

LECTURE 34

VARIATION IN CHROMOSOME NUMBER

A. Aneuploidy

1. Aneuploids are defined as chromosome complements that are *not* exact multiples of the basic haploid set. In theory, all sorts of aneuploids can be defined. Those that are viable to any extent include the following.
 - a) Additions to chromosome number:
 - (i) Primary trisomy [$2n + I$]: examples in humans include Klinefelters (XXY), and Down's syndrome individuals [$2n = 47D$].
 - (ii) Primary tetrasomy [$2n + II$]: examples are mostly in polyploid plants.
 - (iii) Double primary trisomy [$2n + I + I$].
 - b) Reductions in chromosome number:
 - (i) Primary monosomy [$2n - I$]: example in humans is Turners (XO).
 - (ii) Primary nullisomy [$2n - II$]: examples mostly in polyploid plants.
2. Origins of aneuploids:
 - a) Most aneuploids arise from primary nondisjunction at meiosis or from segregation at meiosis of an already-aneuploid parent (= secondary nondisjunction).
3. Viability of aneuploids:
 - a) Generally, viability of most aneuploids is low, at least in diploids. As might be expected, aneuploidy is better tolerated in polyploids or species with polyploid ancestry. Parameters such as phenotypic modification, fertility, and viability decrease for any type of aneuploidy as the ploidy level increases.
4. Genetics of aneuploids:
 - a) The Mendelian genetics of aneuploids is straightforward and can be evaluated in terms of the types of chromosomal configuration expected at meiosis I.
 - (i) Trisomy [$2n + 1$]: trivalent or extra univalent (n and $\underline{n+1}$ gametes)
 - (ii) Tetrasomy [$2n + 2$]: quadrivalent or other configurations
 - (iii) Monosomy [$2n - 1$]: univalent (n and $\underline{n-1}$ gametes)
 - (iv) Nullisomy [$2n - 2$]: all bivalents (one pair missing)

5. Usefulness of aneuploids: limited largely to “Aneuploid Mapping.”

- a) Aneuploid mapping is one of the approaches employed to localize genes to specific chromosomes. It is used primarily in plant species, particularly those with polyploid background or ancestry. The “logic” behind aneuploid mapping is that segregation of an aneuploid is non-Mendelian in the sense that there will be a deviation from “normal” Mendelian segregation.
- (i) The paradigm example is in the jimson weed (*Datura*, $n = 12$) where there exists a primary trisomic for each chromosome and where each primary trisomic confers a distinguishable and different external phenotype on the plant.
 - (ii) The experimental procedure is straightforward: a “quasi-heterozygote” for the gene in question is generated in a situation where one of the chromosomes in the complement is in an aneuploid condition. Segregation of alleles in the quasi-heterozygote is then followed by an appropriate cross (usually a testcross or self-fertilization). If the gene in question is on the aneuploid chromosome, segregation will differ from that expected under normal Mendelian segregation. If the gene is *not* on the aneuploid chromosome, segregation will not differ from that expected for normal Mendelian segregation.
- b) Note that aneuploid mapping is one of four approaches that can be used to localize genes to specific chromosomes in higher organisms.
- (i) These approaches are:
 - Aneuploid mapping
 - Deletion mapping (pseudominance)
 - In situ* hybridization
 - Somatic cell fusion/hybridization

B. Euploidy

1. Euploids are defined as chromosome complements that are exact multiples of the basic haploid set. While all sorts of euploids can be defined, there appears to be a limit to the number of “genomes” that can exist within cells/organisms. The highest polyploids found in a sustaining condition are a few 12-ploids in species of ferns. General types of euploids are as follows.
- a) Monoploids (1N):
- (i) Seldom observed in animals except for haplo-diploids among the Hymenoptera (bees, wasps, etc.).
 - (ii) Observed occasionally in plants but generally of poor viability.
 - (iii) Highly sterile due to segregation grossly aneuploid gametes, stemming from segregation of haploid set of chromosomes at bipolar meiosis,

b) Polyploids (three or more genomes):

- (i) Generally, sustaining polyploid populations or species are even numbered, as odd-numbered polyploids have difficulty segregating balanced sets of chromosomes to two poles at meiosis.
 - (a) Exceptions to this are the all-female, triploid (3N) unisexual “species” discussed previously.
- (ii) However, sustaining, non-triploid polyploids are exceptionally rare in animals, occurring largely in only a few frog species. Two possible reasons for the rarity of sustaining polyploids in animals have been hypothesized.
 - (a) Barriers due to sex determination mechanisms (e.g., *Drosophila*)
 - (b) Low probability (availability) of finding another bisexual like-polyploid mate
- (iii) Alternatively, polyploidy is quite common in plants [$\approx 50\%$ of all species, and $\approx 75\%$ of all grass species, including several economically important cereals].
 - (a) Sex chromosomes (disruption of sex-determination mechanisms) would not an issue in plants, and finding a like-polyploid mate would be “solved” by self-fertilization. This explains why most polyploid plants are capable of selfing.

2. Generative mechanisms of polyploidy:

- a) Somatic doubling: can occur in initial zygote, in stem cell leading to reproductive tissue, or premeiotically in a germ-line gonial cell.
- b) Irregular meiosis, where either meiosis I or II or both is irregular.
- c) Note that with either mechanism, almost any ploidy level imaginable can be generated.

3. Types of polyploids:

- a) Autopolyploids: the same genome is duplicated, e.g., $A/A \rightarrow A/A/A/A$.
 - (i) Many autopolyploids are sterile to varying degrees because of segregation “problems” arising from multivalent configurations of homologous chromosomes at meiosis I.
 - (ii) Some autopolyploids, however, are 100% fertile. In virtually all of these “fertile” autopolyploids, only bivalent configurations are observed at meiosis I. Where known, generation of bivalent meioses in autopolyploids appears to be under genic control.

- (iii) In fertile autopolyploids, formation of bivalents is *random*. Because bivalent pairing is random, there will be several different 2 x 2 segregation patterns possible, depending on the level of polyploidy. In autotetraploids, for example, there will be six different 2 x 2 segregation patterns. This can be demonstrated by using gene markers in a Mendelian cross.

Consider: two autotetraploid lines, homozygous AAAA and aaaa

P₁ AAAA x aaaa

F₁ Aaaa (self) gametes are: AA, Aa, Aa, Aa, Aa, aa

F₂ 35:1, A:a (in phenotype)

- b) Allopolyploids: arise when *different* genomes come together, usually by hybridization.

- (i) Consider an allotetraploid AAA'A', where AA represent genes on chromosomes from one genome, and A'A' represent genes on chromosomes from a second genome. A and A' represent homologous genes but the chromosomes bearing A and A' do not have sufficient homology to pair at meiosis. A and A (and A' and A') are on homologous chromosomes, whereas A and A' are on *homeologous* chromosomes.

- (ii) Allopolyploids originate through what is called the “amphidiploid” cycle, where two species (e.g., AA and A'A') cross (hybridize), but where their “homeologous” chromosomes (A and A') cannot pair at meiosis. The lack of *structural* homology between chromosomes bearing A and A' is generally due to different chromosomal rearrangements that have occurred in the two lineages (species). The result typically is production of aneuploid gametes and sterility in the “hybrids.” However, if there is a chromosome doubling that generates AAA'A', the homeologous chromosomes now have homologous pairing partners at meiosis.

- (iii) A classical example is common breadwheat, a hexaploid derived from three different (diploid) genomes, called A, D, and B.

The A genome is from *Triticum monococcum*.

The D genome is from *Triticum tauschii*.

The origin of the B genome is unknown.

- (iv) Because pairing of chromosomes at meiosis in allopolyploids is *non-random*, patterns of gene segregation differ from those in autopolyploids (where pairing by twos is random).

Consider: two allotetraploid lines, homozygous AAA'A' and aaa'a'

P₁ AAA'A' x aaa'a' gametes all AA' and aa'

F₁ AaA'a' (self) gametes are: AA', Aa', A'a, aa''

F₂ 15:1, A:a (in phenotype)

4. Polyploidy and agriculture:

- a) Many crop or floral plants are natural polyploids. Examples include
 - (i) common breadwheat (6x, arising from three diploid genomes)
 - (ii) New World cotton (4x, arising from two diploid genomes)
 - (iii) marigolds
- b) Triploidy is common in several economically important agronomic plants. Examples include bananas, seedless watermelons, and tulips. Usually, the agronomic variety is sterile or seedless, but can be propagated vegetatively (mitotically).
- c) Polyploidy can be induced and used as an interspecies genetic transfer mechanism, i.e., to transfer useful traits between species. A documented example is the transfer of resistance to the tobacco mosaic virus between *Nicotiana tabacum* (cultivated tobacco) and *Nicotiana glutinosa* (a related species).
 - (i) *Nicotiana tabacum* is TMV^S whereas *Nicotiana glutinosa* is TMV^R
 - (ii) Both species are diploids, and hybrids between them are sterile. The allotetraploid, however, is fertile and is TMV^R. Selection for a higher yield strain is fairly straightforward.