Lecture 6 (FW)
February 9, 2009
Diploid Genetics and Meiosis
Reading: Ch 3, pp. 37-46
Meiosis, pp. 31-36

Midterm Review. There will be a review session, for those who wish to attend on Tuesday, Feb. 24, 5:30-7:00 PM in 159 Mulford Hall..

Lecture 6: Chromosomes and Inheritance (Mendelian Genetics).
Primary Goal: Learn how traits are inherited in sexually reproducing animals.

I. Variation and Blending

A. Persons interested in natural history and agriculture had pondered how various traits are passed from generation to generation for many centuries. Not until the technology of the microscope appeared, and the concept of the cell became accepted were serious theories proposed.

B. Many proposals for the mechanism of heredity involved mixing of fluids. There were debates about how much was contributed by the father, how much by the mother. And there were many examples of blending of traits. For example, if white and red snapdragons were allowed to fertilize one another, the resultant plants were pink.

C. But there were discordant findings. Sometimes a single plant would appear in a field with a completely different trait, something that today we would call a mutant.
Even with the snapdragon, some enterprising scientists showed that if the pink snaps were crossed with one another, sometimes red, or even white plants would appear in the offspring. This is called a reappearance of parental characteristics.

D. How does the simple double helix of DNA fit into all this? We have waited to discuss humans and other familiar animals because it is not so simple as the linear transmission of traits that occurs in viruses and bacteria. There are two parents and reproduction is sexual.

It is crucial to remember that each single chromosome contains one very long strand of double-stranded DNA. Only protein is added. The Central Dogma remains the same.

II. Gregor Mendel

A. In 1866, Mendel published the results of experiments crossing different kinds of peas. He founded modern genetics, though the importance of his experiments was not realized until early in the 20^{th} century. (He died in 1884).

B. Mendel observed the phenomenon of reappearance of parental characteristics, and furthermore, he carefully quantified the results of his experiments.

1. Some nomenclature. P = parental generation; F-1 = first generation of offspring, etc.

2. In the characters Mendel studied (e.g., tall vs. short plants, smooth and wrinkled peas, etc.) there was no evidence of blending, even in the F-1. One character (e.g. smooth) always totally dominated the F-1. If you cross tall and short peas all the F-1 are tall. But when he carried out an F-2, short plants re-appeared, and always in approximately 1/4 of the cases. Mendel proposed a model to explain how this could

work. He postulated that there are **two functional units** (which are particulate; they do not blend) of heredity in each individual, and that the sex cells carry only one of each. So when fertilization occurs and the new plant arises, it receives one kind of genetic unit from each parent. Which of the two units present in each parent that is actually received by the F-1 is completely a matter of chance.

Furthermore, some units are more "powerful" than others, ie., they are **dominant** over the **recessive** form. Sometimes, however, there is no clear dominance and an apparent "blending" may result. The genetic units have come to be called genes, and since there are more than one "kind" of each **gene**, i.e., different flavors, the different kinds of each gene (e.g., short vs. tall) are called **alleles**.

C. We now know Mendel's model is correct. There were two alleles for each trait he studied. The genetic constitution determining a character is called the **genotype**. The actual trait that is observed is called the **phenotype**.

D. There are at least two genes for each trait, and the sex cells only receive, by chance, one of them. This is called **segregation**.

E. If you study more than one trait at a time, as Mendel did, you can observe that the segregation of genes for the different characters is completely independent from one another. This is called **independent assortment**. In other cases, however, the genes for different characters behave as if they are "**linked**". (This is because the genes in questions reside on the same chromosome. We shall come back to this phenomenon later.)

We should also mention now that many traits are determined by more than one gene. Mendel was lucky that he picked phenotypic traits that are determined by a single pair of allelic genes. Many characters are **polygenic**.

III. Meiosis

A. Mendel barely knew about cells, and chromosomes were not known at all. Curiously, however, his theory required that the "factors" be particulate, that there were two in each cell, and in sexually reproducing organisms the sex cells (gametes) would possess only one of each "factor", or gene.

B. This was very prescient. Chromosomes were found to be the carriers of genes in the early 1900's. And biologists realized that the formation of gametes was accomplished with a special variation of cell division, called **meiosis**.

C. The key to understanding meiosis is to realize that: 1) genes (double stranded DNA) are carried on chromosomes, 2) there are two of each kind of chromosome in the nucleus of every cell of the body. For this reason we are called "diploid". Organisms that only have one kind of each chromosome (called haploid), like bacteria, do not normally reproduce sexually (when they do, it is a very unusual kind of "parasexual" process that we shall not discuss). These haploid organisms usually reproduce asexually by mitosis.

D. The only thing difficult about meiosis is that there is a large, special vocabulary used to describe it, which we shall eschew.

1. Meiosis is accomplished by one doubling of the DNA (and hence of the chromosomes), a <u>pairing</u> of each of the two-some that constitute one chromosome type, followed by two divisions.

2. Hence, in the interphase preceding the first meiotic division, the DNA doubles. Each chromosome is now doubled.

Also, as the cell leaves G-2 and enters the division phase, each of the two similar (they are called homologous chromosomes) chromosomes (remember that they are doubled) physically matches up and pairs with its other member. Hence, there are packets of 4 daughter chromosomes of each type. The first division separates these packets into doublets, one to each daughter. And then the daughters undergo another division (without DNA or chromosome duplication) to distribute one of each daughter to the second daughter generation. There is no second S period.

The result is that each of the 4 offspring of the mother cell possess <u>one</u> of each kind of chromosome, not 2. When gametes from the two parents fuse during the mating, the chromosome (and gene) number is reconstituted to the diploid state.

IV. Some terms to know:
dominant, recessive, genotype, phenotype, segregation, independent assortment, linkage, meiosis

V. Reading assignment for next lecture: pp. 51-62.

VI. Sample test questions: Here are some samples of two different kinds of questions from a previous midterm exam on this material.

1 Define the following terms. Give a brief, one or two sentence definition that clearly describes the meaning of the term.
(5 pts ea.)

primer for PCR

G-2 phase

anti-codon

2. . A list of different propositions appears below. They are all either false or at least questionable. Choose ___ of them, analyzing why they are incorrect or questionable<u>. For each answer, briefly (in a sentence or two) give the evidence, example, or reasoning to support your opinion</u>. (6 pts ea.)

The PCR reaction requires one primer, heating to separate DNA strands, and an annealing step.

The transforming principle in *Streptococcus pneumoniae* can be inactivated by protease digestion.

During translation the intron is removed from the mRNA

Power Pt. figures for Lecture 6

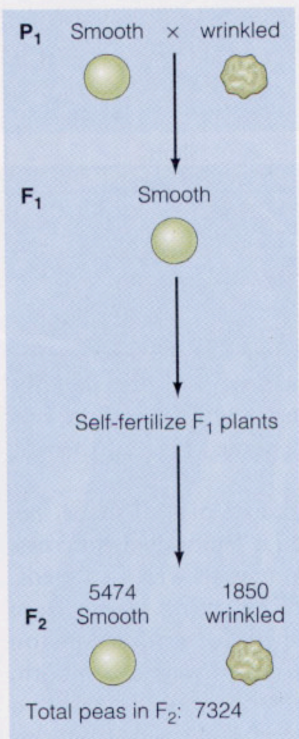


FIGURE 3.3 One of Mendel's crosses. Pure-breeding varieties of peas (smooth and wrinkled) were used as the P1 generation. All the offspring in the F1 generation had smooth seeds. Self-fertilization of F1 plants gave rise to both smooth and wrinkled progeny in the F2 generation. About three-fourths of the offspring were smooth and about one-fourth were wrinkled.

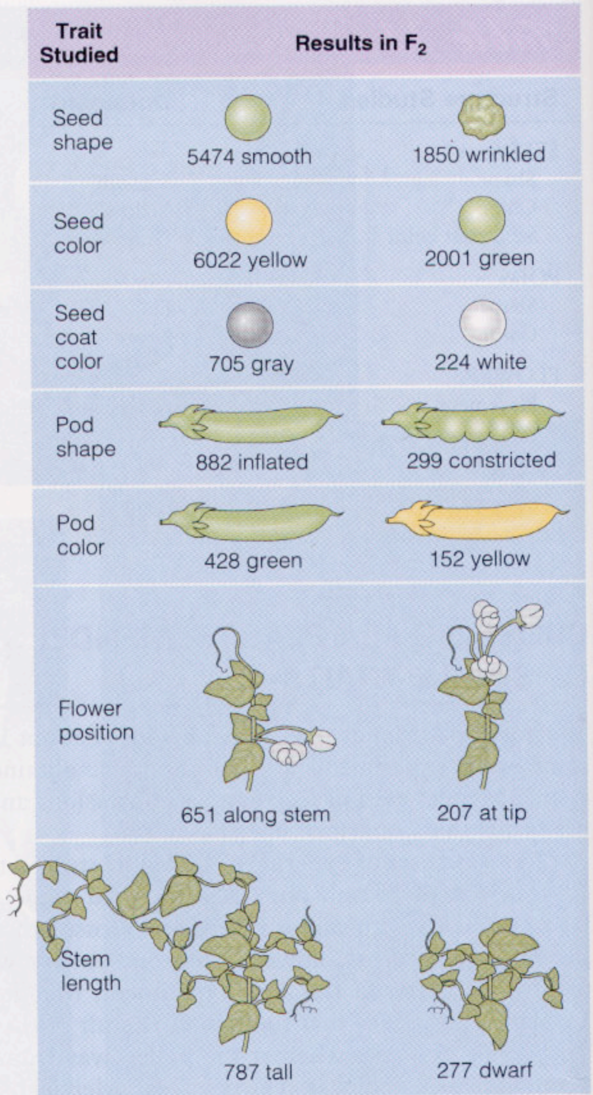
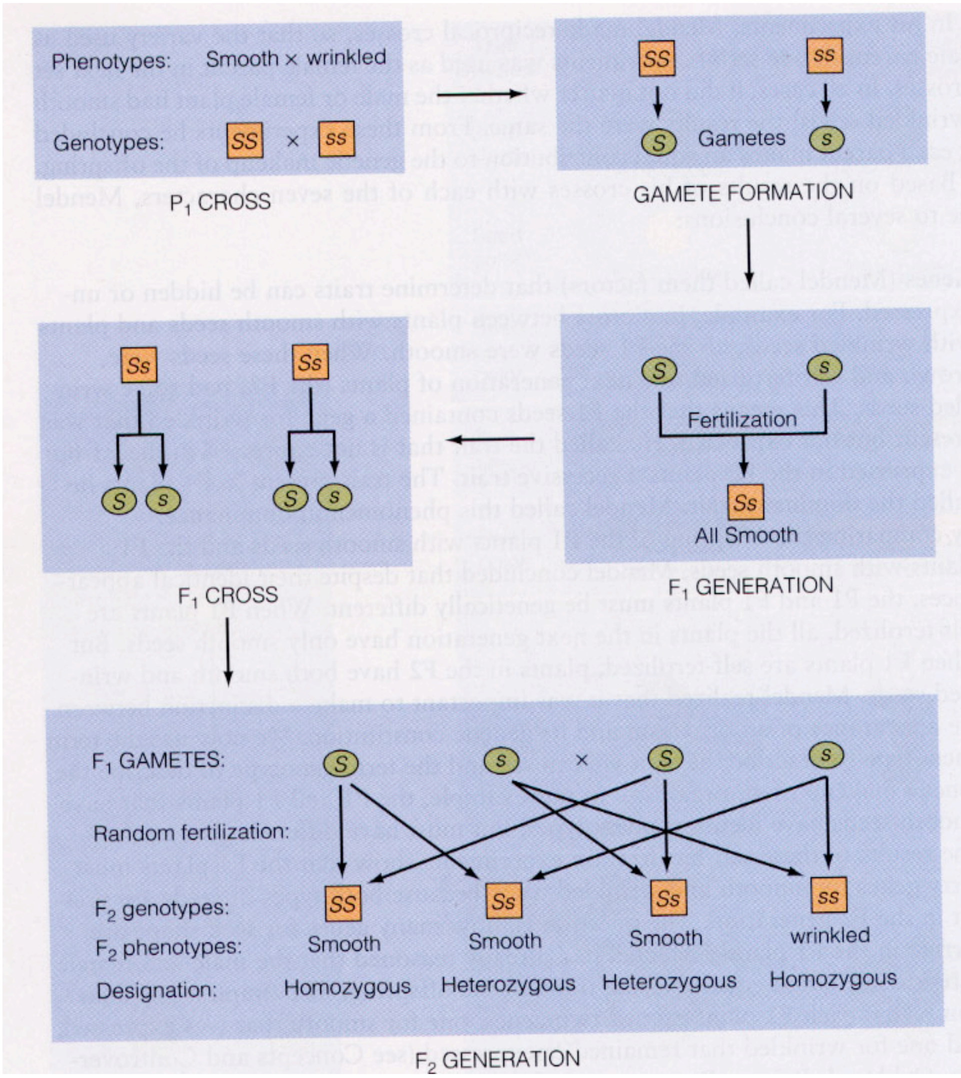


FIGURE 3.4 Results of Mendel's monohybrid crosses in peas. The numbers represent the F2 plants showing a given trait. On average, three-fourths of the offspring showed one trait, and one-fourth showed the other (a 3:1 ratio).



Phenotypes: Smooth × wrinkled
Genotypes: SS × ss
P1 CROSS
SS
ss
S
Gametes
s
GAMETE FORMATION
S
s
Fertilization
Ss
All Smooth
F1 GENERATION
Ss
Ss
S
s
S
s
F1 CROSS
F1 GAMETES:
S
s
×
S
s
Random fertilization:
F2 genotypes:
SS
Ss
Ss
ss
F2 phenotypes:
Smooth
Smooth
Smooth
wrinkled
Designation:
Homozygous
Heterozygous
Heterozygous
Homozygous
F2 GENERATION

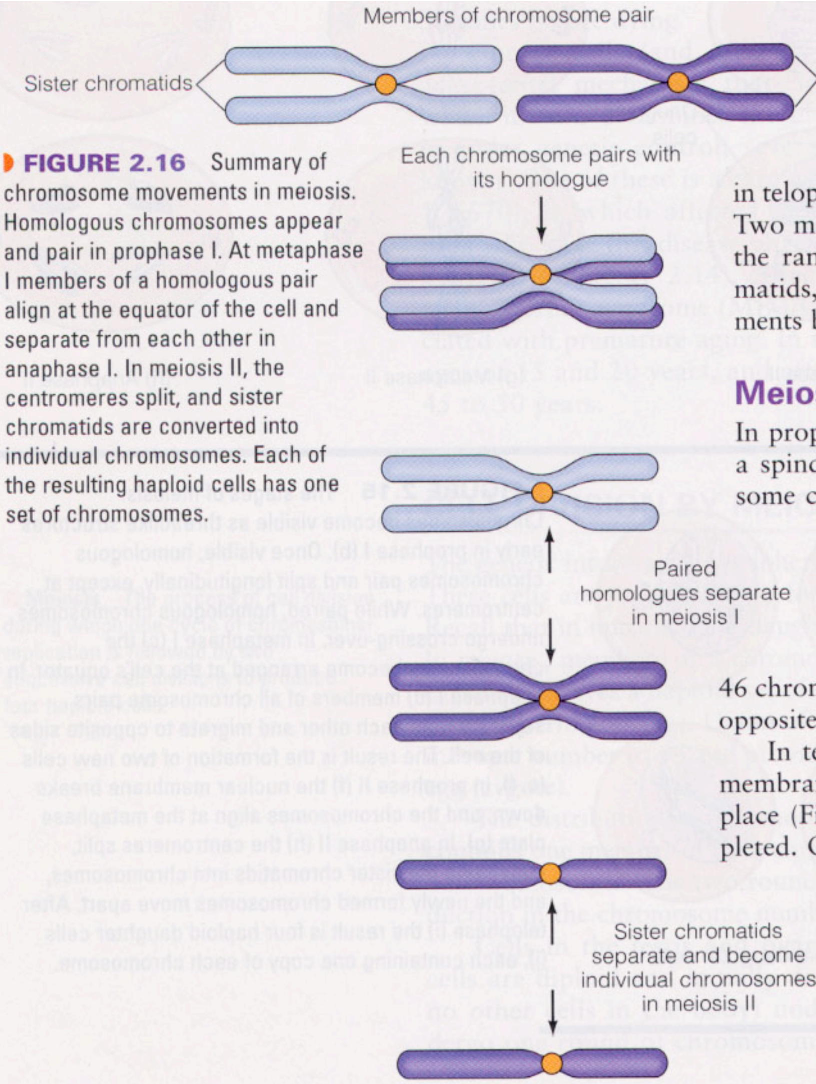


FIGURE 2.16 Summary of chromosome movements in meiosis. Homologous chromosomes appear and pair in prophase I. At metaphase I members of a homologous pair align at the equator of the cell and separate from each other in anaphase I. In meiosis II, the centromeres split, and sister chromatids are converted into individual chromosomes. Each of the resulting haploid cells has one set of chromosomes.

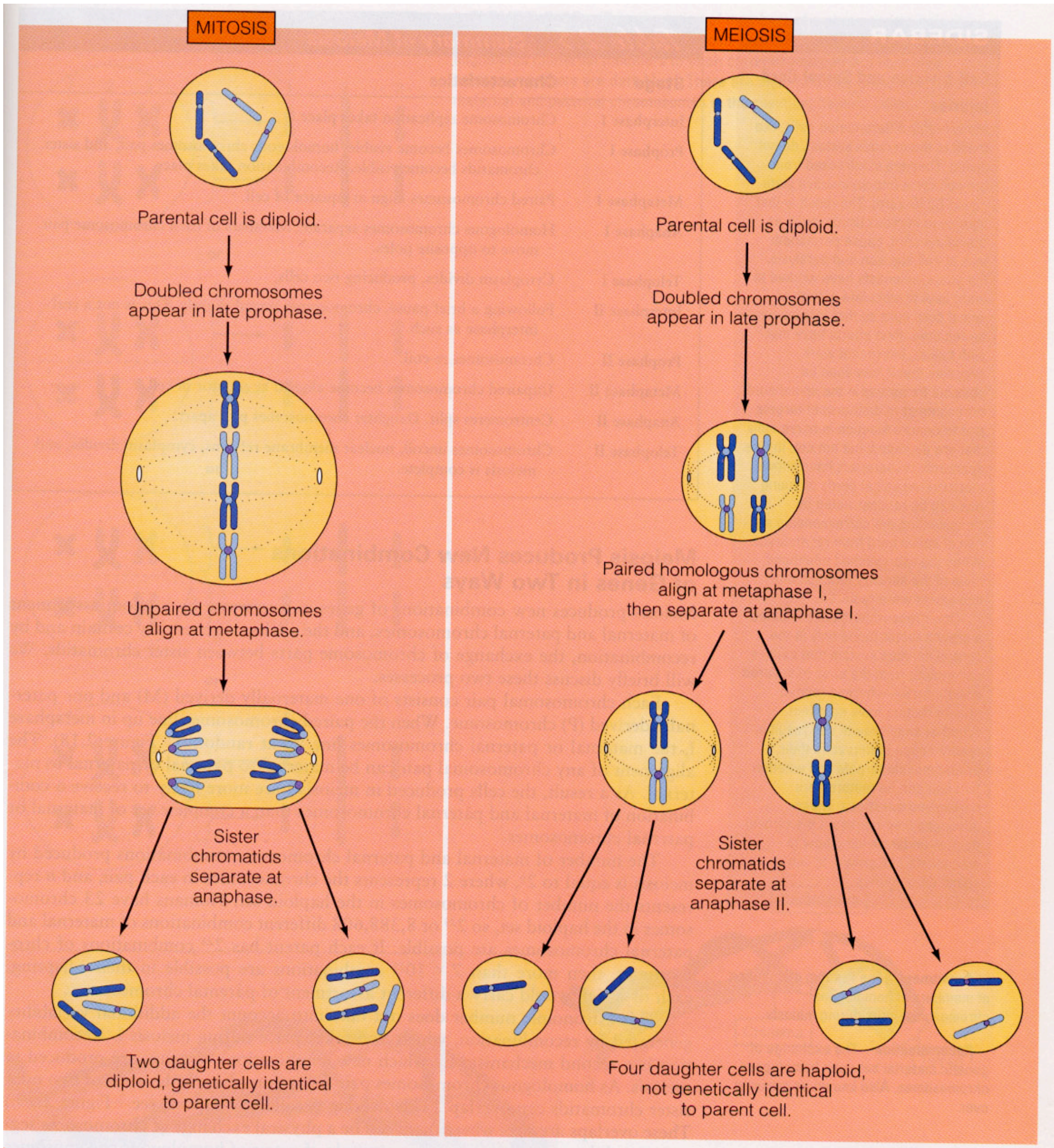
in telophase I, producin
Two major events take
the random separation
matids, and *crossing-ov*
ments between homolo

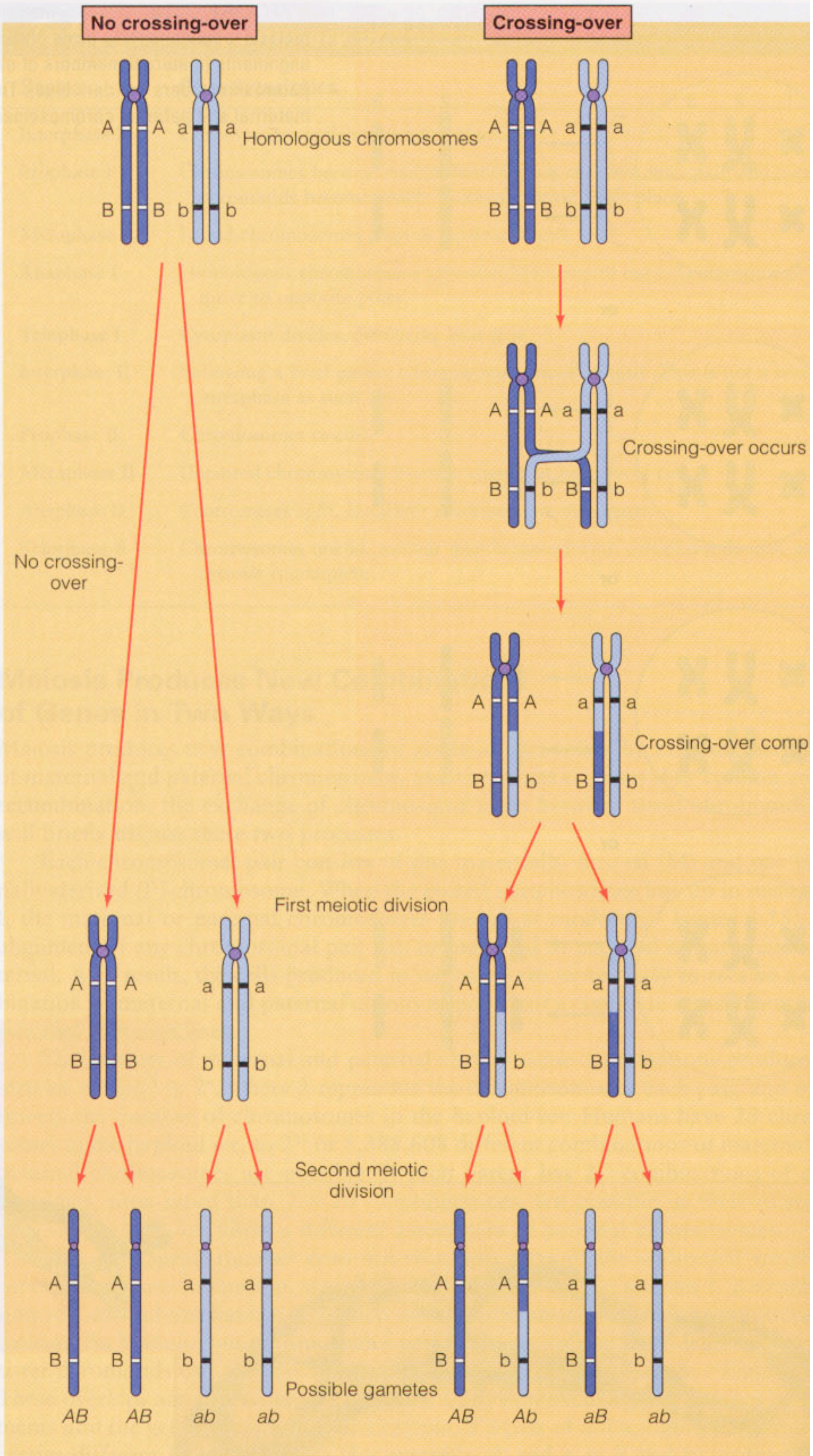
Meiosis II Begins

In prophase II, the chr
a spindle forms (Figur
some consists of two s
tromere. At r
23 unpaired c
fibers attached
(Figure 2.15h
each chromos
46 chromatids are conve
opposite poles of the cell

In telophase II, the
membrane re-forms, and
place (Figure 2.15i). Th
pleted. One diploid cell
gone one r
and two ro
haploid cel
copy of eac

The mo
marized in
of meiosis
ure 2.17 compares the


MITOSIS
Parental cell is diploid.
Doubled chromosomes appear in late prophase.
Unpaired chromosomes align at metaphase.
Sister chromatids separate at anaphase.
Two daughter cells are diploid, genetically identical to parent cell.
MEIOSIS
Parental cell is diploid.
Doubled chromosomes appear in late prophase.
Paired homologous chromosomes align at metaphase I, then separate at anaphase I.
Sister chromatids separate at anaphase II.
Four daughter cells are haploid, not genetically identical to parent cell.



No crossing-over
Crossing-over
A A a a
B B b b
Homologous chromosomes
A A a a
B B b b
A A a a
Crossing-over occurs
B b B b
No crossing-over
A A a a
Crossing-over comp
B b B b
First meiotic division
A A a a
B B b b
A A a a
B b B b
Second meiotic division
A A a a
B B b b
A A a a
B b B b
Possible gametes
AB AB ab ab
AB Ab aB ab