Course Code-EDU161 and Course Name:Botany

Aneuploidy: Meaning, Forms and Importance | Cell Biology

Meaning of 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).

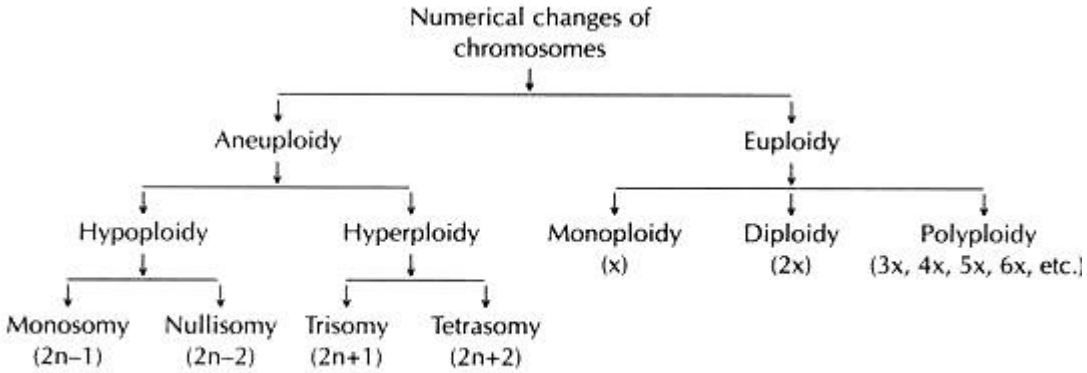


Fig. 11.1: Different kinds of numerical changes of chromosomes (x = basic chromosome number, 2n = somatic chromosome number)

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 ± 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) – tetrasomy (Fig. 11.2).

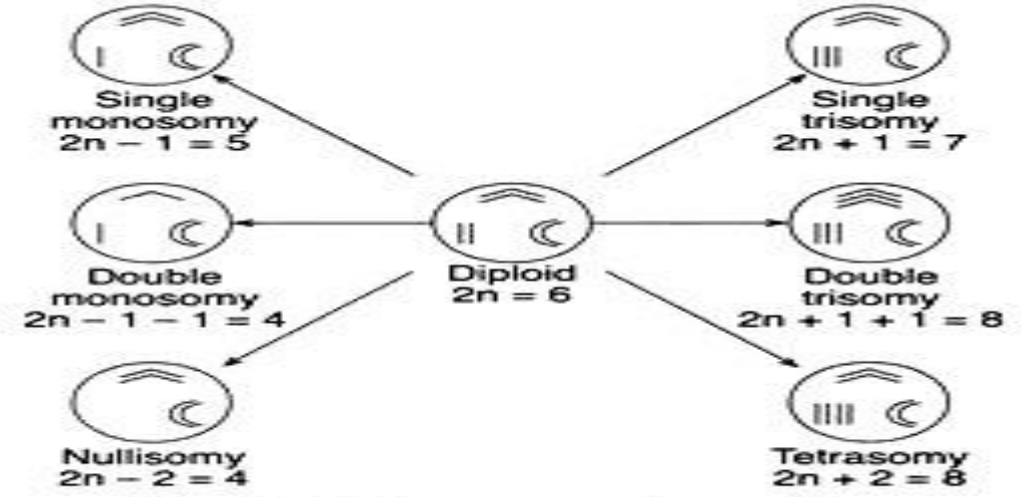


Fig. 11.2 : Different types of aneuploids

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 complete 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 irregular 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 success 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 chromosome formula would be (2n – 2) and not (2n – 1 – 1), which would mean a double monosomic. The number of possible nullisomics in an organism will be equal to the haploid chromosome number.

Origin of Nullisomy

The origin of nullisomics is generally 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.

Trisomy:

Trisomies are those organisms, which have an extra chromosome (2n -t- 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.

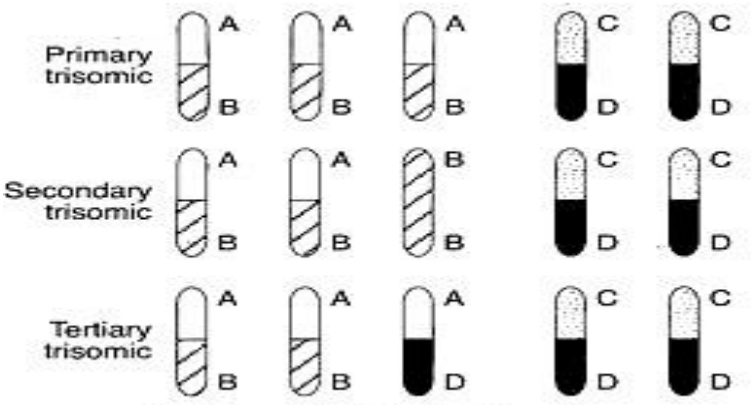


Fig. 11.3 : Three kinds of trisomics

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 (Fig. 11.4).

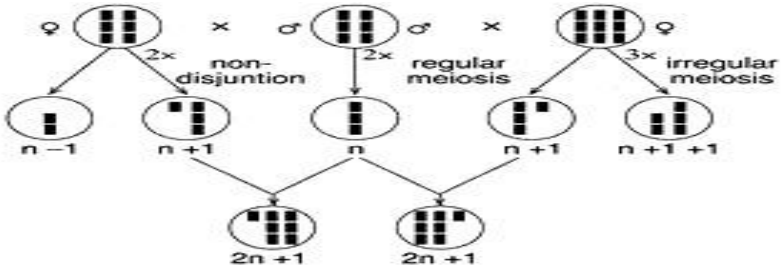


Fig. 11.4: Production of trisomics due to formation of n+1 type of gametes in diploid (2x) and triploid (3x) individuals

Meiotic Behaviour:

Trisomies show irregular' meiosis (Fig. 11.5). 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 having 25 instead of 24 chromosomes.

Gradually, all the 12 kinds of trisomies, which are theoretically 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 is equal to the haploid chromosome 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.

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 aneuploids have played an important role in locating a linkage group and a gene in a particular chromosome. Particularly nullisomics, monosomies and trisomies have been used to determine 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 substitution of whole chromosome or part of the chromosome 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).

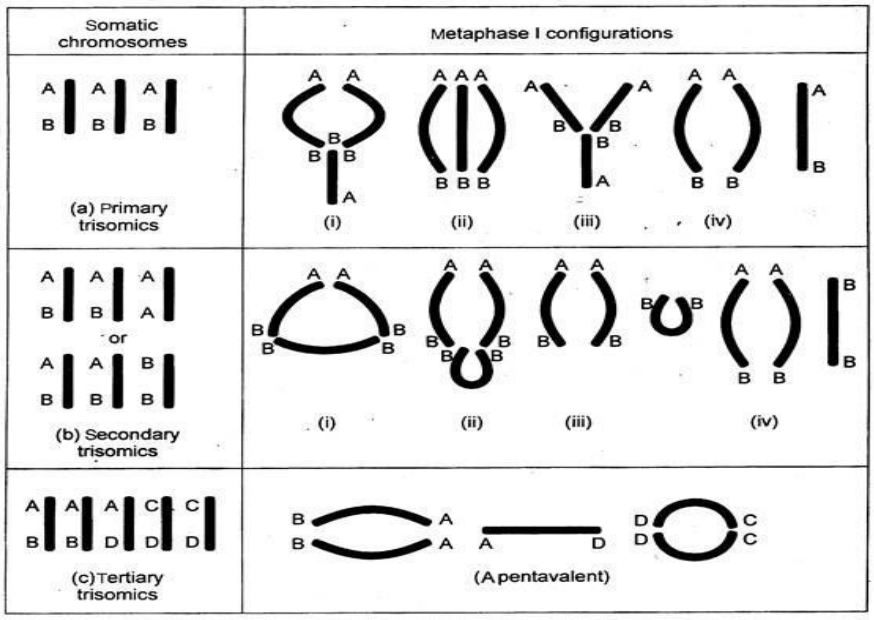


Fig. 11.5: Different types of trisomics and their meiotic configurations at metaphase I

Polyploidy

It is common among plants and has been, in fact, a major source of speciation in the angiosperms. Particularly important is allopolyploidy, which involves the doubling of chromosomes in a hybrid plant. Normally a hybrid is sterile because it does not have the required homologous pairs of chromosomes for successful gamete formation during meiosis. If through polyploidy, however, the plant duplicates the chromosome set inherited from each parent, meiosis can occur, because each chromosome will have a homologue derived from its duplicate set. Thus, polyploidy confers fertility on the formerly sterile hybrid, which thereby attains the status of a full species distinct from either of its parents. It has been estimated that up to half of the known angiosperm species arose through polyploidy, including some of the species most prized by man. Plant breeders utilize this process, treating desirable hybrids with chemicals, such as colchicine, that are known to induce polyploidy.

Polyploid animals are far less common, and the process appears to have had little effect on animal speciation.

Advantages of Polyploidy

Due to the high incidence of polyploidy in some taxa, such as plants, fish, and frogs, there clearly must be some advantages to being polyploid. A common example in plants is the observation of hybrid vigor, or heterosis, whereby the polyploid offspring of two diploid progenitors is more vigorous and healthy than either of the two diploid parents. There are several possible explanations for this observation. One is that the enforced pairing of homologous chromosomes within an allotetraploid prevents recombination between the genomes of the original progenitors, effectively maintaining heterozygosity throughout generations (Figure 3). This heterozygosity prevents the accumulation of recessive mutations in the genomes of later generations, thereby maintaining hybrid vigor. Another important factor is gene redundancy. Because the polyploid offspring now have twice as many copies of any particular gene, the offspring are shielded from the deleterious effects of recessive mutations. This is particularly important during the gametophyte life stage. One might envision that, during the haploid stage of the life cycle, any allele that is recessive for a

deleterious mutation will not be masked by the presence of a dominant, normally functioning allele, allowing the mutation to cause developmental failure in the pollen or the egg sac. Conversely, a diploid gamete permits the masking of this deleterious allele by the presence of the dominant normal allele, thus protecting the pollen or egg sac from developmental dysfunction. This protective effect of polyploidy might be important when small, isolated populations are forced to inbreed.
Another advantage conferred by gene redundancy is the ability to diversify gene function over time. In other words, extra copies of genes that are not required for normal organism function might end up being used in new and entirely different ways, leading to new opportunities in evolutionary selection (Adams & Wendel, 2005).
Interestingly, polyploidy can affect sexuality in ways that provide selective advantages. One way is by disrupting certain self-incompatibility systems, thereby allowing self-fertilization. This might be the result of the interactions between parental genomes in allopolyploids (Comai *et al.*, 2000). Another way is by favoring the onset of asexual reproduction, which is associated with polyploidy in both plants and animals. This switch in reproductive strategies may improve fitness in static environments.

Types

Polyploid types are labeled according to the number of chromosome sets in the nucleus. The letter *x* is used to represent the number of chromosomes in a single set.

- **triploid** (three sets; 3*x*), for example sterile saffron crocus, or seedless watermelons, also common in the phylum Tardigrada[8]
- **tetraploid** (four sets; 4*x*), for example Salmonidae fish,[9] the cotton *Gossypium hirsutum*[10]
- **pentaploid** (five sets; 5*x*), for example Kenai Birch (*Betula papyrifera* var. *kenaica*)
- **hexaploid** (six sets; 6*x*), for example wheat, kiwifruit[11]
- **heptaploid** or **septaploid** (seven sets; 7*x*)
- **octaploid** or **octoploid**, (eight sets; 8*x*), for example *Acipenser* (genus of sturgeon fish), dahlias
- **decaploid** (ten sets; 10*x*), for example certain strawberries
- **dodecaploid** (twelve sets; 12*x*), for example the plants *Celosia argentea* and *Spartina anglica*[12] or the amphibian *Xenopus ruwenzoriensis*.

Autopolyploidy

Autopolyploids are polyploids with multiple chromosome sets derived from a single taxon. Two examples of natural autopolyploids are the piggyback plant, *Tolmiea menzisii*[64] and the white sturgeon, *Acipenser transmontanum*.[65] Most instances of autopolyploidy result from the fusion of unreduced ($2n$) gametes, which results in either triploid ($n + 2n = 3n$) or tetraploid ($2n + 2n = 4n$) offspring.[66] Triploid offspring are typically sterile (as in the phenomenon of 'triploid block'), but in some cases they may produce high proportions of unreduced gametes and thus aid the formation of tetraploids. This pathway to tetraploidy is referred to as the "triploid bridge".[66] Triploids may also persist through asexual reproduction. In fact, stable autotriploidy in plants is often associated with apomictic mating systems.[67] In agricultural systems, autotriploidy can result in seedlessness, as in watermelons and bananas.[68] Triploidy is also utilized in salmon and trout farming to induce sterility.[69][70]

Rarely, autopolyploids arise from spontaneous, somatic genome doubling, which has been observed in apple (*Malus domesticus*) bud sports.[71] This is also the most common pathway

of artificially induced polyploidy, where methods such as protoplast fusion or treatment with colchicine, oryzalin or mitotic inhibitors are used to disrupt normal mitotic division, which results in the production of polyploid cells. This process can be useful in plant breeding, especially when attempting to introgress germplasm across ploidal levels.[72]

Autopolyploids possess at least three homologous chromosome sets, which can lead to high rates of multivalent pairing during meiosis (particularly in recently formed autopolyploids, also known as neopolyploids) and an associated decrease in fertility due to the production of aneuploid gametes.[73] Natural or artificial selection for fertility can quickly stabilize meiosis in autopolyploids by restoring bivalent pairing during meiosis, but the high degree of homology among duplicated chromosomes causes autopolyploids to display polysomic inheritance.[74] This trait is often used as a diagnostic criterion to distinguish autopolyploids from allopolyploids, which commonly display disomic inheritance after they progress past the neopolyploid stage.[75] While most polyploid species are unambiguously characterized as either autopolyploid or allopolyploid, these categories represent the ends of a spectrum between of divergence between parental subgenomes. Polyploids that fall between these two extremes, which are often referred to as segmental allopolyploids, may display intermediate levels of polysomic inheritance that vary by locus.[76][77]

About half of all polyploids are thought to be the result of autopolyploidy,[78][79] although many factors make this proportion hard to estimate.[80]

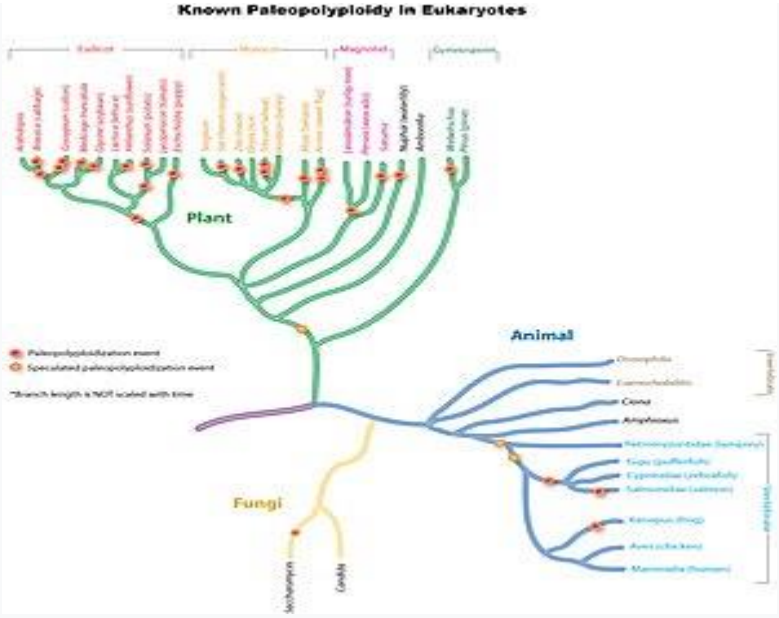
Allopolyploidy

Allopolyploids or amphipolyploids or **heteropolyploids** are polyploids with chromosomes derived from two or more diverged taxa. As in autopolyploidy, this primarily occurs through the fusion of unreduced (2*n*) gametes, which can take place before or after hybridization. In the former case, unreduced gametes from each diploid taxa – or reduced gametes from two autotetraploid taxa – combine to form allopolyploid offspring. In the latter case, one or more diploid F_1 hybrids produce unreduced gametes that fuse to form allopolyploid progeny.[81] Hybridization followed by genome duplication may be a more common path to allopolyploidy because F_1 hybrids between taxa often have relatively high rates of unreduced gamete formation – divergence between the genomes of the two taxa result in abnormal pairing between homoeologous chromosomes or nondisjunction during meiosis.[81] In this case, allopolyploidy can actually restore normal, bivalent meiotic pairing by providing each homoeologous chromosome with its own homologue. If divergence between homoeologous chromosomes is even across the two subgenomes, this can theoretically result in rapid restoration of bivalent pairing and disomic inheritance following allopolyploidization. However multivalent pairing is common in many recently formed allopolyploids, so it is likely that the majority of meiotic stabilization occurs gradually through selection.[73][75]

Because pairing between homoeologous chromosomes is rare in established allopolyploids, they may benefit from fixed heterozygosity of homoeologous alleles.[82] In certain cases, such heterozygosity can have beneficial heterotic effects, either in terms of fitness in natural contexts or desirable traits in agricultural contexts. This could partially explain the prevalence of allopolyploidy among crop species. Both bread wheat and *Triticale* are examples of an allopolyploids with six chromosome sets. Cotton, peanut, or quinoa are allotetraploids with multiple origins. In Brassicaceous crops, the Triangle of U describes the relationships between the three common diploid Brassicas (*B. oleracea*, *B. rapa*, and *B. nigra*) and three allotetraploids (*B. napus*, *B. juncea*, and *B. carinata*) derived from hybridization among the diploid species. A similar relationship exists between three diploid species of *Tragopogon* (*T. dubius*, *T. pratensis*, and *T. porrifolius*) and two allotetraploid species (*T. mirus* and *T.*

miscellus).[83] Complex patterns of allopolyploid evolution have also been observed in animals, as in the frog genus *Xenopus*.[84]

Paleopolyploidy



This phylogenetic tree shows the relationship between the best-documented instances of paleopolyploidy in eukaryotes.

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.[85]

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[26]. Angiosperms (flowering plants) have paleopolyploidy in their ancestry. All eukaryotes probably have experienced a polyploidy event at some point in their evolutionary history.

References:

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