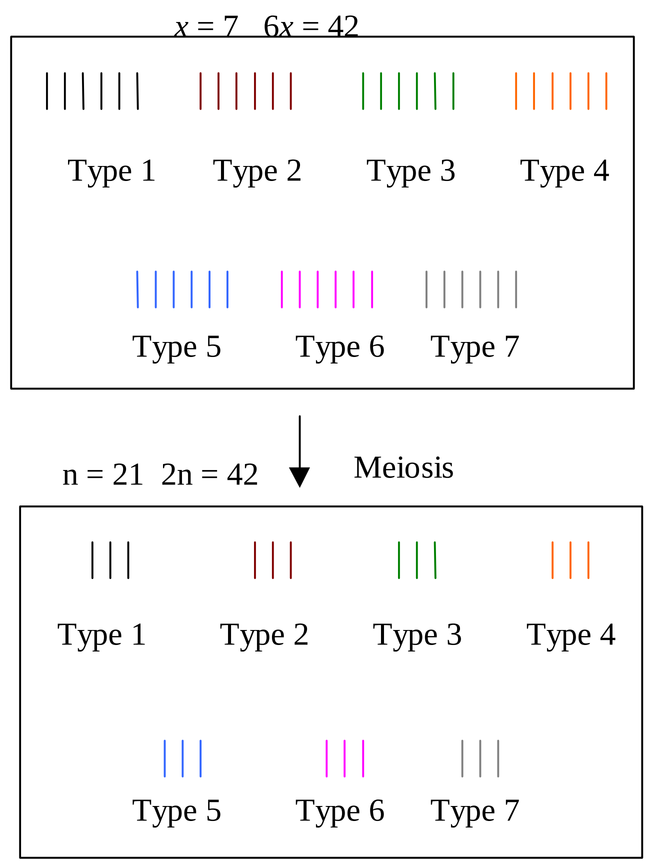
**CHAPTER 9 LECTURE NOTES:**
**CHROMOSOME MUTATION II: CHANGES IN NUMBERS**

I. Aberrant euploidy

A. General info

1. Euploidy refers to the situation in which an organism has one complete set of chromosomes or an integer multiple of a set.
2. The number of chromosomes in a basic set called monoploid number ($x$).
3. The "normal" situations are $1x$ = haploid or $2x$=diploid.
4. Polyploids ($>2x$) are by definition abnormal although they exist in plants in nature.
5. Haploid number (n) refers only to number of chromosomes in gametes. For most diploid organisms that we are familiar with the haploid and monoploid number is the same.

However, this is not always the case: wheat is a hexaploid containing six sets of similar but not identical chromosomes with 7 chromosomes per set ($x$=7 and $6x$ = 42). The gametes however have 21 chromosomes (n=21 and 2n=42).



A. Monoploidy (1*x* = 1 set of chromosomes instead of normal 2)

1. Monoploidy can arise from spontaneous development of an unfertilized egg (parthenogenesis).
2. Lethal in most animals because unmasking of lethal alleles.
3. Monoploids usually sterile because of a problem with meiosis: No pairing partner for each chromosome. Gametes containing all the chromosomes in the set are formed at a frequency of $(1/2)^{x-1}$
4. Exception in animals are male bees, wasps, and ants which develop parthenogenetically and in which gametes are formed by mitosis
5. Monoploids are important in plant breeding. For example:

    Diploids mask advantageous new mutations. Can use monoploidy to circumvent this: Induced in plants anthers by taking cells destined to become pollen and treating with cold → will become small mass of dividing cells – embryoid → Plate on agar and grows into plantlet → look for desirable traits → Use colchicine to redouble chromosomes returning to diploidy in sector of plant that can then be transplanted.

C. General info on polyploidy.

1. In plants

    a) Autoploidy (multiple of sets from one species) vs. alloploidy (multiple sets from different species)

    b) Even # ploidy vs. odd # ploidy: Odd # ploidy always have unpaired chromosome for each chromosome type during meiosis; thus, the probability of producing genetically balanced gametes is low (see below), and odd # ploidies are effectively sterile.

2. In animals

    a) Polyploidy is rare. Reasons for rareness of polyploidy in animals is not certain. Hypothesis is the sex-determination mechanism depends on delicate balance in chromosomes. Also, since most animals do not undergo self-fertilization, there is no chance for polyploids to successfully produce progeny that will be fertile (i.e. 2n X n = 3x (sterile).

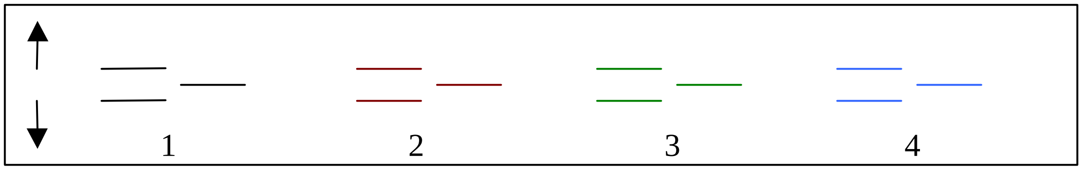
    b) In humans, polyploid zygotes usually die in utero (account for approx. 20% of spontaneous abortions.

    c) There are a few examples of polyploids in animals including flatworms, leeches, brine shrimp, some polyploid frogs and toads, and some fish.

3. Humans have giant polyploid nuclei in such tissues as liver and kidneys. Somatic cell polyploidy called endopolyploidy**.**

D. Triploids (3*x* = 3 sets of chromosomes instead of normal 2 sets)

1. Usually autopolyploids
2. Can arise from spontaneous aberrant meiosis in nature to give a 2n gamete that fuses with a normal n gamete to give a *3x* zygote OR from tetraploid (4*x*) and diploid (2*x*) cross.
3. Usually sterile because the frequency of getting exactly 1 set or exactly 2 sets of all the chromosomes to migrate to one pole is $(1/2)^{x-1}$



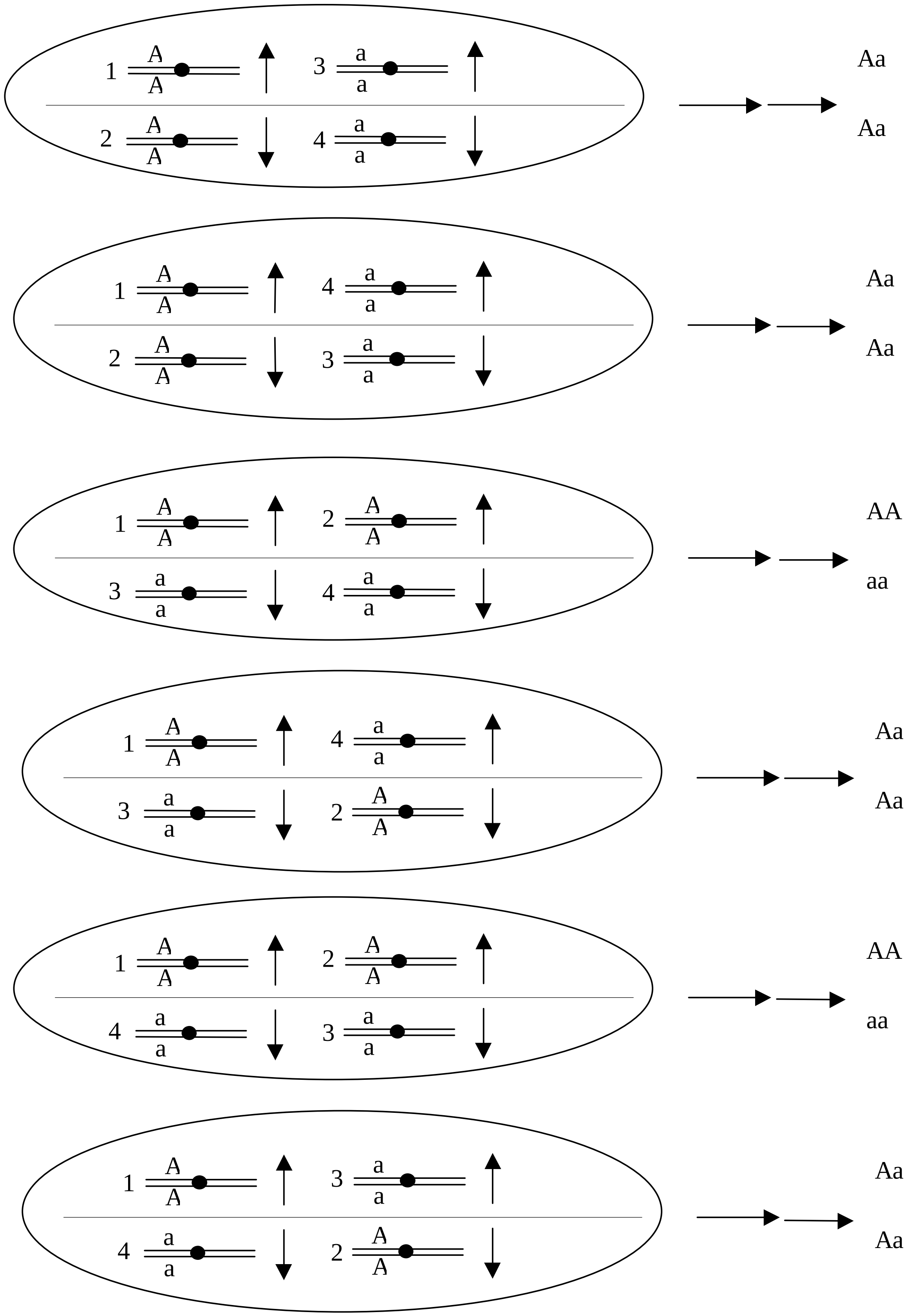

Gametes that contain intermediate between 1n and 2n exhibit genome imbalance because there are different ratios of genes depending upon if the gamete has 1 or 2 copies of each chromosome.

4. Seedless watermelons and bananas are triploid. They are sterile, but are propagated vegatatively.

E. Autotetraploids in plants (4*x* where all 4 sets all derived from one species)

1. Arise from spontaneous aberrant meiosis in nature to give a 2n gamete that fuses with another 2n gamete to give a 4*x* zygote OR by accidental doubling of 2*x* genome to a 4*x* genome in the 1 cell stage of development
2. Important in crop plants because greater # chromosomes = larger plant.
   (alfalfa, coffee, peanuts, MacIntosh apples)
3. Because there are an even # of chromosome sets, pairing of homologous chromosomes allows for normal meiosis.
4. Single locus segregation ratios in are different than simple Mendelian ratios
   a) With a AAaa plant where A/a is close to the centromere:
      Gamete ratios of 4 Aa : 1 AA : 1 aa

   Figure on the next page shows how this is derived:


1 A A
3 a a
2 A A
4 a a
Aa
Aa
1 A A
4 a a
2 A A
3 a a
Aa
Aa
1 A A
2 A A
3 a a
4 a a
AA
aa
1 A A
4 a a
3 a a
2 A A
Aa
Aa
1 A A
2 A A
4 a a
3 a a
AA
aa
1 A A
3 a a
4 a a
2 A A
Aa
Aa

b) With a BBbb plant where B/b is far from the centromere, there are many crossovers between the loci and the centromere. This effectively uncouples the segregation of the B/b alleles from the segregation of the centromere. As such you can consider the chances of obtaining the following gametes as :
BB (12/56) Bb (32/56) and bb (12/56)

F. Allotetraploids (4 sets from different species: 2 sets from species #1 and 2 sets from species #2)

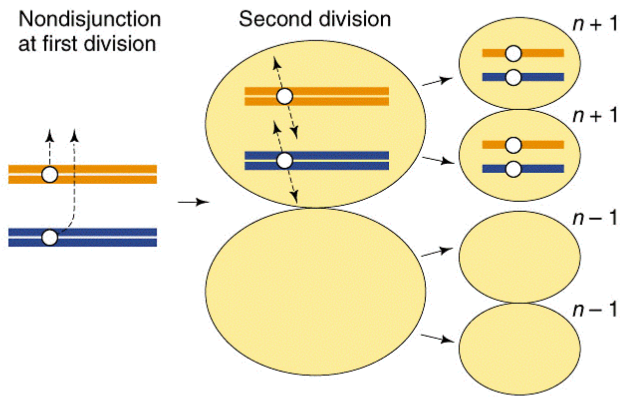
1. Chromosomes are hom**e**ologous (similar but not completely homologous)
2. Important in plant breeding:
   Consider 2 species (2n) which you attempt to cross: $n_1$ X $n_2$ → zygote with $n_1 + n_2$ → ususally sterile because homeologues can not pair → treat with colchicine → $2n_1 + 2n_2$ → now have a allotetraploid (4*x*) (AKA amphidiploid)
3. In nature, the same events as (2) happen except that chromosome doubling is a rare spontaneous event. Allotetraploidy was important in the evolution of plants (cotton, wheat).

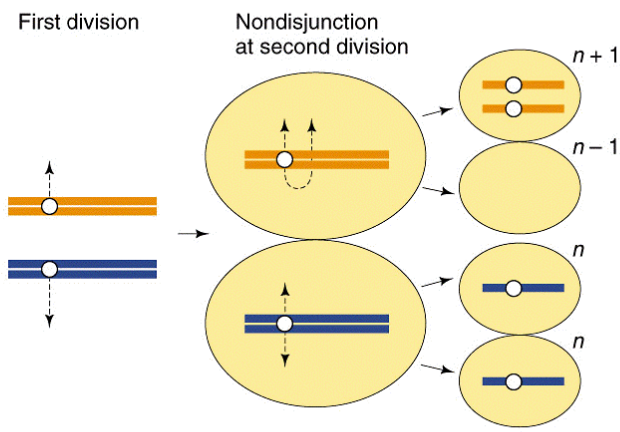
II. Aneuploidy

A. General information

1. The situation in which one or several chromosomes are lost from or added to the normal set of chromosomes

2. Caused by nondisjunction during meiosis to generate n+1 or n-1 gametes which when fertilized with n gamete to generate 2n+1 or 2n-1 zygotes




(from An Introduction to Genetic Analysis, 6th ed. by Griffiths et al. (W. H. Freeman and Company))

3. If autosome is affected, usually lethal in animals because of either chromosome imbalance or unmasking of lethal alleles; exception is the sex chromosomes (see below).

B. Nullsomics

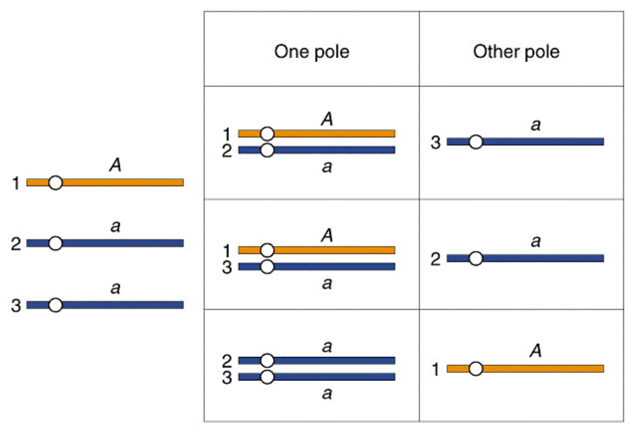
1. Missing two homologous chromosomes for a certain chromosome #
2. 2n-2
3. Lethal in human diploids because a whole chromosome type has been deleted
4. Can be ok in some polyploid plants

C. Monosomics

1. Missing 1 chromosome of a certain chromosome #
2. For diploids: 2n-1
3. In humans, lethal for autosomes (die in utero)
4. Turner syndrome (only 1 X chromosome) Figure 9-17
5. Can be ok in some plants – in fact can use monosomic banks to map recessive alleles to the chromosomes

D. Trisomy

1. Have 1 extra chromosome of a certain chromosome #
2. For diploids: 2n+1
3. sex chromosome trisomies
    a) Klinefelter's syndrome (XXY) Figure 9-20
    b) XYY syndrome (XYY)
    c) Triplo-X (XXX)
4. autosomal trisomies
    a) Trisomy 21 (Down's syndrome) Figure 9-21
    b) Trisomy 13 (Patau syndrome)
    c) Trisomy 18 (Edwards syndrome)
5. Can be ok in some plants
3. Single locus segregation ratios in are different than simple Mendelian ratios
    a) With an Aaa plant where A/a is close to the centromere:
        Gamete ratios of 1A: 2Aa: 2a: 1 aa

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E. Disomics

Aberration in haploid organisms where there is an extra chromosome

F. Somatic Aneuploidy

In above mentioned cases, the chromosome mutations all arose in the germ cells during meiosis. However, aneuploidy can arise in the somatic tissues during mitosis resulting in a genetic mosaic of cell types.