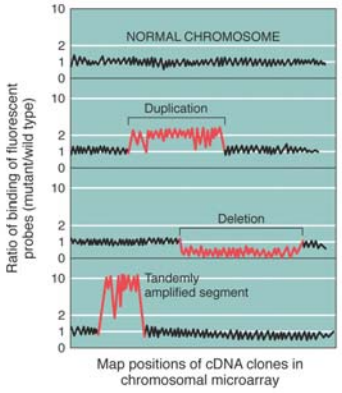
Reminder

- Critical comment essay due next Tuesday, Dec 7 in class
- See web site for general info about structure and guidelines
- Word limit: 500 (about 2 typed pages), use the counter on your word processing software, usually found in menu under "tools"
- Don't give me a book report, I read the papers and I already know what they say

Overview

- Multiples of complete sets of chromosomes are called polyploidy.
  - Even numbers are usually fertile.
  - Odd numbers are usually sterile.
- Aneuploidy refers to the gain or loss of single chromosomes, usually in meiosis.
- Chromosome aberrations include translocations, inversion, deletion, duplication.
  - Each has characteristic meiotic pairing.
  - Crossing-over may result in abnormal gametes, reduced fertility and unmasking of deleterious recessive alleles.
  - Duplication can also provide material for evolutionary divergence.



Chromosome mutations

- Two major types
  - change in number of copies of chromosomes
  - alteration of chromosome structure
- Reveal features of meiosis
- Provide insight into gene function
- Useful tools for experimental analysis
- Provide insight into evolution

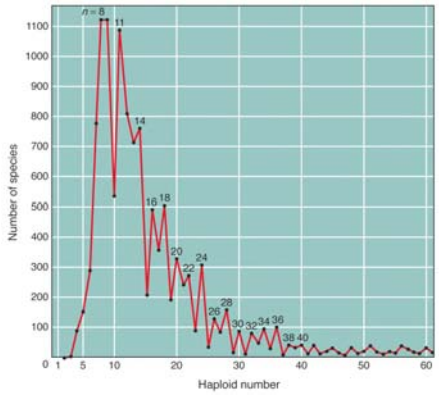
TABLE 11-1 Chromosome Constitutions in a Normally Diploid Organism with Three Chromosomes (Labeled A, B, and C) in the Basic Set

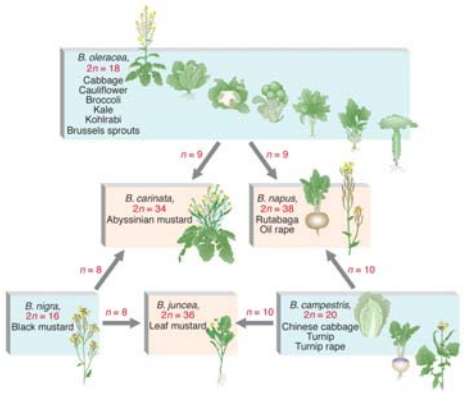
Name	Designation	Constitution	Number of Chromosomes
Euploids			
Monoploid	n	A B C	3
Diploid	$2n$	AA BB CC	6
Triploid	$3n$	AAA BBB CCC	9
Tetraploid	$4n$	AAAA BBBB CCCC	12
Aneuploids			
Monosomic	$2n - 1$	A BB CC	5
		AA B CC	5
		AA BB C	5
Trisomic	$2n + 1$	AAA BB CC	7
		AA BBB CC	7
		AA BB CCC	7

Aberrant euploidy

- Changes in *whole* chromosome sets
- Euploidy: multiples of basic chromosome set
  - haploid
  - diploid
- Aberrant euploid: more or less than normal number
  - monoploid (1*n*)
  - triploid (3*n*)
  - tetraploid (4*n*)
  - pentaploid (5*n*)
  - hexaploid (6*n*)

} polyploidy

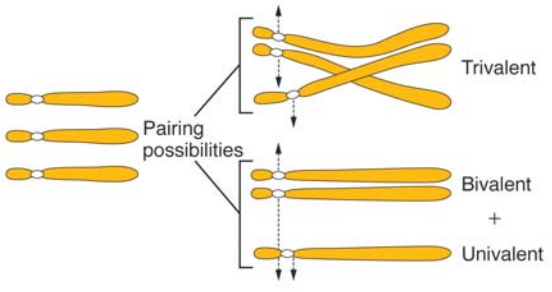




Aneuploidy

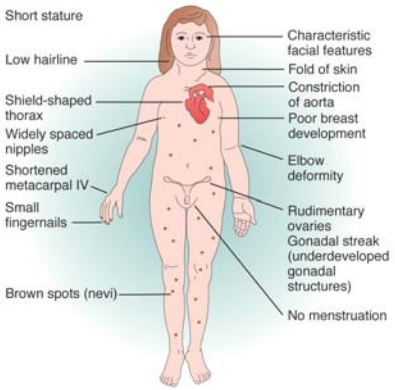
- Chromosome complement differs from normal by part of chromosome set
  - e.g., $2n \pm 1$
  - tolerated in plants
  - usually lethal in animals
- For autosomes:
  - monosomy: $2n - 1$
  - trisomy: $2n + 1$
  - nullisomy: $2n - 2$
  - disomy: $n + 1$ (in haploids)

For sex chromosomes, notation lists copies of each chromosome. Examples: XXY, XXX, XO



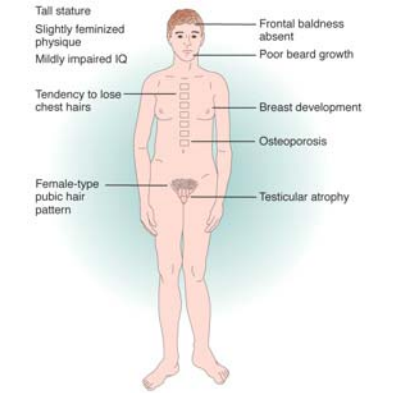
Monosomy

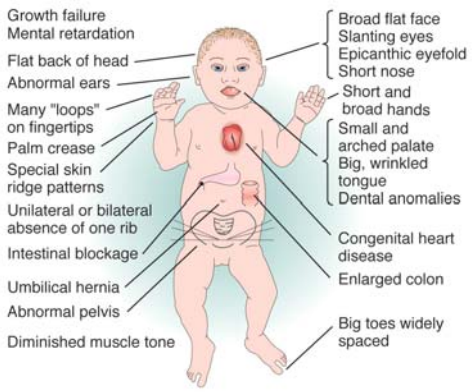
- $2n - 1$
- Usually deleterious owing to unmasking of recessive lethals in animals
  - lethal in utero in humans
- XO: Turner syndrome in humans
  - survives but has some developmental abnormalities
- Used to map genes in plants

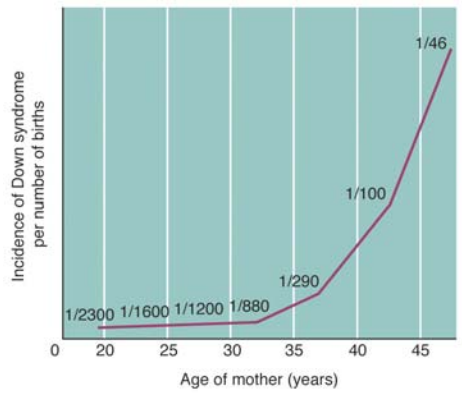


Trisomy

- $2n + 1$
- Often lethal in animals owing to chromosome imbalance
- If viable, may be fertile (meiotic trivalent)
- XXY: Klinefelter syndrome
  - male
  - sterile
- XYY: fertile, no extra Y in gametes
- XXX: fertile, no extra X in gametes
- Trisomy 21: Down syndrome

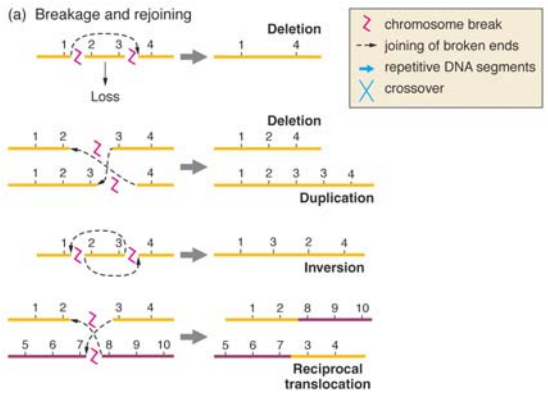


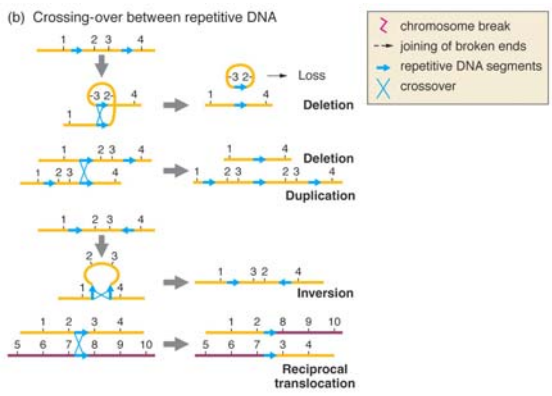




Changes in chromosome structure

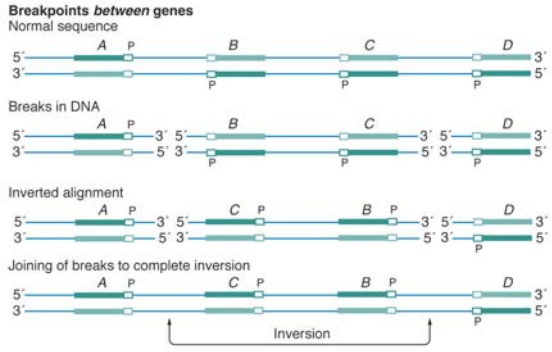
- Also called chromosome rearrangements
  - deletion: loss of segment
  - duplication: gain of segment
  - inversion: reversal of region
  - translocation: movement of segment to another chromosome
- Origin in double-stranded breaks where product has centromere and two telomeres
  - acentric fragments lost at anaphase
  - dicentric fragments dragged to both poles, lost

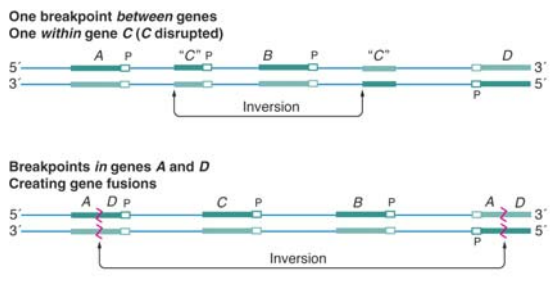


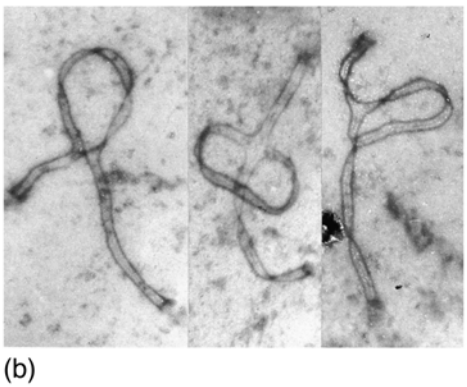


Balanced rearrangement: inversion

- Change in gene order, but gain or loss of DNA
- Inversion loop formed at meiosis I
- Paracentric: centromere outside inversion
  - crossing-over in inversion heterozygote results in one dicentric chromatid and one acentric fragment
  - reduced number of viable gametes
- Pericentric: inversion spans centromere
  - crossing over in inversion results in gene imbalance
  - reduced number of viable gametes

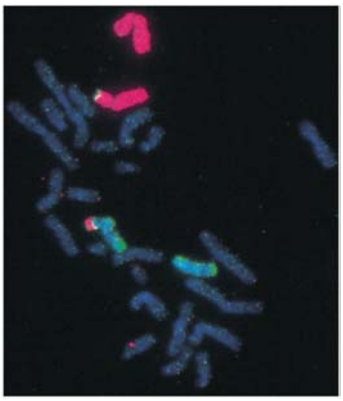


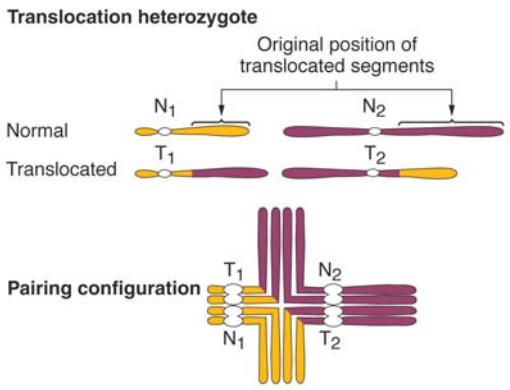




Balanced rearrangement: translocation

- Change in gene order, but no gain or loss of DNA
- Reciprocal translocations: exchange between two nonhomologous chromosomes
- Cross-shaped configuration at meiosis I
- Crossing-over results in gene imbalance, semisterility

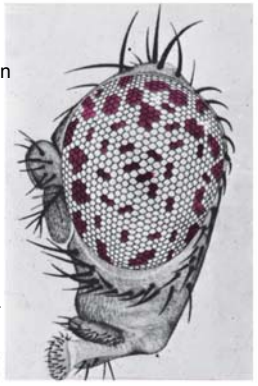




Drosophila eye showing how the White gene varies in expression in w+/w flies.

w+ is near tip of X, but can be translocated to chr.4 depending on where heterochroma-tin boundary is, w+ or w is expressed.

(b)

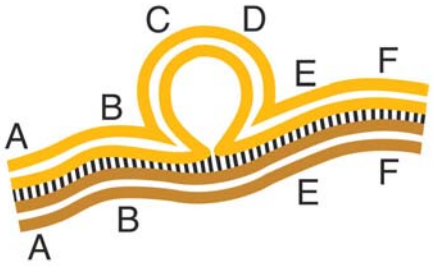


Position-Effect variegation

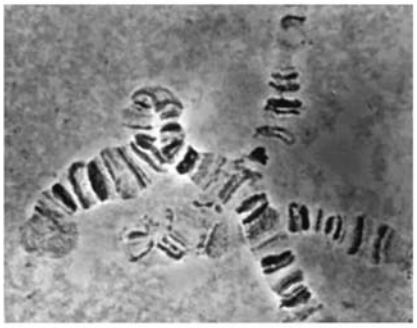
Imbalanced rearrangement: deletion

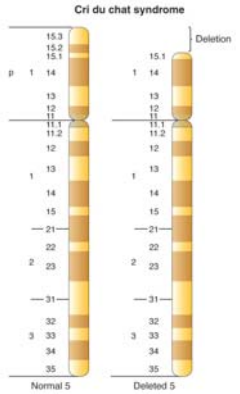
- Loss of segment of DNA
- Intragenic deletion: small deletion within gene
- Multigene deletion
  - many genes deleted
  - often severe consequences
    - gene imbalance
    - expression of deleterious recessive mutation (pseudodominance)
- Visible as deletion loop
- May be used in deletion mapping

(a) Meiotic chromosomes



(b) Polytene chromosomes





Deletions: plants vs animals

- Chromosomal deletion mutations often affect survival differently in plants, compared to animals (i.e. animal sperm)
- Plant sensitivity may be linked to metabolic demands on pollen in reproduction
- Pollen cells must germinate and form proteins to make pollen tube in order to fertilize the ovule
- Genetic abnormalities in pollen cells therefore have greater functional consequences than those in sperm

Imbalanced rearrangement: duplication

- Gain of segment of DNA
- Source of new genes and gene families
- Tandem duplication: adjacent duplications
- Insertional duplication: duplicate gene inserted elsewhere in genome
- May be consequence of unequal crossing-over