RAMA UNIVERSITY

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FACULTY OF AGRICULTURAL SCIENCES

AND ALLIED INDUSTRIES

STRUCTURAL CHROMOSOMAL ABERRATIONS

Structural chromosomal aberrations cause structural abnormalities in chromosome structure. They alter the sequence or the kind of genes present in chromosome. These are further classified into four groups based upon whether they alter the gene sequences, number or location. Changes in the structure of chomosomes. a. Loss or addition of segments of chromosomes. Deletion (deficiency) - Loss of a segment of a chromosome Duplication - repetition of a segment of a chromosome. b. Changes in the normal arrangement of genes in the chromosome. Translocation – Exchange of segments between two non - homologous chromosomes, resulting in new chromosomes. Inversion – Change in the linear order of genes by rotation of a section of a chromosome through 180 degrees. Gene mutations or point mutations – changes in the nucleotide sequence of a gene. a. Deletion b. Insertion c. Substitution d. Inversion a. Loss or addition of segments of chromosomes Deletion of a Gene As the name implies, genes of a chromosome are permanently lost as they become unattached to the centromere and are lost forever Normal chromosome before mutation • Genes not attached to centromere become loose and lost forever • New chromosome lacks certain genes which may prove fatal depending on how important these genes are

Duplication of Genes In this mutation, the mutants genes are displayed twice on the same chromosome due to duplication of these genes. This can prove to be an advantageous mutation as no genetic information is lost or altered and new genes are gained Normal chromosome before mutation • Genes from the homologous chromosome are copied and inserted into the genetic sequence • New chromosome possesses all its initial genes plus a duplicated one, which is usually harmless The next page continues looking at these chromosome mutations and mutations that happen within genes that can prove to be more harmful to the organism at hand. The following pages also investigates polyploidy in species

b. Changes in the normal arrangement of genes in the chromosome Inversion of Genes This is where the order of a particular order of genes are reversed as seen below • Normal chromosome un-altered • The connection between genes break and the sequence of these genes are reversed • The new sequence may not be

viable to produce an organism, depending on which genes are reversed. Advantageous characteristics from this mutation are also possible

Translocation of Genes This is where information from one of two homologous chromosomes breaks and binds to the other. Usually this sort of mutation is lethal • An un-altered pair of homologous chromosomes • Translocation of genes has resulted in some genes from one of the chromosomes attaching to the opposing chromosome

II. Gene mutations or point mutations Alteration of a DNA Sequence The previous examples of mutation have investigated changes at the chromosome level. The sequence of nucleotides on a DNA sequence are also susceptible to mutation. Substitution A substitution is a mutation that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G). Such a substitution could: 1. change a codon to one that encodes a different amino acid and cause a small change in the protein produced. For example, sickle cell anemia is caused by a substitution in the beta-hemoglobin gene, which alters a single amino acid in the protein produced. 2. change a codon to one that encodes the same amino acid and causes no change in the protein produced. These are called silent mutations. change an amino-acid-coding codon to a single "stop" codon and cause an incomplete protein. This can have serious effects since the incomplete protein probably won't function.

Insertion Insertions are mutations in which extra base pairs are inserted into a new place in the DNA.

Deletion Deletions are mutations in which a section of DNA is lost, or deleted

Inversion Frameshift Since protein-coding DNA is divided into codons three bases long, insertions and deletions can alter a gene so that its message is no longer correctly parsed. These changes are called frameshifts. For example, consider the sentence, "The fat cat sat." Each word represents a codon. If we delete the first letter and parse the sentence in the same way, it doesn't make sense. In frameshifts, a similar error occurs at the DNA level, causing the codons to be parsed incorrectly. This usually generates truncated proteins that are as useless as "hef atc ats at" is uninformative

Genetic mutations increase genetic diversity and therefore have an important part to play. They are also the reason many people inherit diseases.

NUMERICAL CHROMOSOMAL ABBERATIONS

POLY PLOIDS

Individual with one set of chromosomes – haploid or Monoploids.

Individual with two sets of chromosomes – True diploid

Most plants and animals posses two sets of chromosomes.

Individuals with more than two sets of chromosomes are called ‘ polyploid’. (poly = many; ploid – fold). Poly ploid in otherwise called as ‘Euploidy’.

HAPLOID (Monoploid)

Haploidy in flowering plants was first recorded by Blakeslee (1937) in *Datura stramonium* . One in very 1000 seedlings of maize was found to be a haploid, developed from the unfertilized egg (i.e by female parthenogenesis).

Polyembryony in plants in a possible source of haploids, due to the occurrence of more than one embryosac within a ovule. Out of the four haploid megaspores derived by meiotic divisions from a single megaspore mother cell, more than one may develop into embryo sacs.

Production of embryos from synergids without fertilization is more common than production of embryos from antipodal. For instance, out of about 30,000 seeds of G. Girsutum, 20 were found to give rise to twin embryos of which from were haploids.

Haploids can be obtained by anther and pollenculture using tissue culture techniques and also by wide species crosses.

Characteristics of haploids

Generally smaller in size than diploids. Their guard cells are also smaller than those of diploids. They are highly sterile because none of chromosomes of true haploid has a homologue.

Classification of haploids : 1. Mono-haploid. 2. Poly haploids.

Mono-haploids: are haploids which arise from time diploids and whose chromosomes are therefore non-homologous to one another. Eg. haploids of maize.

Polyhaploids: Haploids arise from polyploids eg; Haploid of triticum aestivum with one representative of each chromosomes of A. B and D genome.

GENOME

The complete set of chromosome found in the gamete of a true diploid is called a genome. e.g. p. glaucm, 14 chromosome 2 sets n=7, 2n=14.

A gamete contains one set of seven chromosomes (I, II, III, IV, V, VI, VII) is called a genome and if this is represented as ‘ A’ the genomic constitution of the plant in ‘ AA’.

BASIC NUMBER

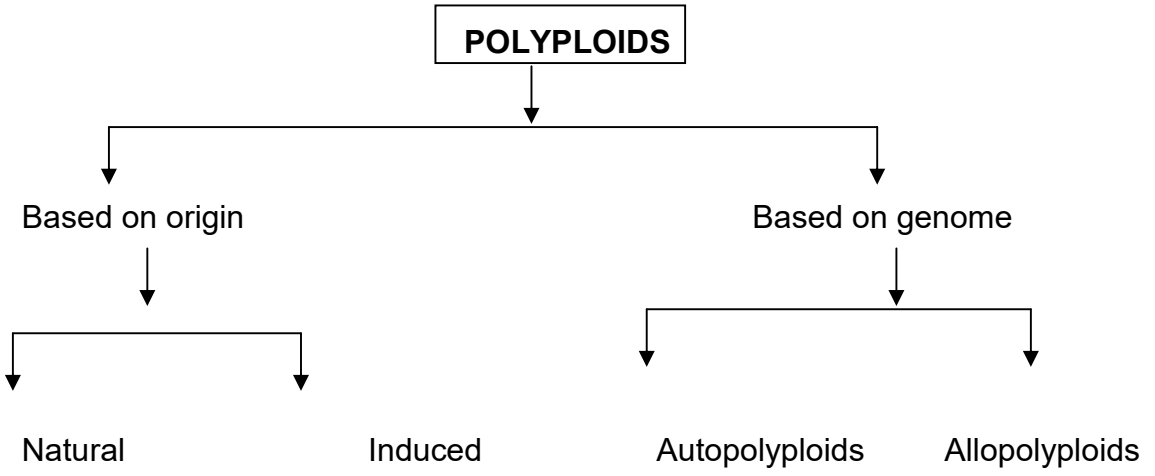
The number of chromosomes constituting a genome is called the basic number. It is the number of chromosomes found in the gamete of a true diploid.

POLYPLOID

Species of solanum which have a chromosome number of 24 are true diploid and species which have somatic no higher than 24, but which are multiples of 12, are called polyploids.

Somatic No. (2n)	Multiples of Basic No.(x-12)	Level of ploidy
24	2x	Diploid
36	3x	Triploid
48	4x	Tetraploid
60	5x	Pentaploid
72	6x	Hexaploid

CLASSIFICATION



Natural polyploids: Polyploids arise in nature by failure of meiosis that result in formation of unreduced gametes. They may also be formed from somatic cells

in which a failure of mitosis has resulted in doubling of the chromosome complement. The cultivated banana and tobacco are examples of natural polyploids.

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 nondisjunction, 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 planted in soil.

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.

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.

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. P1: Species X X Species Y (AA) ↓ (BB) (Diploid) (Diploid) F1: 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.

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).

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 2n+1 is called trisomic,
- 2n−2 is nullisomic, 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.

Note: Non-disjunction 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. Non-disjunction 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.

- 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 + l. The diploid organisms which have one extra chromosome are called trisomics. 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. 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 trisomics were possible in Jimson weed. Through experimental breeding, Blakeslee and his associates succeeded 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.

IMPORTANT QUESTIONS:

1. Define chromosomal abberations and its types.
2. Describe structural chromosomal abberations with examples.
3. Describe numerical chromosomal abberations with their roles.
4. Discuss in detail about the role of aneuploids.
5. Differentiate betweem euploids and aneuploids.
6. Differentiate between autoployploids and allopolyploids.