

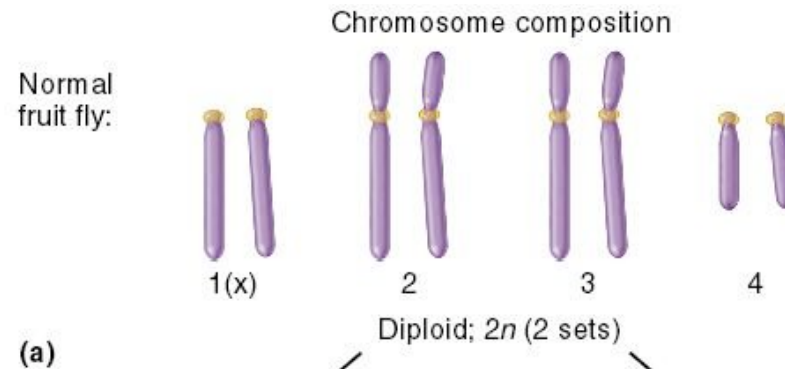
Variations in Chromosome Structure & Function

Ch. 8

Variation in Chromosome Number

- Chromosome numbers can vary in two main ways
 - Euploidy
 - Variation in the number of complete sets of chromosome
 - Variations occur occasionally in animals and frequently in plants
 - Aneuploidy
 - Variation in the number of particular chromosomes within a set
 - Variations are always regarded as abnormal conditions

Polyloid organisms have three or more sets of chromosomes



Individual is said to be trisomic

Polyloid fruit flies:



Triploid; $3n$ (3 sets)

Tetraploid; $4n$ (4 sets)

(b) Variations in euploidy

Aneuploid fruit flies:



Trisomy 2 ($2n + 1$)

Monosomy 1 ($2n - 1$)

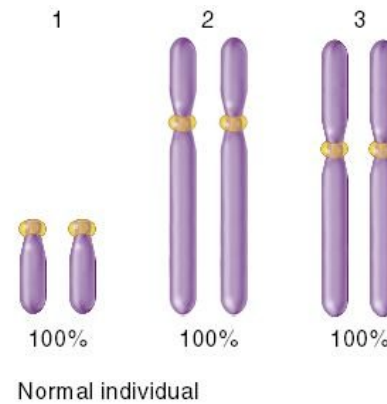
(c) Variations in aneuploidy

Individual is said to be monosomic

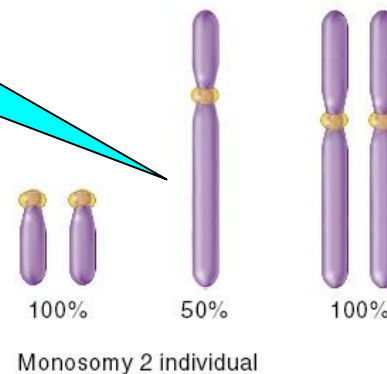
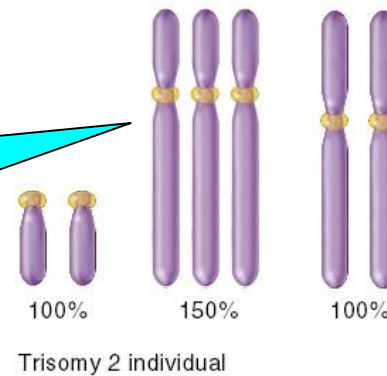
Aneuploidy

- The phenotype of every eukaryotic species is influenced by thousands of different genes
 - The expression of these genes has to be intricately coordinated to produce a phenotypically normal individual
- Aneuploidy commonly causes an abnormal phenotype
 - It leads to an imbalance in the amount of gene products

Aneuploidy



In most cases, these effects are detrimental
They produce individuals that are less likely to survive than a euploid individual



Aneuploidy

- Alterations in chromosome number occur frequently during gamete formation
 - ~ 5-10% of embryos have an abnormal chromosome number
 - ~ 50% of spontaneous abortions are due to such abnormalities
- In some cases, an abnormality in chromosome number produces an offspring that can survive

TABLE 8.1**Aneuploid Conditions in Humans**

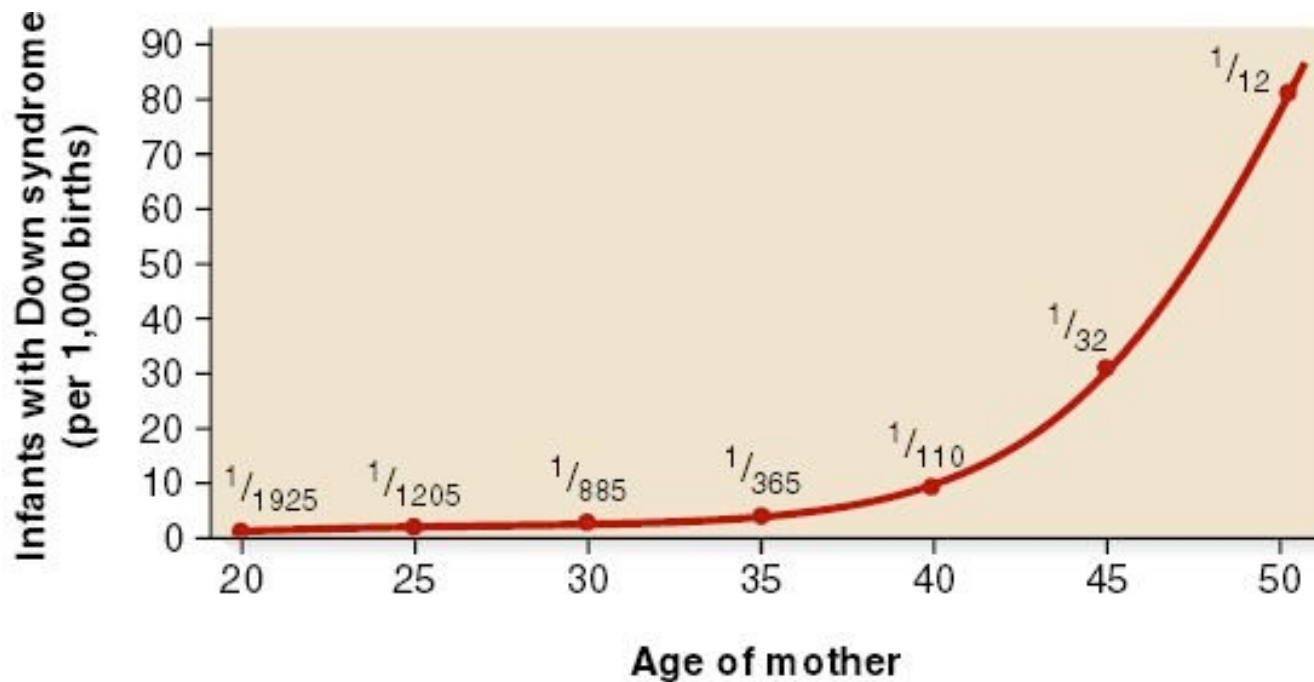
Condition	Frequency	Syndrome	Characteristics
<i>Autosomal</i>			
Trisomy 21	1/800	Down	Mental retardation, abnormal pattern of palm creases, slanted eyes, flattened face, short stature
Trisomy 18	1/6,000	Edward	Mental and physical retardation, facial abnormalities, extreme muscle tone, early death
Trisomy 13	1/15,000	Patau	Mental and physical retardation, wide variety of defects in organs, large triangular nose, early death
<i>Sex Chromosomal</i>			
XXY	1/1,000 (males)	Klinefelter	Sexual immaturity (no sperm), breast swelling
XYY	1/1,000 (males)	Jacobs	Tall
XXX	1/1,500 (females)	Triple X	Tall and thin, menstrual irregularity
X0	1/5,000 (females)	Turner	Short stature, webbed neck, sexually undeveloped

Aneuploidy

- The autosomal aneuploidies compatible with survival are trisomies 13, 18 and 21
 - These involve chromosomes that are relatively small
- Aneuploidies involving sex chromosomes generally have less severe effects than those of autosomes
 - This is explained by X inactivation
 - All additional X chromosomes are converted into Barr bodies
 - The phenotypic effects listed in Table 8.1 may be due to
 - 1. The expression of X-linked genes prior to embryonic X-inactivation
 - 2. An imbalance in the expression of pseudoautosomal genes

Aneuploidy

- Some human aneuploidies are influenced by the age of the parents
 - Older parents more likely to produce abnormal offspring
 - Example: Down syndrome (Trisomy 21)
 - Incidence rises with the age of either parent, especially mothers



Aneuploidy

- Down syndrome is caused by the failure of chromosome 21 to segregate properly
 - This **nondisjunction** most commonly occurs during meiosis I in the oocyte

- The correlation between maternal age and Down syndrome could be due to the age of oocytes
 - Human primary oocytes are produced in the ovary of the female fetus prior to birth
 - They are however arrested in prophase I until the time of ovulation
 - As a woman ages, her primary oocytes have been arrested in prophase I for a progressively longer period of time
 - This added length of time may contribute to an increased frequency of nondisjunction

Euploidy

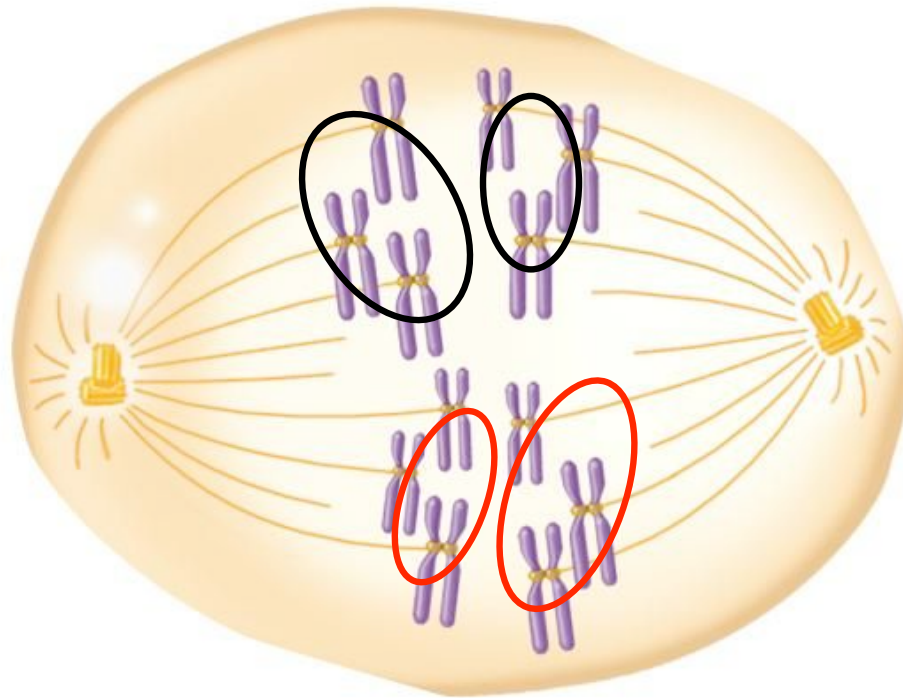
- Most species of animals are diploid ($2n$)
- In many cases, changes in euploidy are not tolerated
 - Polyploidy in animals is generally a lethal condition
- Some euploidy variations are naturally occurring
 - Female bees are diploid
 - Male bees (drones) are **monoploid**
 - Contain a single set of chromosomes

Euploidy

- In contrast to animals, plants commonly exhibit polyploidy
 - 30-35% of ferns and flowering plants are polyploid
 - Many of the fruits and grain we eat come from polyploid plants
- In many instances, polyploid strains of plants display outstanding agricultural characteristics
 - They are often larger in size and more robust

Euploidy

- Polyploids having an odd number of chromosome sets are usually sterile
 - These plants produce highly aneuploid gametes
 - Example: In a triploid organism there is an unequal separation of homologous chromosomes (three each) during anaphase I



**Each cell receives
one copy of some
chromosomes
and two copies of
other chromosomes**

Euploidy

- Sterility is generally a detrimental trait
- It is agriculturally desirable because it may result in
 - 1. Seedless fruit
 - Seedless watermelons and bananas
 - Triploid varieties
 - Asexually propagated by human via cuttings
 - 2. Seedless flowers
 - Marigold flowering plants
 - Triploid varieties
 - Developed by Burpee (Seed producers)

Natural And Experimental Ways To Produce Variations In Chromosome Number

- There are three natural mechanisms by which the chromosome number of a species can vary
 - 1. Meiotic nondisjunction
 - 2. Mitotic abnormalities
 - 3. Interspecies crosses

Meiotic Nondisjunction

- **Nondisjunction** refers to the failure of chromosomes to segregate properly during anaphase
- Meiotic nondisjunction can produce haploid cells that have too many or too few chromosomes
 - If such a gamete participates in fertilization
 - The resulting individual will have an abnormal chromosomal composition in all of its cells

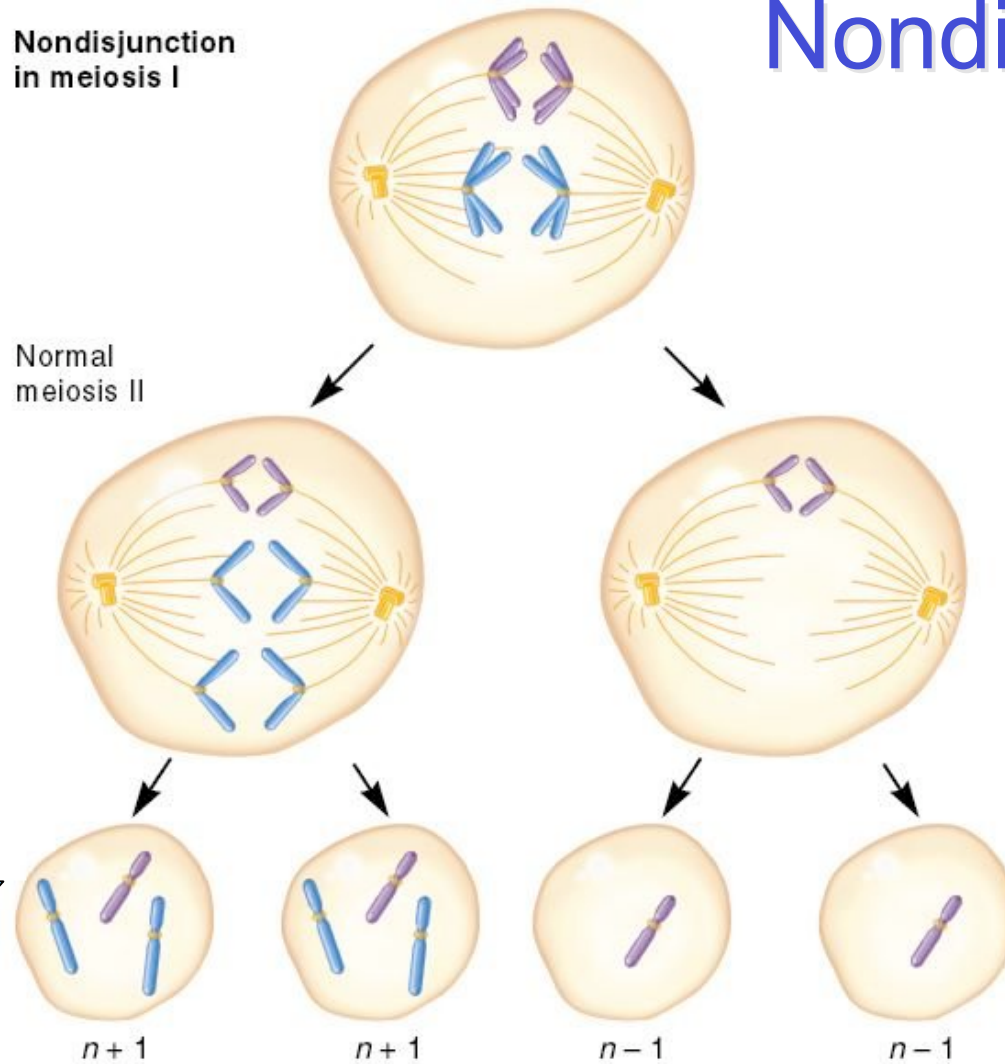
Meiotic

Nondisjunction

Nondisjunction
in meiosis I

Normal
meiosis II

During
fertilization,
these gametes
produce an
individual that
is **trisomic**
for the
missing
chromosome



During
fertilization,
these gametes
produce an
individual that
is **monosomic**
for the
missing
chromosome

(a) Nondisjunction in meiosis I

All four gametes are abnormal

Meiotic

Normal
meiosis I

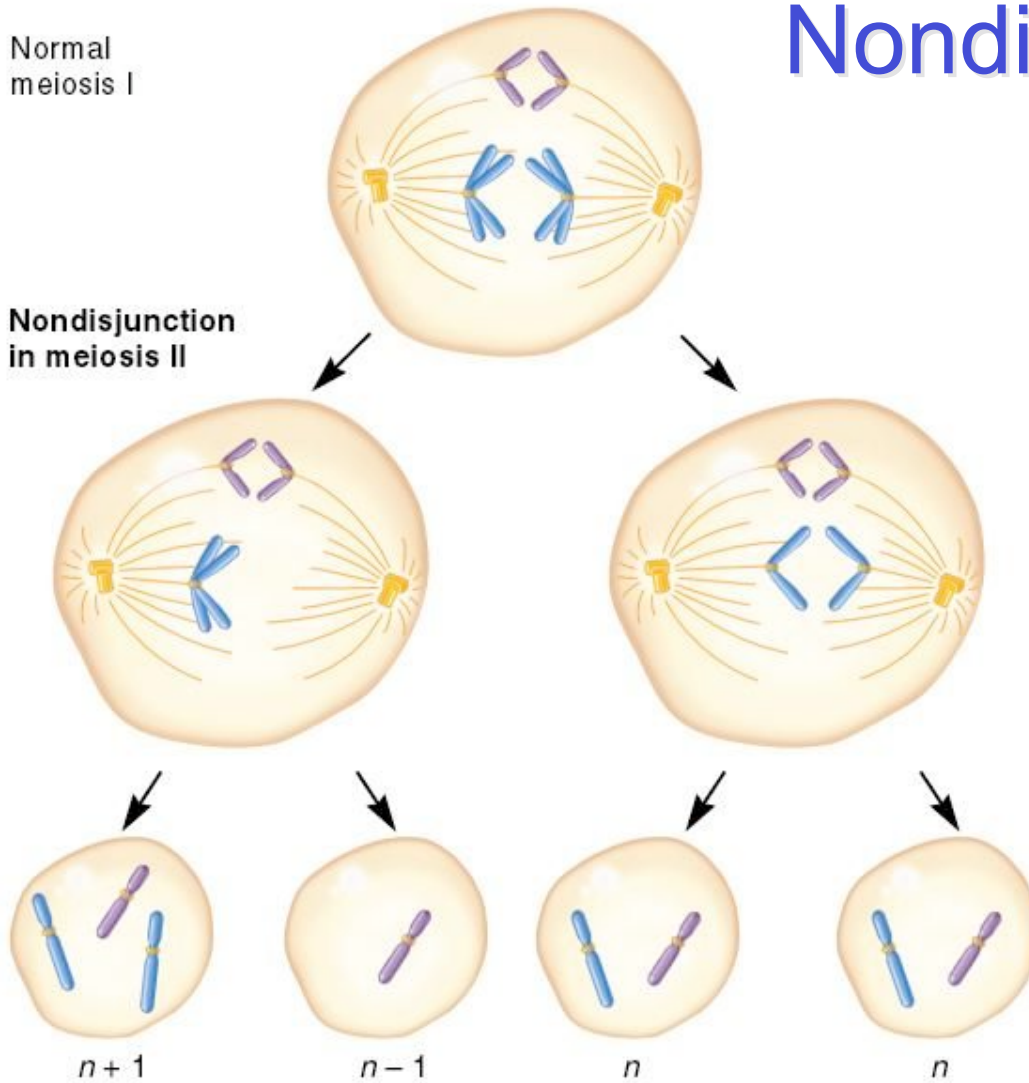
Nondisjunction

Nondisjunction
in meiosis II

50 % Abnormal
gametes

50 % Normal
gametes

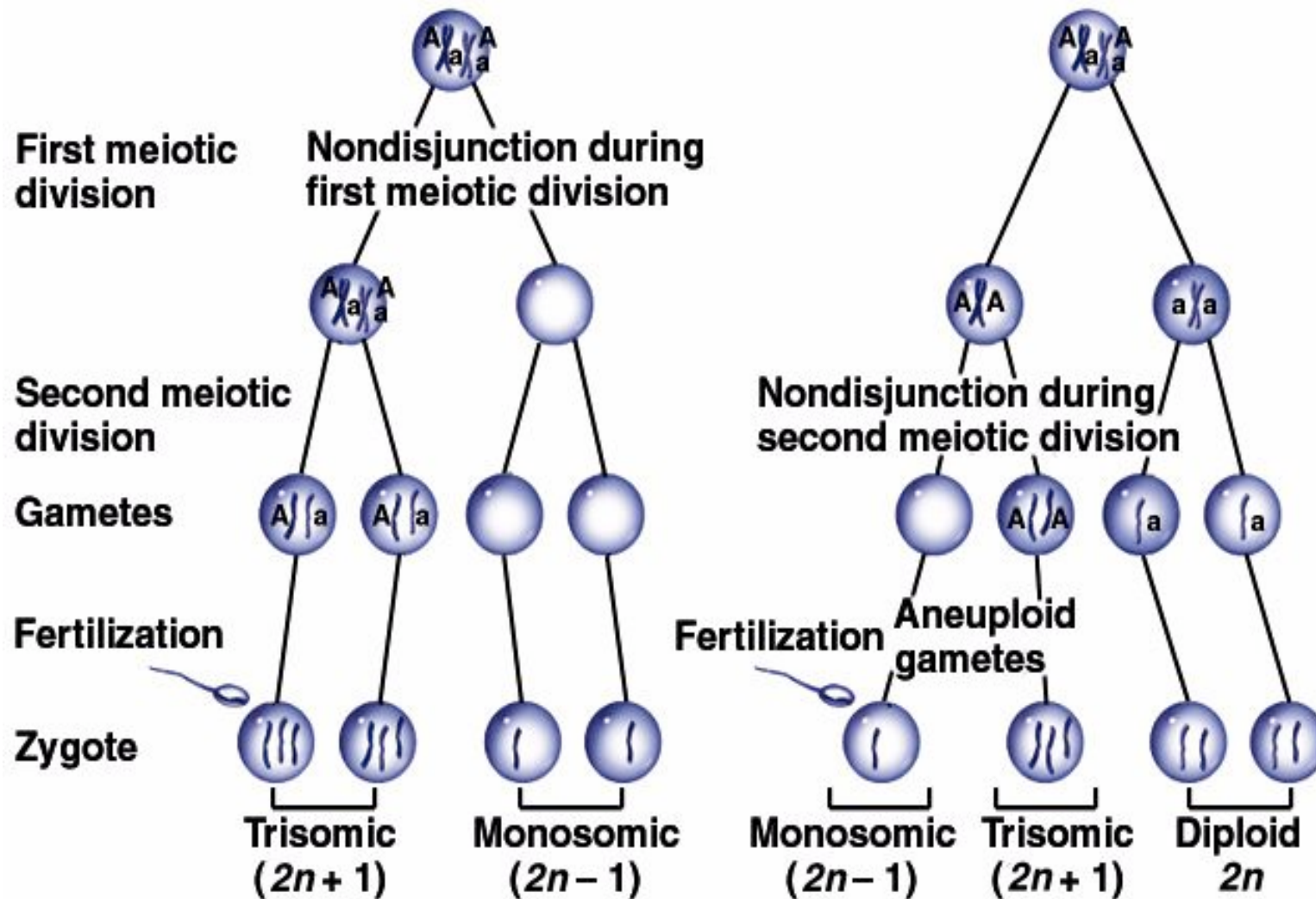
(b) Nondisjunction in meiosis II



Meiotic Nondisjunction

- In rare cases, all the chromosomes can undergo nondisjunction and migrate to one daughter cell
- This is termed **complete nondisjunction**
 - It results in a diploid cell and one without chromosomes
 - The chromosome-less cell is nonviable
 - The diploid cell can participate in fertilization with a normal gamete
 - This yields a triploid individual

Aneuploidy



Mitotic Abnormalities

- Abnormalities in chromosome number often occur after fertilization
 - In this case, the abnormality occurs in mitosis not meiosis
 - 1. Mitotic disjunction
 - Sister chromatids separate improperly
 - This leads to trisomic and monosomic daughter cells
 - 2. Chromosome loss
 - One of the sister chromatids does not migrate to a pole
 - This leads to normal and monosomic daughter cells

Changes in Chromosome Number

Euploidy: Cells that contain only complete sets of chromosomes

Diploidy ($2x$): Two copies of each homolog

Monoploidy (x): One copy of each homolog

Polyploidy: More than the normal diploid number of chromosome sets

Triploidy ($3x$): Three copies of each homolog

Tetraploidy ($4x$): Four copies of each homolog

Aneuploidy: Loss or gain of one or more chromosomes producing a chromosome number that is not an exact multiple of the haploid number

Monosomy ($2n - 1$)

Trisomy ($2n + 1$)

Tetrasomy ($2n + 2$)

Changes in Chromosome Number or Ploidy



Homework Problems

- Chapter 8
- # 1, 20, 21, 22, 23
- DON'T forget to take the online QUIZ!!
- DON'T forget to submit the online iActivity
 - “Karyotypes”