ANEUPLOIDY: A REDEFINITION

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ABSTRACT. A redefinition of the term "aneuploidy" is proposed which will retain the original intended meaning, while accommodating cytological situations encountered since Täckholm (1922) first used the term. Definitions in the literature are frequently at variance, or, where purely numerical criteria are used, inadequate. The term is here defined with reference to the "basic chromosome set". "Aneuploidy" is confined to unbalanced situations (trisomics etc.) whilst the term "dysploidy" is used for the step-wise progression in basic number called an "aneuploid series" by many authors. A new distinction is made between "chromosomal aneuploidy" and "segmental aneuploidy", the latter including "centric" and "acentric" types. These terms are defined and discussed. Their relationship with each other is summarised diagramatically in fig. 1, and with the existing terms "dysploidy" and "heteroploidy" in fig. 2.

The term 'aneuploid' was first applied by Täckholm (1922) to the chromosome numbers of certain polyploid rose hybrids which were not straight multiples of a basic number, and the term 'euploid' for those that were. Since then an increasing number of cytological situations has been discovered which, though as distinct in origin and significance as, for example, trisomy, B-chromosomes, sex-chromosomes and dysploidy (Tischler, 1937), are all aneuploid according to this original definition. Thus, Stebbins (1950) and John & Lewis (1968), for example, use the same term for trisomy, arising from recent non-disjunction, as for members of a dysploid series arising from chromosome rearrangements during past evolutionary changes. In attempts to avoid such ambiguity, many other authors have given their own definitions, but as most of these differ from each other, the confusion in the minds of students reading the literature remains. Furthermore, while in many cases the authors make clear distinctions between the various ways in which the chromosome number can change (e.g. Favarger, 1967), their definitions are often inadequate when used generally. Thus purely numerical definitions (Darlington, 1937; Rieger & Michaelis, 1958; Dawson, 1962) breakdown when applied to a heterozygote for centromere dissociation such as *Nothoscordum fragrans* (Dyer, 1967), where the normal diploid complement ($2n = 19$) has an uneven number, and a trisomic seedling ($2n + 1 = 20$) has an even number which appears to be an exact multiple of one of the gametic complements ($n = 10$).

We consider that a redefinition is required which is consistent with the interpretation intended by Täckholm, while incorporating the wide range of cytological conditions now known. Difficult cases will of course still remain where an understanding of the relationship between chromosomes in a complement is incomplete; for example, where chromosomes are too small for accurate analysis, structural changes are too small to be recognised or additional chromosomes cannot be confidently designated as either A-chromosomes or B-type supernumeraries. Other terms, such as 'heteroploidy' (Winkler, 1916), defined by Sharp (1943) as a chromosome number other

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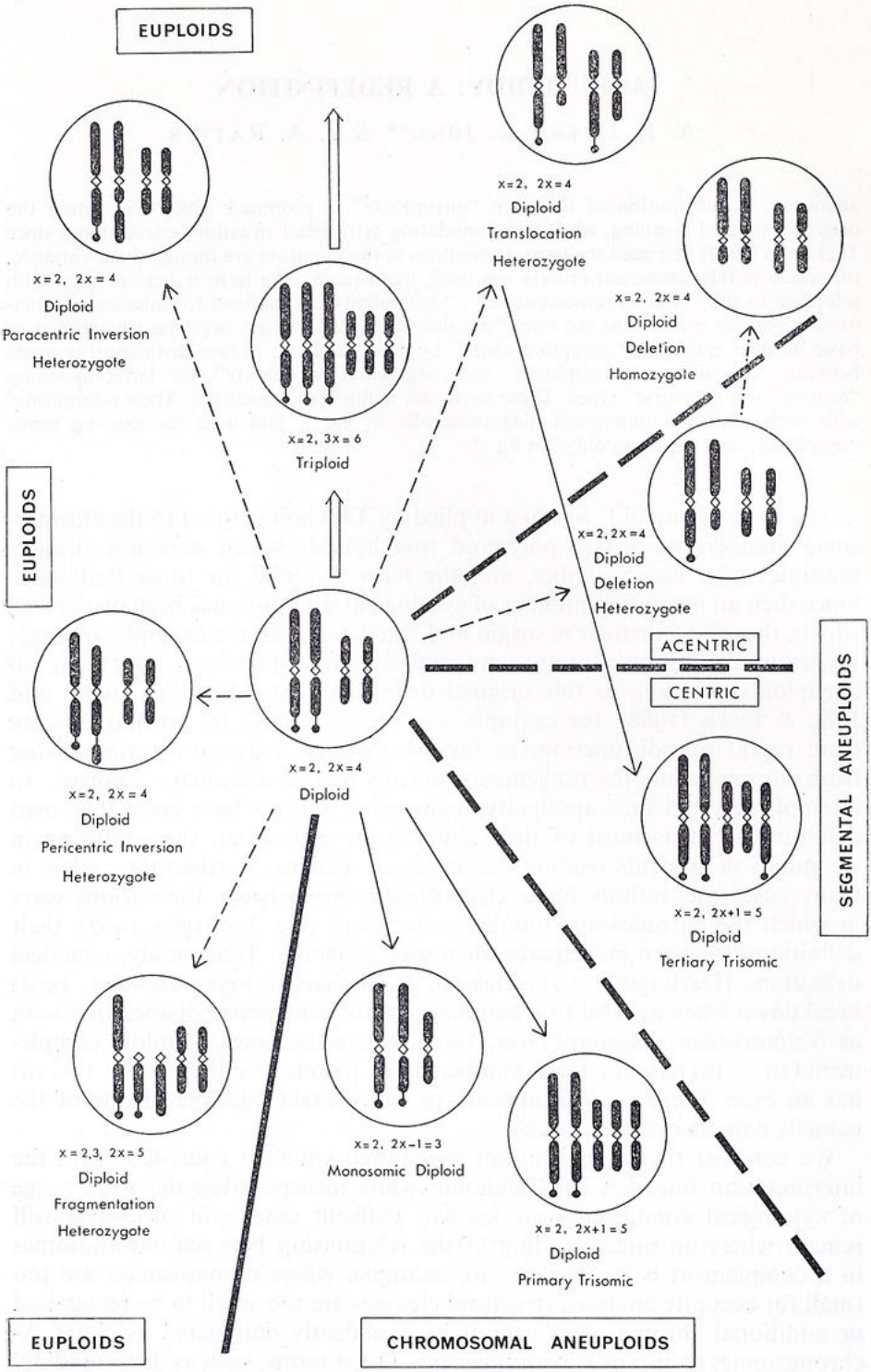

FIG. 1. The euploid and aneuploid derivatives of a structurally homozygous diploid.

spindle failure, to produce polyploids;

inbreeding, to produce homozygotes;

centromere failure, to produce non-disjunction;

chromosome breakage and reunion, to produce structural rearrangement.

than the true monoploid or diploid number, exist for use in a wider context.

In order to re-define 'aneuploidy' in this way, it has proved necessary to describe it in terms of the 'basic chromosome set', more or less equivalent to the basikaryotype of Sinoto & Sato (1940), which is itself defined below. In the discussion of the definitions which follow, examples, which could be almost limitless, have been kept to a minimum. Suitable examples are discussed under the appropriate headings in e.g. Darlington (1937), White (1954) and John & Lewis (1968).

Definitions

Basic chromosome set. A viable complement of non-homologous and indispensable chromosomes, each represented once.

Basic number. The number of chromosomes in a basic set.

Secondary basic chromosome set. The gametic complement of a fertile functionally diploid polyploid.

Secondary basic number. The number of chromosomes in a secondary basic set.

Euploid. Having one or more complete basic chromosome sets or their immediate derivatives by structural rearrangement.

Aneuploid: a) *Chromosomal aneuploid*, having one or more whole chromosomes of a euploid complement absent from or in addition to that complement; b) *Segmental aneuploid*, having one or more chromosome segments of a basic set absent from or in addition to one or more basic sets.

Discussion

Basic chromosome set

A basic set:—

i) is *viable*, in the monoploid or homozygous conditions, where the level of ploidy is normal for the life cycle. Even long established complements may fail in unreduced diploid gametes or unfertilised monoploid zygotes where these arise as spontaneous errors. A basic complement is so called because it has the potential for giving rise to further, derived forms.

In some cases ancestral basic complements no longer survive in the diploid state and are only viable in combinations with related genomes in a polyploid. Polyploids which are fertile and functionally diploid form a point from which a whole new series of evolutionary changes in the chromosome complement can develop. It is sometimes useful to consider the gametic complements of such polyploids as '*secondary basic sets*' whether or not the ancestral diploids have survived.

ii) has chromosomes which are *non-homologous*, homology by definition (Darlington & Mather, 1949) being determined by chromosome morphology and/or pairing behaviour.

iii) has chromosomes which are *indispensable*. This statement holds unless it can be demonstrated convincingly that nullisomic monoploids or diploids exist. The dwarf pomegranate *Punica granatum* var. *nanum* ($2n = 14$) derived from the normal plant with $2n = 16$ may be an example, but not enough information is available.

Supernumerary B-chromosomes, dispensable to the extent that individuals without them can compete with those that possess them, and whole populations and many species do not have them at all, are not therefore part of the basic set.

iv) differs from all others, as a result of structural changes in the chromosomes, to such an extent that a difference in chromosome morphology, behaviour or number is detectable under the microscope. The distinction between these gross changes and those, involving small chromosome segments or even single loci, which are only detectable genetically, is entirely artificial. It has, however, a practical significance while the visible chromosome remains the main basis for comparison in cytogenetical investigations

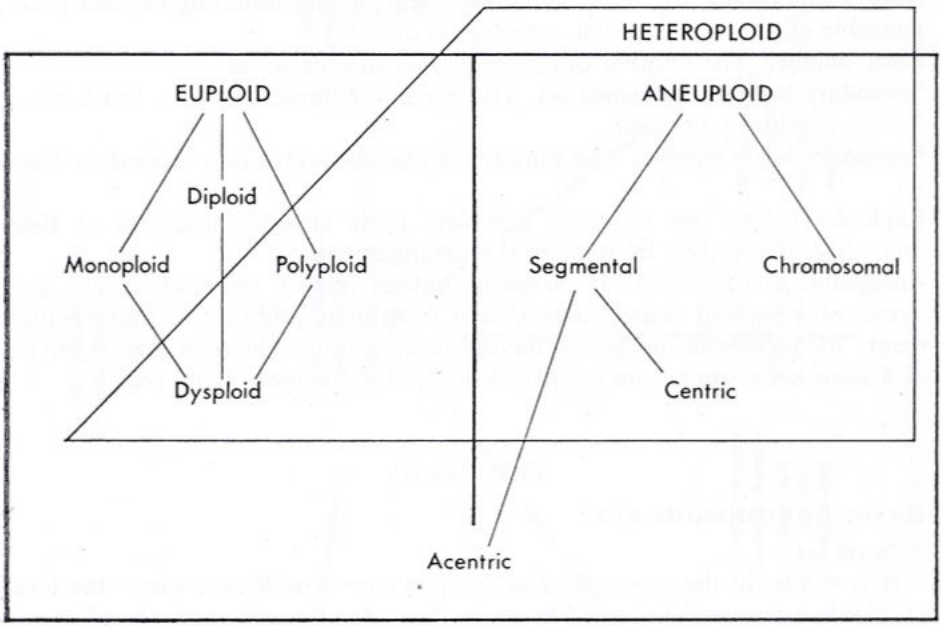

FIG. 2. The relationship of terms referring to ploidy.

into the mechanism and consequences of evolution. The terminology has to be applicable to the data, and only when the whole gene or DNA complement can be accurately and concisely described can the concept of the 'basic genome' usefully replace that of the 'basic chromosome set'.

It is generally accepted that more than one basic set may occur in a genus, species, population or even a polyploid individual, and that this is frequently but not always revealed by the chromosome number. It is also necessary to recognise that two basic sets may occur within a diploid individual (e.g. during establishment of new structural patterns in the chromosomes) and that these too may differ in chromosome number, as obviously must be the case during the establishment of a new basic number in a dysploid series. This is also invariably true of the hybrid products of wide crosses, but these are frequently sterile. Of more significance are examples like the heterogametic sex of dioecious species. Where the sex mechanism is of the XX/XY type, the heterogametic sex has two sets differing in structure and content of one chromosome. Sex chromosomes are not the only ones involved. In

hermaphrodite *Oenothera lamarkiana* there are two basic sets differing in the structure of every chromosome. In *Nothoscordum fragrans*, 2n = 19, structural rearrangements in one autosome have resulted in two basic sets (n = 9, 10) differing in number.

Examples of two basic sets within an individual will be widespread as any structurally altered complement capable of transmission to offspring is likely to be found in the heterozygous form prior to its eventual establishment or elimination. Indeed, every chromosome mutation will initially occur in a heterozygote, but only when the viability of the new complement has been demonstrated, can it be said that both complements constitute 'basic chromosome sets'. As stated by White (1961) ". . . the concept of diploidy does not require that the two haploid sets should be identical, either in a cytological sense or in genetic content, but merely that they should be similar."

Euploid

An organism is clearly euploid if it has one or more, not necessarily identical, basic chromosome sets, whether or not it is genetically homozygous.

Aneuploid

a) *Chromosomal aneuploids.* These include nullisomics (only convincingly demonstrated in allopolyploids) monosomics and polysomics, and result from irregular distribution of chromosomes at division. They do not include complements where the numerical variation only involves B-type supernumeraries. As for "euploid", the term "aneuploid" can be applied equally well at the level of cells, individuals, or populations.

b) *Segmental aneuploid.* There are two situations which resemble chromosomal aneuploidy to some extent, but are not exactly covered by its definition. The first is heterozygosity for deletions or duplications where one or more acentric segments of a basic set have been gained or lost. The second is where centric fragments, sometimes forming new chromosome arm combinations as in the secondary and tertiary polysomics of *Datura* (Blakeslee, 1934), have been gained. In neither situation does the affected genetic material constitute a whole, identifiable chromosome of the basic set, but in its quantitative effect on the gene complement or its qualitative effect on the phenotype, there is no clear distinction between these conditions and chromosomal aneuploidy. Indeed, a duplicated or deleted segment in one chromosome can be longer than another whole chromosome of the same complement. Thus, in Hyacinth, the deleted segment in the 'L' chromosome of the diploid 'Tubergen's Scarlet' is longer than the 'S' chromosome which is polysomic in some aneuploid varieties. To cover such situations, the new term "Segmental aneuploidy", as defined above, is proposed, with a further distinction between those which are *centric* (e.g. a secondary trisomic) and those which are *acentric* (and simultaneously classifiable as euploid, e.g. a plant heterozygous for a duplication of a segment) where it is necessary to emphasise the differences with respect to chromosome number and pairing behaviour.

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