D genomes of wheat. Identification of the chromosome involved in translocation has also been done with the help of aneuploids.

(b) Chromosome substitution in plant breeding: The major contribution of aneuploids has been in the field of plant breeding. The substitu-tion of whole chromosome or part of the chro-mosome using aneuploids has been done. These substitutions resulted in significant modification of yield, resistance, lodging, etc.

(c) Speciation: Aneuploidy can generate variation and source of speciation in vegetatively propagating species. In Crepis, aneuploid variations form a series $X = 3, 4, 5, 6$ and 7 among species. A very extensive aneuploid series has been observed in *Carex* ($n = 6$ to 56).

Euploidy

Euploidy is the presence of chromosome number which is the multiple of the basic chromosome set. An organism with the basic chromosome number 7, may have euploids with chromosome number 7, 14, 21, 28, 35, 42. Euploids are further of different types – monoploids, diploids and polyploids.

In monoploids there is a single set of genome, in diploids there are two sets of genome and in polyploids there are more than two sets of genome.

Symbols of chromosome number:

$2n$ = Somatic chromosome number of a diploid or polyploid species.

n = Gametic chromosome number of a diploid or polyploid species.

X = Basic chromosome number or genomic number.

In a diploid species with chromosome number $2n = 14$, $n = 7$ as well as $x = 7$.

But in a polyploid (hexaploid) species with chromosome number $2n = 6x = 42$, $n = 21$ but $X = 7$.

Monoploidy and Haploidy:

Monoploid individuals have single basic set of chromosome, e.g., in barley $2n = x = 7$ (haploid of a diploid species). Haploids are individuals with chromosome number half of the somatic number, e.g., in wheat ($2n = 3x = 21$). In diploid species, the chromosome number of monoploids and haploids are same, but in polyploid species the chromosome number of monoploids and haploids are different. In polyploid wheat ($6x = 42$), the haploid is $3x = 21$ and manoploid is $x = 7$.

In flowering plants, the diplophase or the sporophytic phase dominates; the haplophase or gametophytic phase is normally limited to the pollen grains and the embryo sacs. In exceptional cases, plants may arise that are entirely haploid. With regard to all their parts, the haploids are smaller and often display poor vigour.

Depending upon the source of the additional chromosome set, **euploids** are classified into two types, auto-polyploids and allopolyploids which are described below:

(a) Auto-polyploids:

Auto-polyploids arise when the additional sets originate from the same species. For example, if the haploid set of a species is designated A, the diploid is AA, triploid AAA, tetraploid AAAA, and so on.

Autotetraploids can arise through one of the following ways:

(i) Fertilisation of an egg by two or more sperms giving rise to a zygote with three or more sets of chromosomes;

(ii) Normal mitotic division in the diploid zygote in which chromosomes duplicate but cell division fails to occur so that four haploid sets of chromosomes produce a tetraploid nucleus; if the same mitotic error occurs during embryo development it results in some tetraploid tissues in an adult diploid individual;

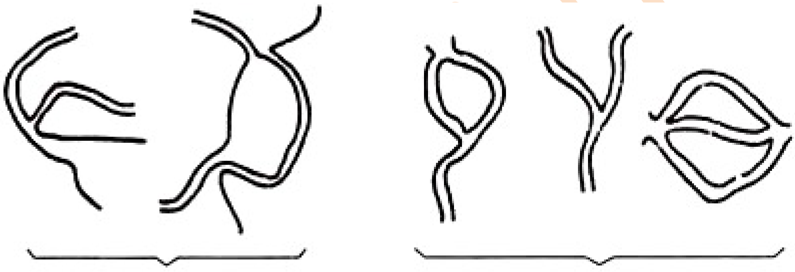
(iii) Failure of meiotic division in germ mother cells so that unreduced diploid gametes are formed instead of haploids.

Although **auto-polypoids** have homologous genomes, yet those having odd numbered sets of chromosomes such as 3n, 5n, 7n, 9n, and so on, show a high degree of sterility. This is because during meiosis, pairing between two homologous chromosomes only results in normal segregation of the haploid set into gametes.

If three homologues are present they may or may not become paired to form a trivalent. Since pairing in any region is restricted to only two homologues at a time, third homologue may fail to pair and remain a univalent, or may pair at some places to form a trivalent.

During anaphasic segregation at meiosis I, two of these homologues may move into one daughter cell, and the third into the other daughter cell. Since all chromosomes of the haploid set have three homologues each, their random distribution or independent assortment will cause the resulting gametes to have varying numbers of different homologues.

In this way a true haploid or a true diploid gamete would be rarely formed. Instead, unbalanced gametes with chromosome numbers ranging between n and 2n would be formed. Such gametes are not viable and triploids consequently are sterile.



Trivalent configurations in meiosis.

It is noteworthy that most popular varieties of seedless watermelons, bananas, Indian carpet grass, and European pears and apples are triploid. These triploid plants have resulted from fertilisation between diploid gametes from tetraploid plants and haploid gametes from diploid plants. Once formed, the triploid plants are healthy and robust and are propagated through asexual cuttings.

Autotetraploids are either fertile or only partially sterile. As there are no univalents and trivalents formed, the four homologues can segregate to form viable diploid gametes, so that tetraploids are often fertile. Groundnut, potatoes and coffee are well known examples of autotetraploid species.

Among cereals, autotetraploid rye is grown in Sweden and Germany. Some of the giant sized plants of Oenothera lamarckiana which De-Vries first noticed in Holland and attributed to a mutation had later turned out to be autotetraploids.

Polyploidy is more common in plants than in animals. More than 50 per cent of angiosperms are known to be polyploids. There are some explanations for this. Plants are mostly hermaphrodites or bisexual organisms in which sex chromosomes do not play a significant role in normal growth and development. An increase in the number of chromosomes is therefore desirable as it increases phenotypic variability and magnifies the expression of some favourable traits. In animals on the other hand, polyploidy leads to a disturbance in the balance between sex chromosomes and autosomes. An increase in the number of sex chromosomes markedly affects sexual development.

Due to this, polyploidy in animals is restricted to those species which are hermaphrodites such as leaches and earthworms, or those which develop partheno-genetically (without fertilisation of egg), as in shrimps, aphids and some lizards. A second reason why polyploidy is more prevalent in plants is that the problem of sterility is easily overcome through asexual methods of reproduction in plants.

Moreover, if one portion of a diploid plant becomes polyploid, for instance a branch bearing fruits, it is possible to propagate that branch through budding and grafting for raising new plants. Such techniques are obviously not applicable to animals except that individual polyploid cells can be excised and cultured in the laboratory. In mammals liver cells are often polyploid.

Even germ cells such as primary spermatocytes are sometimes polyploid in mouse and in man. In plants cells of the tapetum which nourishes the male gametophyte, and endosperm cells which support the growing embryo, are also polyploid. The root nodules of leguminous plants frequently show polyploidy.

From evolutionary point of view polyploidy has played a significant role in evolution of plant species. The origin of some important crop plants such as barley, potatoes, grass (Dactylis glomerata), lotus and many ornamental plants is due to polyploidy.

(b) **Allopolyploidy**:

This is the second type of euploidy where the additional set of chromosomes comes from a different species. For example, suppose a diploid species with two chromosomes sets AA crosses naturally or artificially with another species BB. The offspring produced would be AB which is viable but sterile.

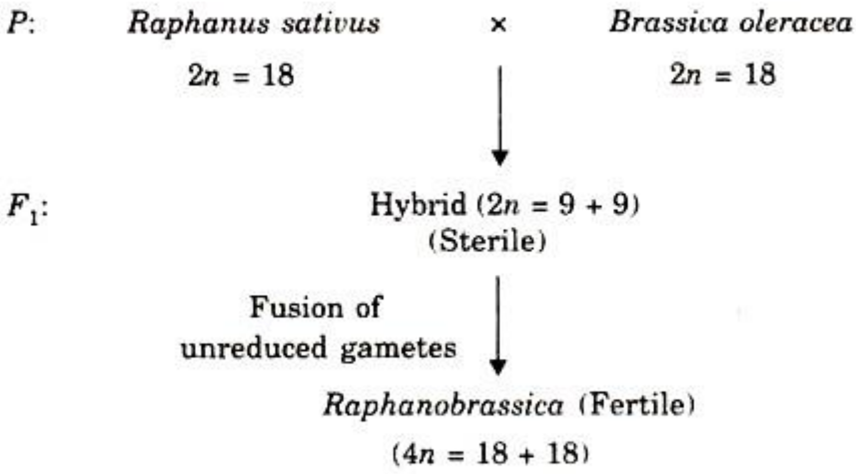
This is because during meiosis the chromosomes belonging to the set A do not find homologous partners in chromosomes of B. Due to failure of pairing at anaphase I, the chromosomes move at random towards the two poles. Thus each gamete gets an unbalanced mixture of A and B chromosomes and sterility results.

There is one way of restoring fertility to a sterile hybrid (AB). If during mitotic division in the AB hybrid all the chromosomes are allowed to divide but cell division is inhibited, the result would be a tetraploid nucleus with two sets of A and two sets of B chromosomes (AABB).

Therefore, when meiosis starts, all chromosomes belonging to one set of A will find homologous partners with the remaining A chromosomes and perfect pairing will result. Similarly, the two sets of B chromosomes will pair with each other and viable fertile gametes would be formed. Such an allopolyploid individual is called an amphidiploid.

It is possible to induce amphidiploidy artificially by treating young buds or seeds with the alkaloid colchicine, a mitotic poison which inhibits spindle formation, consequently cell division. This leads to all the duplicated chromosomes becoming included in a single tetraploid nucleus.

Raphanobrassica is an interesting example of a newly synthesised genus for illustrating allopolyploidy. In 1927 a Russian geneticist Karpechenko made a cross between Raphanus sativus (radish) and Brassica oleracea (cabbage) with the aim of producing a new plant that would have the roots of radish, and in the aerial portions would bear cabbage. The hybrid that was actually formed had the roots of cabbage and tops of radish plant.



The hybrid produced between radish and cabbage proved useless economically. But it proved very important genetically. Both radish and cabbage plants are diploid with 18 chromosomes.

Thus gametes from each parent had 9 chromosomes, and their union produced the F$_1$ hybrid with 18 chromosomes. This hybrid was sterile because the 9 chromosomes of radish did not pair with the 9 chromosomes of cabbage. Sometimes however, viable pollen and ovules were produced having all 18 chromosomes.

Fusion of such unreduced gametes produced tetraploid ($4n = 36$) plants with 18 chromosomes of radish and 18 of cabbage. Pairing took place amongst the radish chromosomes to form 9 pairs; similarly the cabbage chromosomes also formed 9 pairs.

Normal segregation gave rise to viable gametes. The hybrid therefore became fertile and was given the name of a new genus Raphanobrassica. This is a beautiful demonstration of how a new genus can be artificially synthesised through allopolyploidy.

The genus Triticale demonstrates the efforts of man to create a new cereal by crossing wheat and rye. A hexaploid Triticum ($2n + 44$) is crossed to the diploid Secale ($2n = 14$).

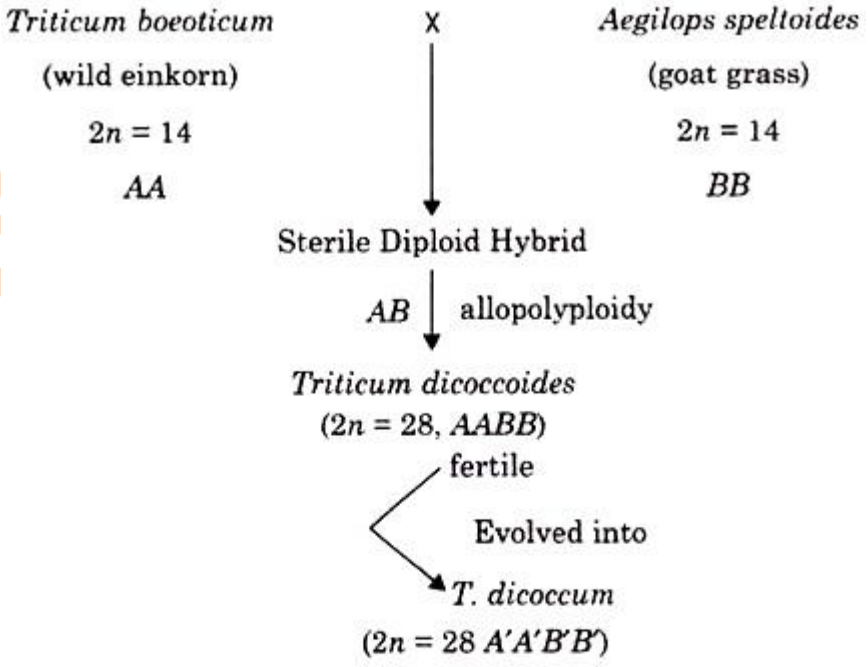
The tetraploid hybrid undergoes chromosome duplication to produce the octoploid Triticale which combines the characters of wheat and rye. It is resistant to diseases affecting both wheat and rye, and the flour made from its grains has very high protein content. Therefore, efforts are being made to develop it for commercial use as a crop plant.

A number of cultivated plants are allopolyploids. One of the most important cereals, wheat, represents an allopolyploid series of diploid, tetraploid and hexaploid species. The series is represented by three groups designated Einkorn (single seeded), Emmer and Vulgare.

The einkorn group consists of two primitive diploid ($2n = 14$) species, namely Triticum monococcum and the wild T. boeoticum. Although not of much use for human consumption because the grain is tightly enclosed in the glumes, the einkorn species are useful as fodder. In some parts of Europe and the Middle East they are used for making dark breads.

The Emmer group consists of seven species of tetraploid wheats of which the most important are Triticum dicoccum (Persian emmer wheat) and T. durum.

The origin of emmer wheats took place through hybridisation between an einkorn wheat and a wild species Aegilops (goat grass) as explained below:



Most of the emmer wheats are grown for animal feed, one T. durum has a high gluten content and is particularly useful for making chapattis in India and noodles in western countries. The vulgare group consists of five species of hexaploid wheats ($2n = 42$) including the economically important bread wheat T. aestivum. It is said to have originated through hybridisation between T. dicoccum (A'A'B'B') and a different species of goat grass Aegilops squarrosa (DD) followed by chromosome doubling. The true bread wheat of today therefore, contains three genomes from three different wheats (A'A'B'B'DD; $2n = 42$).

Bread wheat has 21 pairs of chromosomes which show an interesting behaviour during meiosis. Normally, chromosomes of wheat species coming from different origins do not pair at meiosis. But the chromosomes belonging to A, B and D genomes that are present in hexaploid bread wheat pair with each other under one condition that chromosome No. 5 of B genome should be absent.

Thus chromosome I of A pairs with chromosome I of B; chromosome IA can also pair with ID; and chromosome ID can pair with IB. Similar combinations of pairs exist for other chromosomes of A, B and D genomes. Such chromosomes which belong to different genomes, yet show pairing are called homologous.

Apparently there is a gene on the long arm of chromosome 5B of wheat which suppresses homologous pairing. Riley in 1974 has given the name pairing homologous or ph to this gene. They have also found that at the beginning of meiosis the positions of the chromosomes on the nuclear membrane are determined by this gene, thereby affecting their pairing behaviour.

In cotton it has been possible to trace the origin of American cottons from hybridisation in the past between New World and Old World cottons. The American cultivated cottons have 52 chromosomes whereas the wild American cottons have only 26 chromosomes. The Indian cultivated cottons also have 26 chromosomes but these are morphologically different from the 26 chromosomes of the wild New World varieties.

It appears that sometime in the past the American wild cotton must have crossed with the Old World cultivated cotton to produce a hybrid with 13 New World and 13 Old World chromosomes. Chromosome duplication in this hybrid gave rise to the present day tetraploid ($2n = 52$) cultivated cottons in America.

Clausen and Goodspeed synthesised a new species of Nicotiana (tobacco) by induction of polyploidy as follows:

P: *Nicotiana tabacum* × *N. glutinosa*

$2n = 48$ $2n = 24$

Gametes: $n = 24$ $n = 12$

↓

F_1: Hybrid ($2n = 24 + 12 = 36$)

Triploid, sterile

↓ Colchicine doubling

Nicotiana digluta

($2n = 72$) fertile

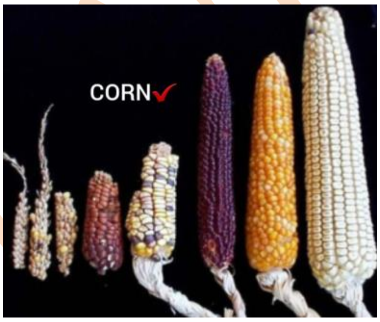
Like auto-polyploids, allopolyploids are also of common occurrence in plants. Among animals they are extremely rare for some well-defined reasons. There are usually no fertilizations of the interspecific type due to different behavioural patterns. Even if fertilisation is induced artificially, the hybrids formed show defects and do not grow normally.

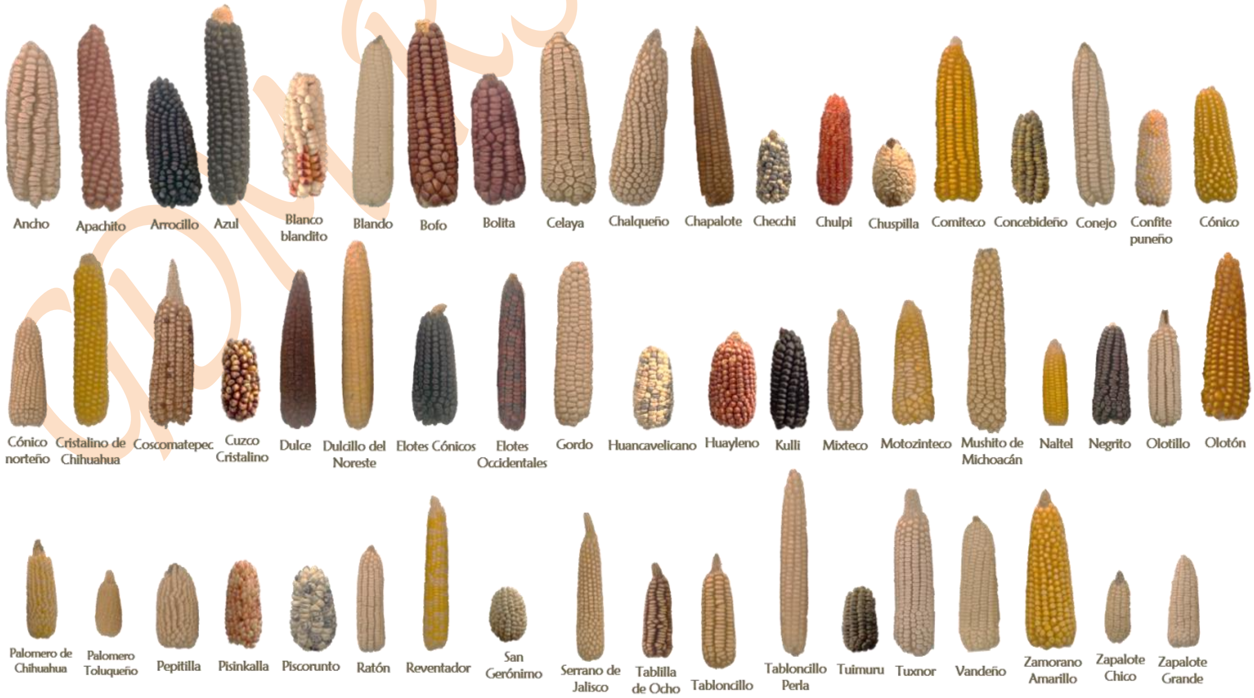
They cannot reproduce vegetatively and do not live long enough to allow chromosome doubling to take place in their germ mother cells to form diploid gametes. Nevertheless, allopolyploidy is found in some animals such as lizards (Cnemidophorns), fishes (Poeciliopsis) and salamanders (Ambystoma), all of which are parthenogenetic.

Polyploidy,

the presence of extra chromosome sets, is fairly common in plants but very rare in animals. One-half of all known plant genera contain polyploid species, and about two-thirds of all grasses are polyploids.

Many of these species reproduce asexually. In animals, where reproduction is primarily by sexual means, polyploidy is rare, probably because it interferes with the sex-determination mechanism. One general effect of polyploidy is that cell size is increased, presumably because there are more chromosomes in the nucleus. Often this increase in size is correlated with an overall increase in the size of the organism. Polyploid species tend to be larger and more robust than their diploid counterparts. These characteristics have a practical significance for humans, who depend on many polyploid plant species for food. These species tend to produce larger seeds and fruits, and therefore provide greater yields in agriculture. Wheat, coffee, potatoes, bananas, strawberries, and cotton are all polyploid crop plants. Many ornamental garden plants, including roses, chrysanthemums, and tulips, are also polyploid.





These are some examples of Polyploid crops, which are used by us in daily life.

STERILE POLYPLOIDS

In spite of their robust physical appearance, many polyploid species are sterile. Extra sets of chromosomes segregate irregularly in meiosis, leading to grossly unbalanced (that is, aneuploid) gametes. If such gametes unite in fertilization, the resulting zygotes almost always die. This in viability among the zygotes explains why many polyploid species are sterile. As an example, let's consider a triploid species with three identical sets of n chromosomes. The total number of chromosomes is therefore 3n. When meiosis occurs, each chromosome will try to pair with its homologues. One possibility is that two homologues will pair completely along their length, leaving the third without a partner; this solitary chromosome is called a univalent. Another possibility is that all three homologues will synapse, forming a trivalent in which each member is partially paired with each of the others. In either case, it is difficult to predict how the chromosomes will move during anaphase of the first meiotic division. The more likely event is that two of the homologues will move to one pole and one homologue will move to the other, yielding gametes with one or two copies of the chromosome. However, all three homologues might move to one pole, producing gametes with zero or three copies of the chromosome. Because this segregational uncertainty applies to each trio of chromosomes in the cell, the total number of chromosomes in a gamete can vary from zero to 3n. Zygotes formed by fertilization with such gametes are almost certain to die; thus, most triploids are completely sterile. In agriculture and horticulture, this sterility is circumvented by propagating the species asexually. The many methods of asexual propagation include cultivation from cuttings (bananas), grafts (Winesap, Gravenstein, and Baldwin apples), and bulbs (tulips). In nature, polyploid plants can also reproduce asexually. One mechanism is apomixis, which involves a modified meiosis that produces unreduced eggs; these eggs then form seeds that germinate into new plants. The dandelion, a highly successful polyploid weed, reproduces in this way.

FERTILE POLYPLOIDS

The meiotic uncertainties that occur in triploids also occur in tetraploids with four identical chromosome sets. Such tetraploids are therefore also sterile. However, some tetraploids are able to produce viable progeny. Close examination shows that these species contain two distinct sets of chromosomes and that each set has been duplicated. Thus, fertile tetraploids seem to have arisen by chromosome duplication in a hybrid that was produced by a cross of two different, but related, diploid species; most often these species have the same or very similar chromosome numbers. Two diploids, denoted A and B, are crossed to produce a hybrid that receives one set of chromosomes from each of the parental species. Such a hybrid will probably be sterile because the A and B chromosomes cannot pair with each other. However, if the chromosomes in this hybrid are duplicated, meiosis will proceed in reasonably good order. Each of the A and B chromosomes will be able to pair with a perfectly homologous partner. Meiotic segregation can therefore produce gametes with a complete set of A and B chromosomes. In fertilization, these “diploid” gametes will unite to form tetraploid zygotes, which will survive because each of the parental sets of chromosomes will be balanced. This scenario of hybridization between different but related species followed by chromosome doubling has evidently occurred many times during plant evolution. In some cases, the process has occurred repeatedly, generating complex polyploids with distinct chromosome sets. One of the best examples is modern bread wheat, ***Triticum aestivum***. This important crop species is a hexaploid containing three different chromosome sets, each of which has been duplicated. There are seven chromosomes in each set, for a total of 21 in the gametes and 42 in the somatic cells. Thus, as we noted at the beginning of this chapter, modern wheat seems to have been formed by two hybridization events. The first involved two diploid species that combined to form a tetraploid, and the second involved a combination between this tetraploid and another diploid, to produce a hexaploid. Cytogeneticists have identified primitive cereal plants in the Middle East that may have participated in this evolutionary process. In 2010, much of the DNA in the wheat genome was sequenced. This genome is very large; roughly five times the size of the human genome. Analysis of all these DNA sequences will help us to understand wheat’s evolutionary history. Because chromosomes from different species are less likely to interfere with each other’s segregation during meiosis, polyploids arising from hybridizations between different species have a much greater chance of being fertile than do polyploids arising from the duplication of chromosomes in a single species. Polyploids created by hybridization between different species are called **allopolyploids** in these polyploids, the contributing genomes are qualitatively different. Polyploids created by chromosome duplication within a species are called **autopolyploids** in these polyploids, a single genome has been multiplied to create extra chromosome sets. Chromosome doubling is a key event in the formation of polyploids. One possible mechanism for this event is for a cell to go through mitosis without going through cytokinesis. Such a cell will have twice the usual number of chromosomes. Through subsequent divisions, it could then give rise to a polyploid clone of cells, which might contribute either to the asexual propagation of the organism or to the formation of gametes. In plants it must be remembered that the germ line is not set aside early in development, as it is in animals. Rather, the reproductive tissues differentiate only after many cycles of cell division. If the chromosomes were accidentally doubled during one of these cell divisions, the reproductive tissues that would ultimately develop might be polyploid. Another possibility is for meiosis to be altered in such a way that unreduced gametes (with twice the normal number of chromosomes) are produced. If such gametes participate in fertilization, polyploid zygotes will be formed. These zygotes may then develop into mature organisms, which, depending on the nature of the polyploidy, may be able to produce gametes themselves. Enhance your understanding of these possibilities by working through Solve It: Chromosome Pairing in Polyploids.

Point to be noted

- Polyploids contain extra sets of chromosomes.
- Many polyploids are sterile because their multiple sets of chromosomes segregate irregularly in meiosis.
- Polyploids produced by chromosome doubling in interspecific hybrids may be fertile if their constituent genomes segregate independently.
- In some somatic tissues—for example, the salivary glands of Drosophila larvae—successive rounds of chromosome replication occur without intervening cell divisions and produce large polytene chromosomes that are ideal for cytogenetic analysis.

Endopolyploidy

Polyploidy occurs in some tissues of animals that are otherwise diploid, such as human muscle tissues. This is known as endopolyploidy. Species whose cells do not have nuclei, that is, prokaryotes, may be polyploid, as seen in the large bacterium *Epulopiscium fishelsoni*. Hence ploidy is defined with respect to a cell.

Neopolyploidy

A polyploid that is newly formed.

Mesopolyploidy

That has become polyploid in more recent history; it is not as new as a neopolyploid and not as old as a paleopolyploid. It is a middle aged polyploid. Often this refers to whole genome duplication followed by intermediate levels of diploidization.

Paleopolyploidy

Ancient genome duplications probably occurred in the evolutionary history of all life. Duplication events that occurred long ago in the history of various evolutionary lineages can be difficult to detect because of subsequent **diploidization** (such that a polyploid starts to behave cytogenetically as a diploid over time) as mutations and gene translations gradually make one copy of each chromosome unlike the other copy. Over time, it is also common for duplicated copies of genes to accumulate mutations and become inactive **pseudogenes**.

In many cases, these events can be inferred only through comparing sequenced genomes. Examples of unexpected but recently confirmed ancient genome duplications include baker's yeast (***Saccharomyces cerevisiae***), mustard weed/thale cress (***Arabidopsis thaliana***), rice (***Oryza sativa***), and an early evolutionary ancestor of the vertebrates (which includes the human lineage) and another near the origin of the teleost fishes. **Angiosperms** (flowering plants) have **paleopolyploidy** in their ancestry. All eukaryotes probably have experienced a polyploidy event at some point in their evolutionary history.

Mixoploidy

Mixoploidy is the case where two cell lines, one diploid and one polyploid, coexist within the same organism. Though polyploidy in humans is not viable, mixoploidy has been found in live adults and children. There are two types: diploid-triploid mixoploidy, in which some cells have 46 chromosomes and some have 69, and diploid-tetraploid mixoploidy, in which some cells have 46 and some have 92 chromosomes. It is a major topic of cytology.

Dihaploidy and polyhaploidy

Dihaploid and **polyhaploid** cells are formed by **haploidisation** of polyploids, i.e., by halving the chromosome constitution.

Dihaploids (which are diploid) are important for selective breeding of tetraploid crop plants (notably potatoes), because selection is faster with diploids than with tetraploids. Tetraploids can be reconstituted from the diploids, for example by somatic fusion.

The term "dihaploid" was coined by Bender to combine in one word the number of genome copies (diploid) and their origin (haploid). The term is well established in this original sense, but it has also been used for doubled monoploids or doubled haploids, which are homozygous and used for genetic research.

Homoploid

Homoploid means "at the same ploidy level", i.e. having the same number of homologous chromosomes. For example, homoploid hybridization is hybridization where the offspring have the same ploidy level as the two parental species. This contrasts with a common situation in plants where chromosome doubling accompanies or occurs soon after hybridization. Similarly, homoploid speciation contrasts with polyploid speciation.

Zygoidy and azygoidy

Zygoidy is the state in which the chromosomes are paired and can undergo meiosis. The zygoid state of a species may be diploid or polyploid. In the azygoid state the chromosomes are unpaired. It may be the natural state of some asexual species or may occur after meiosis. In diploid organisms the azygoid state is monoploid.

Haplodiploidy

Ploidy can also vary between individuals of the same species or at different stages of the life cycle. In some insects it differs by caste. In humans, only the gametes are haploid, but in many of the social insects, including ants, bees, and termites, certain individuals develop from unfertilized eggs, making them haploid for their entire lives, even as adults. In the Australian bulldog ant, *Myrmecia pilosula*, a haplodiploid species, haploid individuals of this species have a single chromosome and diploid individuals have two chromosomes. In Entamoeba, the ploidy level varies from 4n to 40n in a single population. Alternation of generations occurs in most plants, with individuals "alternating" ploidy level between different stages of their sexual life cycle.

Tissue-specific polyploidy

In large multicellular organisms, variations in ploidy level between different tissues, organs, or cell lineages are common. Because the chromosome number is generally reduced only by the specialized process of meiosis, the somatic cells of the body inherit and maintain the chromosome number of the zygote by mitosis. However, in many situations somatic cells double their copy number by means of endoreduplication as an aspect of cellular differentiation. For example, the hearts of two-year-old human children contain 85% diploid and 15% tetraploid nuclei, but by 12 years of age the proportions become approximately equal, and adults examined contained 27% diploid, 71% tetraploid and 2% octaploid nuclei.

Adaptive and ecological significance of variation in ploidy

There is continued study and debate regarding the fitness advantages or disadvantages conferred by different ploidy levels. A study comparing the karyotypes of endangered or invasive plants with those of their relatives found that being polyploid as opposed to diploid is associated with a 14% lower risk of being endangered, and a 20% greater chance of being invasive. Polyploidy may be associated with increased vigor and adaptability. Some studies suggest that selection is more likely to favor diploidy in host species and haploidy in parasite species.

When a germ cell with an uneven number of chromosomes undergoes meiosis, the chromosomes cannot be evenly divided between the daughter cells, resulting in aneuploid gametes. Triploid organisms, for instance, are usually sterile. Because of this, triploidy is commonly exploited in agriculture to produce seedless fruit such as bananas and watermelons. If the fertilization of human gametes results in three sets of chromosomes, the condition is called triploid syndrome.

For further reading:

https://www.nature.com/scitable/topicpage/polyploidy-1552814/
https://www.sciencedirect.com/topics/agricultural-and-biological-sciences/polyploidy
https://www.frontiersin.org/articles/10.3389/fgene.2019.00807/full
https://www.ncbi.nlm.nih.gov/pubmed/550830
Snustad, D. P.; et al. (2006). Principles of Genetics (4th ed.). Hoboken, NJ: John Wiley & Sons. ISBN 978-0-471-69939-2.